

2021 EAU Paediatric Urology Guidelines Search Strategy – Undescended Testes

Database: OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present, Embase <1974 to 2020 June 05>, EBM Reviews - Cochrane Database of Systematic Reviews <2005 to June 03, 2020>

Search Strategy:

-
- 1 (undescended adj3 (testes or testis or testicle)).tw. (6092)
 - 2 ((non-scrotal or nonscrotal) adj3 (testes or testis or testicle)).tw. (72)
 - 3 exp *Cryptorchidism/ (12278)
 - 4 Cryptorchidism.tw,kw. (11538)
 - 5 or/1-4 (20451)
 - 6 exp Child/ or exp Infant/ or exp Minors/ or exp Adolescent/ or exp adolescence/ or exp Pediatrics/ or exp newborn/ or exp Puberty/ or exp Schools/ or high school/ or kindergarten/ or middle school/ or nursery school/ or primary school/ (7310904)
 - 7 (baby or babies or child or children or pediatric* or paediatric* or peadiatric* or infan* or infancy or neonat* or newborn* or new born* or kid or kids or adolescen* or preschool or pre-school or toddler*).tw,kw. (4750428)
 - 8 (postmatur* or prematur* or preterm* or perinat* or boy* or girl* or teen* or minors or prepubescen* or prepuberty* or pubescen* or puber*).tw,kw. (1290517)
 - 9 (elementary school* or high school* or highschool* or kindergar* or nursery school* or primary school* or secondary school* or youth* or young or student* or juvenil* or underage* or (under* adj age*) or "under 16" or "under 18").tw,kw. (2068343)
 - 10 or/6-9 (9986491)
 - 11 5 and 10 (12930)
 - 12 (exp animals/ or exp animal/ or exp nonhuman/ or exp animal experiment/ or animal model/ or animal tissue/ or non human/ or (rat or rats or mice or mouse or swine or porcine or murine or sheep or lambs or pigs or piglets or rabbit or rabbits or cat or cats or dog or dogs or cattle or bovine or monkey or monkeys or trout or marmoset\$1 or basic research or cell lines or in vitro or animal model or canine).tw.) not (humans/ or human/ or (human* or patients or subjects).tw.) (10875069)
 - 13 11 not 12 (12485)
 - 14 conference abstract.pt. or Congresses as Topic/ or Conference Review.pt. or "Journal: Conference Abstract".pt. (3939033)
 - 15 case report/ or case reports/ or case report.ti. (4648524)

16 note/ or editorial/ or letter/ or Comment/ or news/ or (note or editorial or letter or Comment
or news).pt. (4646379)
17 or/14-16 (12520958)
18 13 not 17 (8091)
19 limit 18 to yr="2016 -current" (1066)
20 limit 19 to english language [Limit not valid in CDSR; records were retained] (999)
21 remove duplicates from 20 (584)

1.

Update on Pregnancy after Heart Transplantation.

Punnoose L., Coscia L.A., Kliniewski D., Constantinescu S., Moritz M.J.

Embase

The Journal of heart and lung transplantation : the official publication of the International Society
for Heart Transplantation. S. 39 (4) (pp S142-S143), 2020. Date of Publication: 01 Apr 2020.

[Article]

AN: 631927856

PURPOSE: The purpose of this study was to describe 171 pregnancies in 100 heart transplant
recipients.

METHOD(S): Data regarding pregnancies occurring between 1987 and 2018 were collected by
the Transplant Pregnancy Registry International (TPR) via questionnaires, telephone interviews,
and medical records.

RESULT(S): The mean age at first transplant was 20+/-8.5 yrs. The transplant to conception
interval was 6.8+/-10 yrs (range 0.15-26 yrs) and 37%of the pregnancies were unplanned.
Immunosuppression was calcineurin inhibitor-based with 20% exposed to a mycophenolic acid
product (MPA). Comorbid conditions during pregnancy included: hypertension 46%, preeclampsia
29%, and diabetes requiring insulin 7%. Rejection occurred during 14 pregnancies (8%) and
within 3 months post-partum in 11 pregnancies. Graft loss within 2 years of delivery occurred in 4
recipients; 1 recipient was successfully re-transplanted. Pregnancy outcomes (n=177 includes
multiple births) included: live births 67%, miscarriages 25% (46% with MPA exposure),

terminations 5%, ectopic 1% and stillbirths 1%. Of the 119 newborn, mean gestational age was 36.1+/-3.4 wks and mean birth weight was 2568+/-701 g. Birth defects were reported in 10 children and included: duodenal atresia, AV canal defect, Tetralogy of Fallot (MPA exposure); facial deformities (MPA exposure), laryngomalacia (MPA exposure), cystic hygroma, vermian hypoplasia of the cerebellum, hypospadias, undescended testicle, pectus excavatum, hydronephrosis, and tongue-tie. Seven additional children inherited their mother's cardiac disease; 4 children have received a heart transplant. At last follow-up, mean 8.2 +/- 6.3 yrs, 33 recipients had died (average age of their 36 children at time of maternal death was 10.2 +/- 6.1 yrs), 7 had reduced cardiac function, 8 unknown, and 52 recipients reported adequate transplant function.

CONCLUSION(S): This is the largest reported series of pregnancies in heart transplant recipients to date. The majority of pregnancies reported are successful. MPA exposure presents significant concerns. Pre-pregnancy counseling should include discussion of inheritable cardiac conditions, MPA avoidance, risk of rejection/graft dysfunction, and long-term maternal survival.

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PMC Identifier

32464898 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32464898>]

Institution

(Punnoose) Division of Cardiovascular Medicine, Vanderbilt University School of Medicine, TN, Nashville, United States (Coscia, Kliniewski) Transplant Pregnancy Registry International, Philadelphia, United States

(Constantinescu) Medicine, Lewis Katz School of Medicine, at Temple University, Philadelphia, United States

(Moritz) Surgery, Lehigh Valley Health Network, Allentown, United States

Publisher

NLM (Medline)

Year of Publication

2020

2.

Evaluation of human chorionic gonadotropin (HCG) therapeutic results in patients with unilateral cryptorchidism (undescended testis).

Sharifiaghdas F., Sharifiaghdas S., Maleki E.R., Narouie B., Shikhzadeh A., Mehrabi S.

Embase

Open Urology and Nephrology Journal. 13 (1) (pp 13-17), 2020. Date of Publication: 2020.

[Article]

AN: 2004466507

Background: Cryptorchidism or undescended testis is an evolutionary defect where one or both testes fail to descend into the scrotum. HCG causes the testes to fail, possibly due to weight gain, an increase in testicular vasculature, and stimulating the testosterone and di-hydro-testosterone. The present study has been conducted to evaluate the therapeutic effects of HCG on patients with unilateral cryptorchidism.

Method(s): In a prospective descriptive study, 211 patients of 8 months to 7 years old with unilateral cryptorchidism whose parents refused surgery received HCG therapy. The patients were followed up 1 month, 3months, and 12 months after the first injection. They were examined in terms of the location of testes, possible relapses, sides of undescended testes, treatment complications, and HCG therapeutic results.

Result(s): Four weeks after the first injection, 160 patients (75.12%) out of 211 cases had the descent of testes into the inguinal canal and the scrotum. 69.5% of non-palpable abdominal testes descended into the inguinal canal, 69.7% of patients with inguinal testes, 78% of patients with supra inguinal testes and 100% of patients with retractile testis experienced the descent of testes into the scrotum.

Conclusion(s): The therapeutic response to HCG was successful in more than 50% of the cases in all the groups. Therefore, the need for performing surgical procedures on children with unilateral cryptorchidism would be decreased and they can be treated by a cost-effective and less invasive method. Moreover, at least one-year follow-up of the patients is required to ensure outcomes of the treatment.

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Status

In-Process

Institution

(Sharifiaghdas, Maleki, Shikhzadeh) Department of Urology, Urology and Nephrology Research Center, Shahid Labbafinejad Medical Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran, Islamic Republic of (Sharifiaghdas) Department of Pediatrics, Torfeh General Hospital, Tehran, Iran, Islamic Republic of

(Narouie) Department of Urology, Zahedan University of Medical Sciences, Zahedan, Iran, Islamic Republic of

(Mehrabi) Department of Emergency, School of Medicine, Arak University of Medical Sciences, Arak, Iran, Islamic Republic of

Publisher

Bentham Science Publishers (P.O. Box 294, Bussum 1400 AG, Netherlands)

Year of Publication

2020

3.

Androgen Insensitivity Syndrome: A rare genetic disorder.

Fulare S., Deshmukh S., Gupta J.

Embase

International Journal of Surgery Case Reports. (no pagination), 2020. Date of Publication: 2020.

[Article]

AN: 2006069977

Background: Androgen Insensitivity Syndrome (AIS) is a rare X-linked recessive androgen receptor (AR) disorder with 46XY karyotype. Partial AIS affects 5-7 per 1,000,000 genetically male individuals whereas Complete AIS affects 2-5 per 100,000 genetically male individuals. CAIS CAIS is characterized by complete resistance to the action of androgens. Presentation of case: 17-year patient presented with swelling in bilateral inguinal region. Patient also complained of primary amenorrhea with serum FSH and LH levels being raised, serum testosterone level much above normal range. MRI Pelvis revealed agenesis of vagina, uterine body, both ovaries and cervix. Bilateral testes were noted in bilateral superficial inguinal ring. Bilateral orchidectomy was done and the patient was advised estrogen substitution therapy.

Discussion(s): CAIS is usually diagnosed at puberty, when the patient presents with primary amenorrhea. Karyotype has to be mapped in order to differentiate from other genetic disorders. Orchidectomy should be done to avoid risk of malignancy of undescended intra-abdominal testes (3.6 % at 25 years old, and 33 % at 50 years old, reported by various studies). Hormonal substitution therapy should be administered. Comprehensive psychiatric assessment and intervention go a long way in alleviating distress and enhancing quality of life.

Conclusion(s): Androgen Insensitivity Syndrome requires expert and sympathetic handling. Close collaboration between surgeon, gynaecologist and psychologist is essential for proper management of complete androgen insensitivity syndrome.

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Status

Article-in-Press

Author NameID

Deshmukh, Satish; ORCID: <http://orcid.org/0000-0001-5266-6603>

Institution

(Fulare, Deshmukh, Gupta) NKP Salve Institute of Medical Sciences, Nagpur, India

Publisher

Elsevier Ltd

Year of Publication

2020

4.

Postnatal germ cell development in cryptorchid boys.

Dong L.-H., Hildorf S., Clasen-Linde E., Kvist K., Cortes D., Thorup J., Andersen C.Y.

Embase

Asian Journal of Andrology. 22 (3) (pp 258-264), 2020. Date of Publication: 01 May 2020.

[Article]

AN: 631670255

Cryptorchidism is associated with infertility in adulthood. Early orchiopexy is suggested to reduce the risk. Information is lacking on the potential link between infant germ cell maturation and the risk of future infertility. The objective of the study was to evaluate age-related germ cell development in cryptorchidism. Immunostaining for markers of germ cell development (octamer-binding transcription factor 3/4 [OCT3/4], placental alkaline phosphatase [PLAP], KIT proto-oncogene [C-KIT], podoplanin [D2-40], Lin-28 homolog A [LIN28], and G antigen 7 [GAGE-7]) was performed in testicular biopsies from 40 cryptorchid boys aged 4-35 months. Germ cell numbers and distributions were evaluated in cross sections of seminiferous tubules, with and without immunostaining. OCT3/4, D2-40, and LIN28 were generally expressed in the early stages of germ cell development, as shown by positive expression in germ cells in the central region of seminiferous tubules. In contrast, PLAP and GAGE-7 were expressed in both central and peripheral parts of the tubules in the early stages of development and expressed mainly in a peripheral position with advancing age. Germ cell maturation was delayed in this study population as compared with that observed in our previous study on germ cell markers in a healthy population. The number of GAGE-7-positive germ cells per tubular cross section obtained by

immunostaining was significantly higher than that obtained by standard hematoxylin and eosin staining. Double immunostaining revealed heterogeneity in germ cell development in cryptorchid testes. These results shed light on the pathophysiology of germ cell development in boys with cryptorchidism.

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Status

Embase

Institution

(Dong, Andersen) Laboratory of Reproductive Biology, Copenhagen University Hospital, Rigshospitalet, Copenhagen 2100, Denmark (Hildorf, Cortes, Thorup, Andersen) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen 2200, Denmark

(Clasen-Linde) Department of Pathology, Copenhagen University Hospital, Rigshospitalet, Copenhagen 2100, Denmark

(Kvist, Thorup) Department of Pediatric Surgery, Copenhagen University Hospital, Rigshospitalet, Copenhagen 2100, Denmark

(Cortes) Department of Pediatrics, Copenhagen University Hospital Hvidovre, Copenhagen 2650, Denmark

Publisher

Wolters Kluwer Medknow Publications (B9, Kanara Business Centre, off Link Road, Ghatkopar (E), Mumbai 400 075, India)

Year of Publication

2020

5.

Urogenital Abnormalities in Adenosine Deaminase Deficiency.

Pajno R., Pacillo L., Recupero S., Cicalese M.P., Ferrua F., Barzaghi F., Ricci S., Marzollo A., Pecorelli S., Azzari C., Finocchi A., Cancrini C., Di Matteo G., Russo G., Alfano M., Lesma A., Salonia A., Adams S., Booth C., Aiuti A.

Embase

Journal of Clinical Immunology. 40 (4) (pp 610-618), 2020. Date of Publication: 01 May 2020.

[Article]

AN: 2004740288

Background: Improved survival in ADA-SCID patients is revealing new aspects of the systemic disorder. Although increasing numbers of reports describe the systemic manifestations of adenosine deaminase deficiency, currently there are no studies in the literature evaluating genital development and pubertal progress in these patients.

Method(s): We collected retrospective data on urogenital system and pubertal development of 86 ADA-SCID patients followed in the period 2000-2017 at the Great Ormond Street Hospital (UK) and 5 centers in Italy. In particular, we recorded clinical history and visits, and routine blood tests and ultrasound scans were performed as part of patients' follow-up. Results and Discussion: We found a higher frequency of congenital and acquired undescended testes compared with healthy children (congenital, 22% in our sample, 0.5-4% described in healthy children; acquired, 16% in our sample, 1-3% in healthy children), mostly requiring orchidopexy. No urogenital abnormalities were noted in females. Spontaneous pubertal development occurred in the majority of female and male patients with a few cases of precocious or delayed puberty; no patient presented high FSH values. Neither ADA-SCID nor treatment performed (PEG-ADA, BMT, or GT) affected pubertal development or gonadic function.

Conclusion(s): In summary, this report describes a high prevalence of cryptorchidism in a cohort of male ADA-SCID patients which could represent an additional systemic manifestation of ADA-SCID. Considering the impact urogenital and pubertal abnormalities can have on patients' quality of life, we feel it is essential to include urogenital evaluation in ADA-SCID patients to detect any abnormalities, initiate early treatment, and prevent long-term complications.

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Status

In-Process

Institution

(Pajno, Russo) Department of Pediatrics, Endocrine Unit, IRCCS San Raffaele Scientific Institute, Via Olgettina, 60, Milan 20132, Italy (Pacillo) Department of Pediatrics, "Pietro Barilla" Children Hospital, University of Parma, via Gramsci, Parma 14, Italy

(Pacillo, Finocchi, Cancrini, Di Matteo) Unit of Immune and Infectious Diseases, Scientific Institute for Research and Healthcare (IRCCS) Childrens' Hospital Bambino Gesù, University Department of Pediatrics (DPUO), Rome, Italy

(Pacillo, Finocchi, Cancrini, Di Matteo) Department of Systems Medicine, University of Rome Tor Vergata, Rome, Italy

(Recupero, Cicalese, Ferrua, Barzaghi, Aiuti) Pediatric Immunohematology and Stem Cell Program, IRCCS San Raffaele Scientific Institute, Via Olgettina, 60, Milan 20132, Italy
(Recupero, Cicalese, Ferrua, Barzaghi, Aiuti) San Raffaele Telethon Institute for Gene Therapy (SR-Tiget), IRCCS San Raffaele Scientific Institute, Milan, Italy
(Ricci, Azzari) Division of Pediatric Immunology, Department of Health Sciences, University of Florence and Meyer Children's Hospital, Florence, Italy
(Marzollo) Pediatric Hematology-Oncology Unit, Department of Women's and Children's Health, University of Padova, Via Giustiniani 3, Padova 35128, Italy
(Pecorelli) Department of Pediatric Surgery, Ospedale dei Bambini - Spedali Civili, Brescia, Piazzale Spedali Civili 1, Brescia 25123, Italy
(Alfano, Salonia) Division of Experimental Oncology/Unit of Urology, URI, IRCCS Ospedale San Raffaele, Milan, Italy
(Lesma) Unit of Pediatric Surgery, Department of Urology, IRCCS Ospedale San Raffaele, Milan, Italy
(Salonia, Aiuti) Università Vita-Salute San Raffaele, Milan, Italy
(Adams) SIHMDS-Haematology, Great Ormond Street Hospital for Children, London, United Kingdom
(Booth) Department of Paediatric Immunology, Great Ormond Street Hospital, London, United Kingdom
Publisher
Springer
Year of Publication
2020

6.

Androgen Treatment in Adolescent Males With Hypogonadism.

Rey R.A., Grinspon R.P.

Embase

American journal of men's health. 14 (3) (pp 1557988320922443), 2020. Date of Publication: 01 May 2020.

[Article]

AN: 631883302

During adolescence, androgens are responsible for the development of secondary sexual characteristics, pubertal growth, and the anabolic effects on bone and muscle mass. Testosterone is the most abundant testicular androgen, but some effects are mediated by its conversion to the more potent androgen dihydrotestosterone (DHT) or to estradiol. Androgen deficiency, requiring replacement therapy, may occur due to a primary testicular failure or secondary to a hypothalamic-pituitary disorder. A very frequent condition characterized by a late activation of the gonadal axis that may also need androgen treatment is constitutional delay of puberty. Of the several testosterone or DHT formulations commercially available, very few are employed, and none is marketed for its use in adolescents. The most frequently used androgen therapy is based on the intramuscular administration of testosterone enanthate or cypionate every 3 to 4 weeks, with initially low doses. These are progressively increased during several months or years, in order to mimic the physiology of puberty, until adult doses are attained. Scarce experience exists with oral or transdermal formulations. Preparations containing DHT, which are not widely available, are preferred in specific conditions. Oxandrolone, a non-aromatizable drug with higher anabolic than androgenic effects, has been used in adolescents with preserved testosterone production, like Klinefelter syndrome, with positive effects on cardiometabolic health and visual, motor, and psychosocial functions. The usual protocols applied for androgen therapy in boys and adolescents are discussed.

PMC Identifier

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Author NameID

Rey, Rodolfo A.; ORCID: <http://orcid.org/0000-0002-1100-3843>

Institution

(Rey, Grinspon) Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), CONICET - FEI - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina (Rey) Departamento de Biologia Celular, Histologia, Embriologia y Genetica, Facultad de Medicina, Universidad de Buenos Aires, Argentina

Publisher

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Year of Publication

2020

Effect of the occupational environment of parents on cryptorchidism.

Mitsui T., Araki A., Miyashita C., Ito S., Kitta T., Moriya K., Shinohara N., Takeda M., Kishi R.

Embase

Pediatrics international : official journal of the Japan Pediatric Society. (no pagination), 2020.

Date of Publication: 24 May 2020.

[Article]

AN: 631868960

OBJECTIVES: The early detection and treatment of cryptorchidism are necessary to preserve male fertility. This study aimed to assess the effect of parents' occupational environment on the incidence of cryptorchidism in their sons.

METHOD(S): The study enrolled 51,316 newborn males, whose mothers were recruited in the Japan Environment and Children's Study. We analyzed cryptorchidism incidence in male newborns according to 14 categories of occupations. We also analyzed the effect of the mother's occupational environment during gestation, including working time and night-shift work, on cryptorchidism incidence. Information on occupations was obtained from self-administered questionnaires. Cryptorchidism was identified through a survey at birth or 1 month after birth using medical records.

RESULT(S): Cryptorchidism was identified in 305 male babies (0.59%) at birth or 1 month after birth. Weight, height, head circumference, and chest circumference at birth were significantly lower in male babies with cryptorchidism than in those without the condition. Gestational age was also shorter in mothers whose babies developed cryptorchidism. Moreover, maternal age at delivery and smoking during gestation also had an effect on cryptorchidism incidence. However, multivariate analysis of the 14 categories of occupations of parents during gestation showed no significant effect on cryptorchidism incidence in their male babies.

CONCLUSION(S): This study revealed that the occupational environment of parents did not significantly affect the incidence of cryptorchidism in their sons. However, this study might have underestimated mild and transient cases of cryptorchidism. Further studies are necessary to investigate the risk factors of cryptorchidism in relation to parents' occupation.

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Status

Article-in-Press

Author NameID

Mitsui, Takahiko; ORCID: <http://orcid.org/0000-0003-0408-3678>

Institution

(Mitsui, Takeda) Department of Urology, University of Yamanashi Graduate School of Medical Science, Japan (Mitsui, Araki, Miyashita, Ito, Kishi) Hokkaido University Center for Environmental and Health Sciences, Sapporo, Japan

(Kitta, Moriya, Shinohara) Department of Urology, Hokkaido University Graduate School of Medicine, Sapporo, Japan

Publisher

NLM (Medline)

Year of Publication

2020

8.

Gonadotropin Treatment For The Male Hypogonadotropic Hypogonadism.

Boeri L., Capogrosso P., Salonia A.

Embase

Current pharmaceutical design. (no pagination), 2020. Date of Publication: 23 May 2020.

[Article]

AN: 631861263

Hypogonadotropic hypogonadism (HH) is caused by a dysfunction in the hypothalamus and/or the pituitary gland and it can be congenital or acquired. This condition is biochemically characterized by low or inappropriately normal gonadotropins levels along with low total testosterone levels. If fertility is not an issue, testosterone therapy is the treatment of choice to induce and maintain secondary sexual characteristics and sexual function. Spermatogenesis is frequently impaired in patients with HH, but usually responsive to hormonal therapy such as gonadotropin therapy or GnRH supplementary/replacement therapy. When gonadotropins are the choice of treatment conventional therapy includes human chorionic gonadotropin (hCG) along with different FSH formulation: human menopausal gonadotropins (hMG), highly purified urinary FSH preparations (hpFSH) (e.g., urofollitropin) or recombinant FSH (rFSH). The combination of FSH and hCG demonstrated to be associated with better outcomes than single compounds, whereas similar results were obtained with different FSH preparations in male individuals both regarding the ability to stimulate spermatogenesis and eventually inducing physiology pregnancy. Gonadotropins can be administered either subcutaneously or intramuscularly. The combination therapy with hCG and FSH for a period of 12-24 months was found to promote testicular growth

in almost all patients, spermatogenesis in approximately 80% and pregnancy rates in the range of 50%. Gynecomastia is the most common side effect of gonadotropin therapy and is due to hCG stimulation of aromatase causing increased secretion of estradiol. The therapeutic success is higher in patients with post-puberal HH, in those without previously undescended testes, in patients with higher baseline testicular volume, who underwent repeated cycles of therapy and in patients with higher baseline inhibin B serum concentrations. Reversal of hypogonadism can occur in up to 10% of patients but its physiopathologic mechanism has yet to be elucidated. In conclusion, gonadotropins therapy is effective in promoting puberty and in supporting spermatogenesis onset and preservation in HH patients with either hypothalamic or pituitary conditions.

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Institution

(Boeri, Capogrosso, Salonia) Division of Experimental Oncology/Unit of Urology, IRCCS Ospedale San Raffaele, Milan. Italy, URI, Switzerland

Publisher

NLM (Medline)

Year of Publication

2020

9.

The lack of HSD17B3 in male mice results in disturbed Leydig cell maturation and endocrine imbalance akin to humans with HSD17B3 deficiency.

Sipila P., Junnila A., Hakkarainen J., Huhtaniemi R., Mairinoja L., Zhang F.P., Strauss L., Ohlsson C., Kotaja N., Huhtaniemi I., Poutanen M.

Embase

FASEB Journal. 34 (5) (pp 6111-6128), 2020. Date of Publication: 01 May 2020.

[Article]

AN: 2004457877

Hydroxysteroid (17beta) dehydrogenase type 3 (HSD17B3) deficiency causes a disorder of sex development in humans, where affected males are born with female-appearing external genitalia, but are virilized during puberty. The hormonal disturbances observed in the Hsd17b3 knockout mice (HSD17B3KO), generated in the present study, mimic those found in patients with HSD17B3 mutations. Identical to affected humans, serum T in the adult HSD17B3KO mice was within the normal range, while a striking increase was detected in serum A-dione concentration. This resulted in a marked reduction of the serum T/A-dione ratio, a diagnostic hallmark for the patients with HSD17B3 deficiency. However, unlike humans, male HSD17B3KO mice were born with normally virilized phenotype, but presenting with delayed puberty. In contrast to the current belief, data from HSD17B3KO mice show that the circulating T largely originates from the testes, indicating a strong compensatory mechanism in the absence of HSD17B3. The lack of testicular malignancies in HSD17B3KO mice supports the view that testis tumors in human patients are due to associated cryptorchidism. The HSD17B3KO mice presented also with impaired Leydig cell maturation and signs of undermasculinization in adulthood. The identical hormonal disturbances between HSD17B3 deficient knockout mice and human patients make the current mouse model valuable for understanding the mechanism of the patient phenotypes, as well as endocrinopathies and compensatory steroidogenic mechanisms in HSD17B3 deficiency.

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Status

Embase

Institution

(Sipila, Junnila, Hakkarainen, Huhtaniemi, Mairinoja, Zhang, Strauss, Kotaja, Huhtaniemi, Poutanen) Institute of Biomedicine, Research Centre for Integrative Physiology and Pharmacology, University of Turku, Turku, Finland (Sipila, Strauss, Poutanen) Turku Center for Disease Modeling (TCDM), Institute of Biomedicine, University of Turku, Turku, Finland (Ohlsson, Poutanen) Institute of Medicine, the Sahlgrenska Academy, Gothenburg University, Gothenburg, Sweden

(Zhang) GM-Unit of Laboratory Animal Centre, STEMM Research Program, Faculty of Medicine, University of Helsinki, Helsinki, Finland

Publisher

John Wiley and Sons Inc. (P.O.Box 18667, Newark NJ 07191-8667, United States)

Year of Publication

2020

10.

Safety of Antimicrobials During Pregnancy: A Systematic Review of Antimicrobials Considered for Treatment and Postexposure Prophylaxis of Plague.

Yu P.A., Tran E.L., Parker C.M., Kim H.-J., Yee E.L., Smith P.W., Russell Z., Nelson C.A., Broussard C.S., Yu Y.C., Meaney-Delman D.

Embase

Clinical infectious diseases : an official publication of the Infectious Diseases Society of America. 70 (1 Supplementement) (pp S37-S50), 2020. Date of Publication: 21 May 2020.

[Article]

AN: 631836554

BACKGROUND: The safety profile of antimicrobials used during pregnancy is one important consideration in the decision on how to treat and provide postexposure prophylaxis (PEP) for plague during pregnancy.

METHOD(S): We searched 5 scientific literature databases for primary sources on the safety of 9 antimicrobials considered for plague during pregnancy (amikacin, gentamicin, plazomicin, streptomycin, tobramycin, chloramphenicol, doxycycline, sulfadiazine, and trimethoprim-sulfamethoxazole [TMP-SMX]) and abstracted data on maternal, pregnancy, and fetal/neonatal outcomes.

RESULT(S): Of 13 052 articles identified, 66 studies (case-control, case series, cohort, and randomized studies) and 96 case reports were included, totaling 27 751 prenatal exposures to amikacin (n = 9), gentamicin (n = 345), plazomicin (n = 0), streptomycin (n = 285), tobramycin (n = 43), chloramphenicol (n = 246), doxycycline (n = 2351), sulfadiazine (n = 870), and TMP-SMX (n = 23 602). Hearing or vestibular deficits were reported in 18/121 (15%) children and 17/109 (16%) pregnant women following prenatal streptomycin exposure. First trimester chloramphenicol exposure was associated with an elevated risk of an undescended testis (odds ratio [OR] 5.9, 95% confidence interval [CI] 1.2-28.7). Doxycycline was associated with cardiovascular malformations (OR 2.4, 95% CI 1.2-4.7) in 1 study and spontaneous abortion (OR 2.8, 95% CI 1.9-4.1) in a separate study. First trimester exposure to TMP-SMX was associated with increased risk of neural tube defects (pooled OR 2.5, 95% CI 1.4-4.3), spontaneous abortion (OR 3.5, 95% CI 2.3-5.6), preterm birth (OR 1.5, 95% CI 1.1-2.1), and small for gestational age (OR 1.6, 95% CI 1.2-2.2). No other statistically significant associations were reported.

CONCLUSION(S): For most antimicrobials reviewed, adverse maternal/fetal/neonatal outcomes were not observed consistently. Prenatal exposure to streptomycin and TMP-SMX was associated with select birth defects in some studies. Based on limited data, chloramphenicol and doxycycline may be associated with adverse pregnancy or neonatal outcomes; however, more data are needed to confirm these associations. Antimicrobials should be used for treatment and PEP of plague during pregnancy; the choice of antimicrobials may be influenced by these data as well as information about the risks of plague during pregnancy.

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Institution

(Yu, Parker, Kim, Yee, Russell, Yu) Division of Preparedness and Emerging Infections, National Center for Emerging and Zoonotic Infectious Diseases (NCEZID), Centers for Disease Control and Prevention (CDC), Atlanta, GA, United States (Tran, Broussard, Meaney-Delman) Division of Birth Defects and Infant Disorders, National Center on Birth Defects and Developmental Disabilities, CDC, Atlanta, GA, United States

(Tran) Eagle Global Scientific, LLC, San Antonio, TX, United States

(Parker, Kim, Yee) Chenega Professional and Technical Services, LLC, Atlanta, GA, United States

(Smith) NCEZID, CDC, Atlanta, GA, United States

(Russell) Oak Ridge Institute for Science and Education CDC Fellowship Program, Atlanta, GA, United States

(Nelson) Division of Vector-Borne Diseases, NCEZID, CDC, Fort Collins, CO, United States

Publisher

NLM (Medline)

Year of Publication

2020

11.

The endocrine manifestations of spinal muscular atrophy, a real-life observational study.

Brener A., Lebenthal Y., Shtamler A., Levy S., Stein R., Fattal-Valevski A., Sagi L.

Embase

Neuromuscular Disorders. 30 (4) (pp 270-276), 2020. Date of Publication: April 2020.

[Article]

AN: 2005503666

The introduction of nusinersen, the first therapeutic modality for Spinal Muscular Atrophy (SMA) patients has raised hopes and led to construction of a multi-professional medical SMA service, including pediatric endocrinology. Our study aimed to provide a comprehensive description of the endocrine manifestations of SMA patients with variable degree of sarcopenia. Real-life clinical and laboratory data of 62 SMA patients (age range 3 months to 31 years, 24 type 1, 21 type 2, 17 type 3) were collected including: weight-status, self-reported information on puberty, current pubertal stage, Homeostatic Model Assessment of Insulin Resistance (HOMA-IR), basal gonadotropin and androgen levels. Precocious pubarche (mean age at onset 3.9 +/- 2.8 years) was found in 24% (15/62) of the SMA cohort [45.9%(11/24) type 1 and 19%(4/21) type 2]. A higher HOMA-IR predicted precocious pubarche after adjustment for SMA type and age (OR=1.42; 95% CI, 1.05, 1.93, P = 0.025). Bilateral cryptorchidism was found in 60% of type 1 and 30% of type 2 boys; type 3 young adult males attained full puberty. Most of the young women had normal pubertal development and regular menses, regardless of degree of obesity. Our findings suggest that isolated precocious pubarche is associated with early-onset insulin resistance linked to severity of muscular atrophy.

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Embase

Author NameID

Lebenthal, Yael; ORCID: <http://orcid.org/0000-0001-5963-0574>

Institution

(Brener, Lebenthal, Stein) Pediatric Endocrinology and Diabetes Unit, Dana-Dwek Children's Hospital, Tel Aviv Sourasky Medical Center, Tel Aviv 64239-06, Israel (Brener, Lebenthal, Fattal-Valevski, Sagi) Sackler Faculty of Medicine, Tel Aviv University, Tel Aviv 69978-01, Israel (Shtamler, Fattal-Valevski, Sagi) Pediatric Neurology Institute, Dana-Dwek Children's Hospital, Tel Aviv Sourasky Medical Center, Tel Aviv 64239-06, Israel (Levy) Statistical Education Unit, The Academic College of Tel Aviv Yaffo, Tel-Aviv 68182-11, Israel

Publisher

Elsevier Ltd

Year of Publication

12.

Female Authorship Publishing Trends and Forecasting in Pediatric Urology: Are We Closer to Gender Equality?.

Rickard M., Hannick J.H., Blais A.-S., Wang J., Santos J.D., Lorenzo A.J.

Embase

Urology. 139 (pp 141-150), 2020. Date of Publication: May 2020.

[Article]

AN: 2005190021

OBJECTIVE: To review the literature of 5 pediatric urology topics and conduct gender based and forecasting analyses of first and corresponding authors.

METHOD(S): A PubMed search was performed for hypospadias, hydronephrosis, vesicoureteral reflux, bladder and bowel dysfunction, and cryptorchidism over 3 decades from 1990 to 2019.

The 50 most relevant "best match" papers from each decade were extracted by topic. Author gender, specialty, and advanced degrees, along with journal and publication variables were collected. Forecasting analyses were conducted through the Holt-Winters method.

RESULT(S): Among 750 papers analyzed, 78% of corresponding and 70% of first authors were male. A significant upward trend was observed for female-authored publications in both first and corresponding positions over time ($P < .01$). Forecasting analyses predicted a continuing upward trend for female corresponding (55%) and first authors (83%) by 2049. Most studies originated from pediatric urology (59%), followed by pediatric surgery (9%) and endocrinology/genetics (5%). Papers focused in The Journal of Urology (30%) with the majority originating from the United States (38%). Most were retrospective (44%) and discussed medical (54%) versus surgical management (20%).

CONCLUSION(S): The majority of pediatric urology literature has been generated by male authors. A persistent, rising trend in female authorship across all examined pediatric urology topics was noted. These encouraging findings are projected to continue to increase in the future, suggesting a movement toward equal and fair gender representation in authorship in pediatric urology.

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Status

Embase

Institution

(Rickard, Hannick, Blais, Wang, Santos, Lorenzo) Division of Urology, The Hospital for Sick Children, Toronto, Ontario, Canada (Hannick) Division of Pediatric Urology, UH Rainbow Babies and Children's Hospital, Cleveland, OH, United States

(Blais) Division of Urology, Centre Hospitalier Universitaire de Quebec, Quebec City, Quebec, Canada

(Wang) Department of Statistical Sciences, University of Toronto, Toronto, Ontario, Canada

Publisher

Elsevier Inc. (E-mail: usjcs@elsevier.com)

Year of Publication

2020

13.

Maternal serum concentrations of bisphenol A and propyl paraben in early pregnancy are associated with male infant genital development.

Fisher B.G., Thankamony A., Mendiola J., Petry C.J., Frederiksen H., Andersson A.M., Juul A., Ong K.K., Dunger D.B., Hughes I.A., Acerini C.L.

Embase

Human reproduction (Oxford, England). 35 (4) (pp 913-928), 2020. Date of Publication: 28 Apr 2020.

[Article]

AN: 631622288

STUDY QUESTION: Are maternal serum phthalate metabolite, phenol and paraben concentrations measured at 10-17 weeks of gestation associated with male infant genital developmental outcomes, specifically cryptorchidism, anogenital distance (AGD), penile length and testicular descent distance, at birth and postnatally? SUMMARY ANSWER: Maternal serum bisphenol A (BPA) concentration at 10-17 weeks of gestation was positively associated with congenital or postnatally acquired cryptorchidism, and n-propyl paraben (n-PrP) concentration was associated with shorter AGD from birth to 24 months of age. WHAT IS KNOWN ALREADY:

Male reproductive disorders are increasing in prevalence, which may reflect environmental influences on foetal testicular development. Animal studies have implicated phthalates, BPA and parabens, to which humans are ubiquitously exposed. However, epidemiological studies have generated conflicting results and have often been limited by small sample size and/or measurement of chemical exposures outside the most relevant developmental window. STUDY DESIGN, SIZE, DURATION: Case-control study of cryptorchidism nested within a prospective cohort study (Cambridge Baby Growth Study), with recruitment of pregnant women at 10-17 postmenstrual weeks of gestation from a single UK maternity unit between 2001 and 2009 and 24 months of infant follow-up. Of 2229 recruited women, 1640 continued with the infancy study after delivery, of whom 330 mothers of 334 male infants (30 with congenital cryptorchidism, 25 with postnatally acquired cryptorchidism and 279 unmatched controls) were included in the present analysis. PARTICIPANTS/MATERIALS, SETTING, METHODS: Maternal blood was collected at enrolment, and serum levels of 16 phthalate metabolites, 9 phenols (including BPA) and 6 parabens were measured using liquid chromatography/tandem mass spectrometry. Logistic regression was used to model the association of cryptorchidism with serum chemical concentrations, adjusting for putative confounders. Additionally, offspring AGD, penile length and testicular descent distance were assessed at 0, 3, 12, 18 and 24 months of age, and age-specific Z scores were calculated. Associations between serum chemical levels and these outcomes were tested using linear mixed models. MAIN RESULTS AND THE ROLE OF CHANCE: Maternal serum BPA concentration was associated with offspring all-type cryptorchidism both when considered as a continuous exposure (adjusted odds ratio per log₁₀ µg/l: 2.90, 95% CI 1.31-6.43, P= 0.009) and as quartiles (phet= 0.002). Detection of n-PrP in maternal serum was associated with shorter AGD (by 0.242 standard deviations, 95% CI 0.051-0.433, P= 0.01) from birth to 24 months of age; this reduction was independent of body size and other putative confounders. We did not find any consistent associations with offspring outcomes for the other phenols, parabens, and phthalate metabolites measured. LIMITATIONS, REASONS FOR CAUTION: We cannot discount confounding by other demographic factors or endocrine-disrupting chemicals. There may have been misclassification of chemical exposure due to use of single serum measurements. The cohort was not fully representative of pregnant women in the UK, particularly in terms of smoking prevalence and maternal ethnicity. WIDER IMPLICATIONS OF THE FINDINGS: Our observational findings support experimental evidence that intrauterine exposure to BPA and n-PrP during early gestation may adversely affect male reproductive development. More evidence is required before specific public health recommendations can be made. STUDY FUNDING/COMPETING INTEREST(S): This work was supported by a European Union Framework V programme, the World Cancer Research Fund International, the Medical Research Council (UK), Newlife the Charity for Disabled Children, the Mothercare Group Foundation, Mead Johnson Nutrition and the National Institute for Health Research Cambridge

Comprehensive Biomedical Research Centre. Visiting Fellowship (J.M.): Regional Programme 'Jimenez de la Espada' for Research Mobility, Cooperation and Internationalization, Seneca Foundation-Science and Technology Agency for the Region of Murcia (No. 20136/EE/17). K.O. is supported by the Medical Research Council (UK) (Unit Programme number: MC_UU_12015/2).

The authors declare no conflict of interest.

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32325494 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32325494>]

Institution

(Fisher, Thankamony, Petry, Ong, Dunger, Hughes, Acerini) Department of Paediatrics, University of Cambridge, Box 116, Level 8, Addenbrooke's Hospital, Hills Road, Cambridge, CB2 0QQ, UK (Mendiola) Division of Preventive Medicine and Public Health, Department of Public Health Sciences, University of Murcia School of Medicine, IMIB-Arrixaca, Avda. Teniente Flomesta, Murcia 30003, Spain

(Frederiksen, Andersson, Juul) Department of Growth and Reproduction & International Center for Research and Research Training in Endocrine Disruption of Male Reproduction and Child Health (EDMaRC), Rigshospitalet, University of Copenhagen, Blegdamsvej 9, Copenhagen DK-2100, Denmark

(Ong) MRC Epidemiology Unit, University of Cambridge, Box 285, Institute of Metabolic Science, Cambridge Biomedical Campus, Cambridge, CB2 0QQ, UK

(Dunger) Metabolic Research Laboratories, University of Cambridge, Box 289, Institute of Metabolic Science, Cambridge Biomedical Campus, Cambridge, CB2 0QQ, UK

Publisher

NLM (Medline)

Year of Publication

2020

14.

Distinguishing between hidden testes and anorchia: The role of endocrine evaluation in infancy and childhood.

Jespersen K., Ljubicic M.L., Johannsen T.H., Christiansen P., Skakkebaek N.E., Juul A.

Embase

European journal of endocrinology. (no pagination), 2020. Date of Publication: 01 May 2020.

[Article]

AN: 631785326

OBJECTIVE: Non-palpable testes remain a diagnostic challenge, often involving exploratory laparoscopy. We evaluated the diagnostic value of a wide range of reproductive hormones in order to distinguish between bilateral cryptorchidism and bilateral anorchia. DESIGN: In this retrospective study, we identified and included 36 boys with non-palpable testes (20 with cryptorchidism, 3 with congenital hypogonadotropic hypogonadism (CHH), and 13 with anorchia) at first examination during childhood.

METHOD(S): Information on karyotype, phenotype, surgical results from laparoscopy, and biochemistry was retrieved from patient files. We compared serum concentrations of AMH, inhibin B, FSH, LH, testosterone (T), estradiol, and hCG stimulation testing in cryptorchid and anorchid boys to serum concentrations in a large, age-matched control group. Receiver operating characteristic curves were used to determine cut-off values of each reproductive hormone as a predictor of the presence of functional testicular tissue.

RESULT(S): Concentrations of AMH in 0-1 year olds: ≥ 155 pmol/L and $>1-15$ year olds: ≥ 19 pmol/L, inhibin B (≥ 22 pg/mL and ≥ 4 pg/mL), FSH (≤ 28.9 IU/L and ≤ 20.3 IU/L) and hCG-induced T ($>1-15$ year olds: ≥ 2 nmol/L) were significantly sensitive and specific markers in predicting the presence of functional testicular tissue in boys with non-palpable testes. In infancy, anorchid infants had significantly elevated gonadotropin levels while CHH had low levels.

CONCLUSION(S): Our findings suggest that laparoscopy may not be necessary in all boys with non-palpable testes if reproductive hormones unequivocally confirm the presence of functional testicular tissue while proving the absence may still be a diagnostic challenge.

PMC Identifier

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Status

Article-in-Press

Institution

(Jespersen) Department of Growth and Reproduction, Rigshospitalet, Kobenhavn, Denmark

(Ljubicic) Ljubicic, Department of Growth and Reproduction, Rigshospitalet, Kobenhavn 2100, Denmark

(Johannsen) Department of Growth and Reproduction, Rigshospitalet, Kobenhavn, Denmark

(Christiansen) Christiansen, Department of Growth and Reproduction, Rigshospitalet, Kobenhavn, Denmark

(Skakkebaek) Skakkebaek, Department of Growth and Reproduction, Rigshospitalet, Kobenhavn, Denmark

(Juul) Department of Growth and Reproduction, Rigshospitalet, Kobenhavn, Denmark

Publisher

NLM (Medline)

Year of Publication

2020

15.

Clinical, hormonal and genetic characteristics of androgen insensitivity syndrome in 39 Chinese patients.

Liu Q., Yin X., Li P.

Embase

Reproductive Biology and Endocrinology. 18 (1) (no pagination), 2020. Article Number: 34. Date of Publication: 28 Apr 2020.

[Article]

AN: 631621945

Background: Abnormal androgen receptor (AR) genes can cause androgen insensitivity syndrome (AIS), and AIS can be classified into complete androgen insensitivity syndrome (CAIS), partial androgen insensitivity syndrome (PAIS) and mild AIS. We investigated the characteristics of clinical manifestations, serum sex hormone levels and AR gene mutations of 39 AIS patients, which provided deeper insight into this disease.

Method(s): We prospectively evaluated 39 patients with 46, XY disorders of sex development (46, XY DSD) who were diagnosed with AIS at the Department of Endocrinology of Shanghai Children's Hospital from 2014 to 2019. We analysed clinical data from the patients including hormone levels and AR gene sequences. Furthermore, we screened the AR gene sequences of the 39 AIS patients to identify probable mutations.

Result(s): The 39 AIS patients came from 37 different families; 19 of the patients presented CAIS, and 20 of them presented PAIS. The CAIS patients exhibited a higher cryptorchidism rate than the PAIS (100 and 55%, $P = 0.001$). There were no significant difference between the CAIS and PAIS groups regarding the levels of inhibin B (INHB), sex hormone-binding globulin (SHBG), basal luteinizing hormone (LH), testosterone (T), or basal dihydrotestosterone (DHT), the T:DHT

ratio, DHT levels after human chorionic gonadotropin (HCG) stimulation or T levels after HCG stimulation. However, the hormone levels of AMH ($P = 0.010$), peak LH ($P = 0.033$), basal FSH ($P = 0.009$) and peak FSH ($P = 0.033$) showed significant differences between the CAIS group and the PAIS group. Twenty-one reported pathogenic and 9 novel AR mutations were identified. Spontaneous AR mutations were found in 5 AIS patients, and 21 patients inherited mutations from their mothers, who carried heterozygous mutations.

Conclusion(s): Forty-six XY DSD patients with cryptorchidism and female phenotypes were highly suspected of having AIS. We demonstrated that CAIS patients could not be distinguished by their hormone levels alone. Compared with PAIS patients, CAIS patients exhibited higher basal FSH, peak FSH, and peak LH hormone levels but lower AMH expression. We identified 21 reported pathogenic AR mutations and 9 novel AR mutations that led to different types of AIS. Missense mutations were the major cause of AIS and mostly occurred in exon 7 of the AR gene. These findings provided deeper insight into the diagnosis and classification of AIS and will even contributed to its clinical assessment.

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PMC Identifier

32345305 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32345305>]

Status

Embase

Institution

(Liu, Yin, Li) Department of Endocrinology, Shanghai Children's Hospital, Children's Hospital Affiliated to Shanghai Jiao Tong University, Shanghai 200062, China

Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2020

16.

Individualized treatment of pediatric inguinal hernia reduces adolescent recurrence rate: an analysis of 3006 cases.

Chu C.B., Chen J., Shen Y.M., Liu S.J., Sun L., Nie Y.S., Liu J., Cao J.X., Du H.D., Zou Z.Y., Yuan X.

Embase

Surgery Today. 50 (5) (pp 499-508), 2020. Date of Publication: 01 May 2020.

[Article]

AN: 2003911821

Purpose: We implemented the individualized treatment (IT) regimen for children with inguinal hernia and the Lichtenstein hernioplasty using an acellular tissue matrix patch (LHAP) for those with high risks. This retrospective study compares the complications of conventional laparoscopic high hernia sac ligation (LHSL) with those of the IT regimen for the management of pediatric inguinal hernia and investigates whether the recurrence rate of inguinal hernias in children treated by IT is lower than that of those treated by LHSL.

Method(s): The subjects of this retrospective study were 3006 children who underwent LHSL or IT for inguinal hernia between February, 2008 and February, 2016 at the Beijing Chao-Yang Hospital (Beijing, China). They comprised 1516 (50.4%) children who underwent LHSL between February, 2008 and December, 2012, and 1490 (49.6%) who underwent IT between January, 2013 and June, 2016. We analyzed the patients' data, including clinical characteristics and postoperative complications. The mean follow-up was 85.31 months for the LHSL group and 43.34 months for the IT group ($P < 0.01$). Given the difference in the follow-up periods, the log-rank test was used to analyze the recurrence rate.

Result(s): The mean age, weight, and height of these children at the time of surgery were 6 years old, 24.17 kg, and 114.48 cm in the LHSL group and 6 years old, 24.57 kg, and 115.18 cm in the IT group, respectively ($P = 0.647$, $P = 0.393$, $P = 0.505$). The mean age, body weight, and height for adolescents at the time of surgery were 14.7 years old, 57.19 kg, and 168.37 cm in the LHSL group and 14.9 years old, 57.96 kg and 169.21 cm in the IT group, respectively ($P = 0.099$, $P = 0.061$, $P = 0.059$). The male/female ratio was 5.1:1 (1268/248) in the LHSL group and 4.9:1 (1241/249) in the IT group ($P = 0.795$). The side ratio of inguinal hernia (right/left/bilateral) was about 10:7:8 (602/430/484) in the LHSL group and 3.8:2.8:3.4 (567/422/501) in the IT group ($P = 0.551$). The comorbidities of the male patients included hydrocele (206), cryptorchidism (15), umbilical hernia (12), congenital heart disease (16), and other congenital diseases (25). The comorbidities in the female patients included round ligament cysts (11). There was no significant difference between the groups in postoperative complications including hydrocele ($P = 0.687$), hematoma ($P = 0.061$), surgical site infection ($P = 0.742$), testicular atrophy (not found), and umbilical trocar hernia ($P = 0.585$). There were two cases of recurrence in the IT group and eight in the LHSL group ($P = 0.07$). The frequency of postoperative recurrence of adolescent inguinal hernia was 3.16% (7/221) in the LHSL group, 0 (0/223) in the IT group ($P = 0.008$), and 0 (0/128) in the LHSL subgroup in the IT group ($P = 0.045$).

Conclusion(s): The favorable outcomes of IT, which had a lower recurrence rate than LHSL for adolescent inguinal hernia, demonstrate that this is a reasonable treatment regimen for pediatric inguinal hernia.

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PMC Identifier

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Status

Embase

Institution

(Chu, Chen, Shen, Liu, Sun, Nie, Liu, Cao, Du, Zou, Yuan) Department of Hernia and Abdominal Wall Surgery, Beijing Chaoyang Hospital, Capital Medical University, 5 Jingyuan Road, Shijingshan District, Beijing 100043, China

Publisher

Springer (Singapore)

Year of Publication

2020

17.

Laparoscopic Orchiopexy Versus Open Orchiopexy for Palpable Undescended Testis in Children: A Prospective Comparison Study.

Yang Z., Li S., Zeng H., Yin J., Xu W., Li J., Xie J., Liu C.

Embase

Journal of Laparoendoscopic and Advanced Surgical Techniques. 30 (4) (pp 453-457), 2020.

Date of Publication: April 2020.

[Article]

AN: 631587948

Background: Can laparoscopic orchiopexy achieve a better testicular position and a higher success rate than open orchiopexy for palpable undescended testis in children? We conducted a prospective comparison study with a large volume of cases to answer this question.

Method(s): A total of 256 patients with palpable undescended testis who were admitted between January 1, 2017 and December 31, 2017 were included in this study. Among them, 124 patients

underwent laparoscopic orchiopexy and 132 patients underwent open inguinal orchiopexy. The outcome evaluated index included final testicular position, success rate, and complications. Result(s): Of 256 patients, the mean age was 2.4 years; 218 patients had unilateral palpable testis, and the other 38 patients had bilateral palpable testis. There were no significant differences between laparoscopic orchiopexy group and open orchiopexy group with respect to age, side, preoperative testicular position, and testicular volume. The final testicular position in laparoscopic group was better than that in open group (lower position rate: 89.3% versus 77.9%, $P = .01$). There was no significant difference in success rate (laparoscopic group: 100%; and open group: 98.5%). There were 3 complications in the laparoscopic group and 6 complications in open group ($P > .05$). No testicular atrophy was found in either group. No testicular ascent occurred in laparoscopic group while there were 2 cases in open group, which required additional surgery for correction. Of patients who underwent surgery at the age of 3 years or older ($n = 75$), laparoscopic surgery was associated with markedly better testicular position than open surgery (lower position rate: 88.1% versus 69.6%, $P = .03$).

Conclusion(s): Laparoscopic orchiopexy is associated with better testicular position and comparable success rate comparing to open orchiopexy for palpable undescended testis in children. This procedure could be recommended for palpable undescended testis, especially in older children.

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PMC Identifier

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Status

Embase

Institution

(Yang, Liu) Department of Urology, Third Affiliated Hospital of Southern Medical University, Tianhe District, Guangzhou, Guangdong 510630, China (Yang, Li, Yin, Xu, Li, Xie) Department of Urology, Laboratory of Pelvic Floor Muscle Function, Shenzhen Children's Hospital, Shenzhen, China

(Zeng) Department of Radiology, Shenzhen Children's Hospital, Shenzhen, China

Publisher

Mary Ann Liebert Inc. (E-mail: info@liebertpub.com)

Year of Publication

2020

18.

Non-palpable testis: is management consistent and objective?.

Mah L.W., Durbin-Johnson B., Kurzrock E.A.

Embase

Journal of Pediatric Urology. 16 (1) (pp 62-68), 2020. Date of Publication: February 2020.

[Article]

AN: 2004512544

Introduction: Diagnostic laparoscopy is recommended for boys with non-palpable testis (NPT) by American and European guidelines with the decision to pursue inguinal exploration based upon testicular vessel appearance. We hypothesized that management decisions are not consistent with the guidelines, and that assessment of vessels is subjective.

Material(s) and Method(s): Pediatric urologist management decisions were evaluated via an electronic survey to determine impact of contralateral testicle size, sonographic findings, surgeon region, and years in practice. In a digital image survey, surgeons were asked to interpret gonadal vessel status of 32 consecutive cases of NPT with absent abdominal testis as normal, atretic, or blind-ending to determine intra- and inter-rater reliability.

Result(s): Of the 339 participants, more Europeans (49%) chose sonography as the first management step for NPT compared to US surgeons (12%). Regardless of sonographic findings, over 80% chose laparoscopy as the first step. In the presence of normal, atretic, and blind-ending vessels, the decisions to proceed with inguinal/scrotal exploration were 88%, 68%, and 17%, respectively. Contralateral hypertrophy and sonography findings had no significant impact on the decision to proceed with inguinal/scrotal exploration. The visual gonadal vessel survey showed surgeon interpretation of normal or blind-ending vessels had moderate inter-rater reliability. Surgeons did not agree on normal status 37% of the time and did not agree on atretic status 66% of the time. There was no statistical difference between European and US respondents ($P = 0.23$). Intra-rater reliability was fair for blind-ending vessels. When the first interpretation was blind-ending, the same surgeon changed interpretation of the same image 39% of the time. There was no statistical difference by years of practice.

Conclusion(s): Non-visualization of NPT on sonography and contralateral testis size had no significant impact upon management decisions. Surgeons chose to pursue inguinal/scrotal exploration based upon laparoscopic gonadal vessel status. However, these interpretations were subjective with low inter- and intra-rater reliability.[Formula presented]

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Status

Embase

Institution

(Mah, Kurzrock) Department of Urologic Surgery, University of California Davis Children's Hospital, Sacramento, CA, United States (Durbin-Johnson) Division of Biostatistics, University of California Davis, Davis, CA, United States

Publisher

Elsevier Ltd

Year of Publication

2020

19.

Revisiting the success rate of one-stage Fowler-Stephens orchiopexy with postoperative Doppler ultrasound and long-term follow-up: a 15-year single-surgeon experience.

Wu C.Q., Kirsch A.J.

Embase

Journal of Pediatric Urology. 16 (1) (pp 48-54), 2020. Date of Publication: February 2020.

[Article]

AN: 2003957994

Introduction: The optimal management of a high undescended testicle (UDT) remains debated. When tethering is due to shortened spermatic vessels, a one-stage or two-stage Fowler-Stephens Orchiopexy (FSO) can be performed. Published series suggest a higher success rate with a two-stage FSO, though its superiority has not been ascertained. The authors examine patient and surgical factors associated with success of one-stage FSO and compare our outcomes with the established literature.

Method(s): We retrospectively reviewed the charts of 43 boys who underwent one-stage FSO for 45 testes from 2003 to 2018 by a single surgeon at our institution. Patient factors, surgical approach, and postoperative results were reviewed. Radiographic success was a testis with intact vascular flow or normal echotexture on scrotal ultrasound. Clinical success was a non-atrophic, intrascrotal testis at postoperative follow-up. Radiographic and clinical success was compared. Data analysis was performed with Fisher's exact test and t-test for categorical and continuous variables respectively.

Result(s): Follow-up (mean 29 months; 0.5 months-14.5 years) was available for 40 patients (42 testes; 21 bilateral, 21 unilateral). Average age at surgery was 21.8 months (4 months-10.4 years). Associated conditions were seen in 14 patients. Twenty-nine patients had postoperative scrotal Doppler ultrasound with radiographic success in 25/29 (86.2%). Overall clinical success was 34/42 (81%) with 4 (9.5%) each of atrophy and ascent. A second surgery to address ascent in four patients allowed correction in three for a success rate of 88%. Radiographic and clinical success was significantly associated ($P = 0.01$). Clinical success was also significantly associated with inguinal rather than laparoscopic testis mobilization ($P = 0.03$) but not to patient's age, associated conditions, unilaterality/bilaterality, or pre-operative hypoplasia or location. Greater than 3 years of follow-up was available in 11 (26.2%) cases, of which seven had successful results. All adverse outcomes occurred within 8 months after surgery. Success was otherwise durable.

Conclusion(s): This is the largest series of one-stage FSO to date. The success rate of one-stage FSO in this series matches that published in the literature for two-stage FSO; equal success is achieved with fewer surgeries. Postoperative Doppler ultrasound demonstrating intact vascular flow or normal testis echotexture is significantly associated with clinical success and highlights the utility of postoperative ultrasound in patients with an equivocal exam.[Formula presented]

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PMC Identifier

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Status

Embase

Institution

(Wu, Kirsch) Children's Healthcare of Atlanta, Emory University School of Medicine, United States

Publisher

Elsevier Ltd

Year of Publication

2020

New insights into the expression of androgen and estrogen receptors of the appendix testis in congenital cryptorchidism.

Sinopidis X., Panagidis A., Kourea E., Kostopoulou E., Rojas-Gil A.P., Skiadopoulos S., Georgiou G., Spiliotis B.E.

Embase

Journal of Pediatric Endocrinology and Metabolism. 33 (4) (pp 503-508), 2020. Date of Publication: 01 Apr 2020.

[Article]

AN: 2005118454

The appendix testis (AT) is the most common vestigial remnant of the human testis. Variations in the presence and expression of AT androgen receptor (AR) and estrogen receptor (ER) have been reported in cryptorchidism. We studied the possible association of AR and ER expression of the AT with cryptorchidism. ATs were resected from 40 boys who underwent inguinoscrotal surgery, (20 patients with congenital unilateral cryptorchidism [UC] and 20 controls with orthotopic testes and hydrocele). AR and ER expression was evaluated with immunohistochemistry, and the percentage and intensity of AR and ER expression were evaluated by the Allred scoring method. AT length was compared between the two groups. Correlation of AR and ER expression was evaluated independently in patients and controls. The Allred score for AR trended toward lower values in UC compared to controls ($p = 0.193$), while ER scores presented statistically significant lower values in UC compared to controls ($p = 0.017$). No significant difference or trend was found in the expression of both receptors between high and low cryptorchidism ($p = 0.981$ for AR, $p = 0.824$ for ER) and for the appendiceal length between UC and controls ($p = 0.369$). The findings of a trend for lower AR expression and a statistically significant lower expression of ER in UC may suggest an association of AR and ER with cryptorchidism and may provide an insight into the process of testicular descent.

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PMC Identifier

32109207 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32109207>]

Status

Embase

Institution

(Sinopidis) Department of Pediatric Surgery, University of Patras, School of Medicine, Rion, Patras 26504, Greece (Panagidis, Georgiou) Department of Pediatric Surgery, Children's Hospital, Patras, Greece

(Kourea) Department of Pathology, University of Patras, School of Medicine, Patras, Greece

(Kostopoulou, Spiliotis) Research Laboratory of the Division of Pediatric Endocrinology and Diabetes, Department of Pediatrics, University of Patras, School of Medicine, Patras, Greece

(Rojas-Gil) Faculty of Health Sciences, Department of Nursing, University of Peloponnese,
Tripoli, Greece

(Skiadopoulos) Department of Medical Physics, University of Patras, School of Medicine, Patras,
Greece

Publisher

De Gruyter (E-mail: peter.golla@degruyter.com)

Year of Publication

2020

21.

Identifying and addressing training deficiencies in the examination of cryptorchidism-a quality improvement study.

Sharma D., Sohn J.K., Zillioux J.M., Herndon C.D.A., Corbett S.T., Kern N.G.

Embase

Journal of Pediatric Urology. 16 (1) (pp 61.e1-61.e8), 2020. Date of Publication: February 2020.

[Article]

AN: 2004235150

Background: Accurate and timely diagnosis of cryptorchidism by primary care providers (PCPs) is critical to oncologic and fertility outcomes. Physical exam is the mainstay of diagnosis, but little is known about PCPs' skills in examining cryptorchid patients. Patients referred to surgeons for cryptorchidism often have normal or retractile testes on exam, and delayed or missed diagnosis of cryptorchidism may contribute to advanced age at surgical intervention. Previous studies on cryptorchidism have not investigated the baseline training, confidence, and/or exam skills of providers.

Objective(s): The authors aimed to define baseline training and provider confidence in the exam of cryptorchid patients and to improve examiner confidence using bedside teaching with a pediatric urologist. Secondly, baseline training and confidence were correlated to skill. Study design: Medical students, family medicine, pediatrics, and urology residents, and pediatric attendings completed surveys on baseline training and self-reported confidence in the examination of cryptorchid patients at an academic institution from 2017 to 2018. N.G.K. (pediatric urologist) proctored examinations of cryptorchid patients and provided standardized grades and individualized feedback. Surveys were readministered after 3 months. Non-

parametric comparison tests were performed to determine intervention effect and compare subgroups.

Result(s): Ninety-two respondents participated. 62% reported little to no formal training on the scrotal exam, 50% were self-taught, and 20% defined undescended testis incorrectly. Confidence increased with level of training, comparing attendings to residents to students ($P < 0.001$). Those who learned from a mentor had higher baseline confidence than those who did not ($P < 0.01$). Baseline confidence and amount of formal training positively correlated with exam skill as graded during proctored sessions ($n = 59$, $P < 0.01$). Provider confidence was higher after proctored exams (Fig. 2, $n = 32$, $P < 0.0001$).

Discussion(s): Significant training deficiencies exist in the examination of cryptorchid patients. A single proctored exam with a pediatric urologist can improve provider confidence and may improve exam skills. A rotation with pediatric urology, including proctored exams of cryptorchid patients, has become standard practice for pediatric trainees at the authors institution as a result of this study.

Conclusion(s): While further studies are required to assess the effectiveness of bedside teaching and its impact on accurate and timely diagnosis of cryptorchidism, implementation of the authors quality improvement recommendations at other teaching institutions would help address training deficiencies in the examination of cryptorchid patients.[Formula presented]

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PMC Identifier

31848108 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31848108>]

Status

Embase

Institution

(Sharma, Zillioux, Corbett, Kern) University of Virginia School of Medicine, Department of Urology, PO Box 800422, Charlottesville, VA 22908, United States (Sohn) University of Virginia School of Medicine, Department of Pediatrics, PO Box 801355, Charlottesville, VA 22908, United States

(Herndon) Children's Hospital of Richmond at Virginia Commonwealth University, Department of Urology, 8700 Stony Point Parkway, Richmond, VA 23235, United States

Publisher

Elsevier Ltd

Year of Publication

2020

22.

Hypogonadism and Cryptorchidism.

Rodprasert W., Virtanen H.E., Makela J.-A., Toppari J.

Embase

Frontiers in Endocrinology. 10 (no pagination), 2020. Article Number: 906. Date of Publication: 15 Jan 2020.

[Review]

AN: 630727755

Congenital cryptorchidism (undescended testis) is one of the most common congenital urogenital malformations in boys. Prevalence of cryptorchidism at birth among boys born with normal birth weight ranges from 1.8 to 8.4%. Cryptorchidism is associated with a risk of low semen quality and an increased risk of testicular germ cell tumors. Testicular hormones, androgens and insulin-like peptide 3 (INSL3), have an essential role in the process of testicular descent from intra-abdominal position into the scrotum in fetal life. This explains the increased prevalence of cryptorchidism among boys with diseases or syndromes associated with congenitally decreased secretion or action of androgens, such as patients with congenital hypogonadism and partial androgen insensitivity syndrome. There is evidence to support that cryptorchidism is associated with decreased testicular hormone production later in life. It has been shown that cryptorchidism impairs long-term Sertoli cell function, but may also affect Leydig cells. Germ cell loss taking place in the cryptorchid testis is proportional to the duration of the condition, and therefore early orchiopexy to bring the testis into the scrotum is the standard treatment. However, the evidence for benefits of early orchiopexy for testicular endocrine function is controversial. The hormonal treatments using human chorionic gonadotropin (hCG) or gonadotropin-releasing hormone (GnRH) to induce testicular descent have low success rates, and therefore they are not recommended by the current guidelines for management of cryptorchidism. However, more research is needed to assess the effects of hormonal treatments during infancy on future male reproductive health.

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Status

Embase

Institution

(Rodprasert, Virtanen, Makela, Toppari) Research Centre for Integrative Physiology and Pharmacology, Institute of Biomedicine, University of Turku, Turku, Finland (Rodprasert, Virtanen, Makela, Toppari) The Population Research Centre, University of Turku, Turku, Finland

(Toppari) Department of Pediatrics, Turku University Hospital, Turku, Finland

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2020

23.

A Faster and Simpler Operation Method for Laparoscopic Inguinal Hernia Repair in Children.

Wang K., Cai J., Lu Y.-C., Li X.-W., Zhai G.-M., He W.-B., Guo X.-S., Wu G.-Q.

Embase

Journal of laparoendoscopic & advanced surgical techniques. Part A. (no pagination), 2020. Date of Publication: 27 Mar 2020.

[Article]

AN: 631362320

Background: Inguinal hernias (IHs) are common in infants and children. The key step in inguinal hernia repair is high ligation of the hernia sac. The current main treatment methods for IHs are open and laparoscopic surgery. Over the past two decades, laparoscopic herniorrhaphy has increased in popularity. Herein, we introduced a new method to laparoscopically treat IHs. The goal of this study was to investigate the clinical effects and advantages of this new operation technique for IHs, which is called the "hernia sac ligation by single-incision laparoscopic surgery with a double-line band method."

Patients and Methods: We retrospectively reviewed the records of all children who underwent initial laparoscopic herniorrhaphy at our center over a 1-year period. A single surgeon performed all surgeries using the modified single-incision laparoscopic technique. Intraoperative findings and complications, operative times, and postoperative complications were reviewed for all children.

Result(s): All 119 surgeries were successfully completed (58 bilateral and 61 unilateral). In total, 54 out of 58 children had contralateral openings discovered at time of surgery and underwent unplanned bilateral laparoscopic hernia repair. This clinical study included 99 boys patients and 20 girls patients (boy-to-girl ratio was 4.95:1). The age range at the time of surgery was 0.5 to 10 years, and the average age was 2.63 years. No patient had any intraoperative complication. Postoperative complications occurred in 1 boy (0.56%) who had a hernia recurrence that required open repair. The addition of auxiliary operating forceps was required for 8 boys (6.72%). No child

had scrotum edema, wound infection, stitch granuloma, or iatrogenic cryptorchidism. Overall, a 93.3% operative success rate was noted with the modified technique.

Conclusion(s): The modified technique is a safe and effective operation method, which can significantly shorten the operation time, reduce recurrence rates, and result in minimal scarring. Additionally, the procedure is expected to be less expensive.

PMC Identifier

32216722 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32216722>]

Institution

(Wang, Lu, Li, Zhai, He, Guo, Wu) Department of Pediatric Surgery, Dongguan Maternal and Child Health Hospital, Dongguan, Guangdong, China (Cai) Reproductive Medicine Center, Dongguan Maternal and Child Health Hospital, Dongguan, Guangdong, China

Publisher

NLM (Medline)

Year of Publication

2020

24.

Laparoscopic orchiopexy of palpable undescended testes_experience of a single tertiary institution with over 773 cases.

You J., Li G., Chen H., Wang J., Li S.

Embase

BMC Pediatrics. 20 (1) (no pagination), 2020. Article Number: 124. Date of Publication: 16 Mar 2020.

[Article]

AN: 631232651

Background: Discuss the superiority of laparoscopic orchiopexy in the treatment of inguinal palpable undescended testes.

Method(s): Inclusion criteria: Preoperative examination and color Doppler ultrasound examination confirmed that the testes were located in the inguinal canal and could not be pulled into the scrotum, except for retractive and ectopic testes. The surgical steps were depicted as follow. The retroperitoneal wall was carved by ultrasonic scalpels, separates the spermatic vessels closed to the inferior pole of the kidney if necessary, dissects the peritoneum of vas deferens, cuts the

testicular gubernaculum, and pulls back the testicle into the abdominal cavity. Besides, protect the vas deferens, and descend the testes to the scrotum and fix them without tension.

Result(s): There were 773 patients with 869 inguinal undescended palpable testes, 218 cases on the left side, 459 cases on the right side and 96 cases with bilateral undescended testes, whose age ranged from 6 months to 8 years, with an average of 20 months. All testes were successfully operated, no converted to open surgery. The average operation time was (34.8 +/- 5.4) min.

There were 692 testes have an ipsilateral patent processus vaginalis (89.5%); In 677 cases of unilateral cryptorchidism, 233 cases (34.4%) have a contralateral patent processus vaginalis, and laparoscopic percutaneous extraperitoneal closure the hernia sac carry out during the surgery.

There was no subcutaneous emphysema during the operation, no vomiting, no abdominal distension, no wound bleeding and obvious pain after surgery, especially wound infection is rarely. Doppler ultrasound was evaluated regularly after surgery. The patients were followed up for 6 to 18 months. All the testes were located in the scrotum without testicular retraction and atrophy. No inguinal hernia or hydrocele was found in follow-up examination.

Conclusion(s): Laparoscopic orchiopexy manage inguinal palpable cryptorchidism is safe and effective, and there are obvious minimally invasive advantages. Furthermore, It could discover a contralateral patent processus vaginalis, and treat at the same time, which avoid the occurrence of metachronous inguinal hernia.

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PMC Identifier

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Status

Embase

Institution

(You, Li, Chen, Wang, Li) Department of Pediatric Urology Surgery, Wuhan Children's Hospital (Wuhan Maternal and Child Healthcare Hospital), Tongji Medical College, Huazhong University of Science and Technology, No.100, Hong Kong Road, Jiang'an District, Wuhan 430016, China

Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2020

5alpha-Reductase type 2 deficiency in families from an isolated Andean population in Venezuela.
Avendano A., Gonzalez-Coira M., Paradisi I., Rojas A., Da Silva G., Gomez-Perez R., Ceballos J.O.

Embase

Annals of Human Genetics. 84 (2) (pp 151-160), 2020. Date of Publication: 01 Mar 2020.

[Article]

AN: 2003479473

5alpha-Reductase type 2 deficiency causes a 46,XY disorder of sex development (DSD) characterized by ambiguous external genitalia, rudimentary prostate, and normal internal genitalia. The disease prevalence worldwide is low, but in a small and isolated village of the Venezuelan Andes, a higher incidence has been found. DNA analysis of the SRD5A2 gene was performed in three inbred affected individuals clinically diagnosed with DSD. The entire coding regions, the p.L89V polymorphism (rs523349) and five intragenic SNPs (rs2300702, rs2268797, rs2268796, rs4952220, rs12470196) used to construct haplotypes were analyzed by Sanger sequencing. To assess the probable ethnic origin of the mutation in this geographic isolate, a population structure analysis was performed. Homozygosity for the p.N193S mutation was found in all patients, with a mutation carrier frequency of 1:80 chromosomes (0.0125) in the geographic focus, suggesting a founder phenomenon. The results of the population structure analysis suggested a mutation origin closer to the Spanish populations, according to the clusters grouping. The genotype-phenotype correlation in the patients was not absolute, being hypospadias and cryptorchidism the main traits that differentiate affected individuals.

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Status

Embase

Author NameID

Paradisi, Irene; ORCID: <http://orcid.org/0000-0001-9039-8945>

Institution

(Avendano, Gonzalez-Coira, Da Silva) Medical Genetics Unit, Medicine Faculty, Los Andes University, Merida, Venezuela (Paradisi) Venezuelan Institute for Scientific Research (IVIC), Laboratory of Human Genetics, Caracas, Venezuela

(Rojas) National Center for Scientific Calculation (CeCalcULA), Los Andes University, Merida, Venezuela

(Gomez-Perez) Endocrinology Service, Los Andes University Hospital (I.A.H.U.L.A.), Merida, Venezuela

(Ceballos) Andrology Laboratory, Los Andes University, Electronic Microscopic Center, Merida, Venezuela

Publisher

Blackwell Publishing Ltd

Year of Publication

2020

26.

The pediatric patient and future fertility: optimizing long-term male reproductive health outcomes.

Nassau D.E., Chu K.Y., Blachman-Braun R., Castellan M., Ramasamy R.

Embase

Fertility and Sterility. 113 (3) (pp 489-499), 2020. Date of Publication: March 2020.

[Review]

AN: 2005226288

Globally, male factor infertility accounts for 20%-70% of couples struggling to conceive. Certain male pediatric developmental conditions, such as cryptorchidism, hypospadias, testicular and other childhood cancers, infections, and pediatric varicocele have been associated with future infertility. Early fertility preservation, especially in those with pending chemotherapy or genetic conditions such as Klinefelter syndrome, should be strongly considered in patients expected to experience testicular loss. Although optimal treatment timing may be unknown owing to a paucity of long-term prospective studies, early diagnosis and targeted treatment may optimize fertility potential in adulthood.

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PMC Identifier

32192588 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32192588>]

Status

Embase

Institution

(Nassau) Department of Urology, Lenox Hill Hospital, Donald and Barbra Zucker School of Medicine at Hofstra/Northwell, New York, NY, United States (Chu, Blachman-Braun, Ramasamy) Department of Urology, University of Miami Miller School of Medicine, Miami, FL, United States

(Castellan) Division of Pediatric Urology, Nicklaus Children's Hospital, Miami, FL, United States

Publisher

Elsevier Inc. (E-mail: usjcs@elsevier.com)

Year of Publication

2020

27.

Transscrotal transverse incision for the treatment of middle and low cryptorchidism in children: experience from 796 cases.

Wang Y.-J., Chen L., Zhang Q.-L., Lin Y., Cui X., Chen J.-C., Zhou C.-M.

Embase

BMC surgery. 20 (1) (pp 51), 2020. Date of Publication: 17 Mar 2020.

[Article]

AN: 631276074

BACKGROUND: The purpose of this study was to summarize our clinical experience with transscrotal transverse incision in the treatment of low and middle cryptorchidism in children.

METHOD(S): A total of 796 children with low or middle cryptorchidism participated in this study from March 2012 to May 2018. Transscrotal transverse incision was used to treat low and middle cryptorchidism. Symptoms and signs were followed up at 1 week, 1 month, 3months and every six to 12months thereafter.

RESULT(S): Testicular descent fixation through transverse scrotal incision was successfully performed in all 796 children. All patients were discharged 1-2days after the operation. During hospitalization and follow-up, 35 patients had complications, including 7 cases of cryptorchidism recurrence, 5 cases of poor scrotal incision healing, and 23 cases of scrotal haematoma. There were no complications, such as bladder injury, testicular atrophy, inguinal hernia or hydrocele.

CONCLUSION(S): Transscrotal transverse incision is a safe and feasible method for the treatment of middle and low cryptorchidism. It has the advantages of less trauma and an aesthetic scar after operation.

PMC Identifier

32183767 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32183767>]

Institution

(Wang, Chen, Zhang, Lin, Cui, Chen, Zhou) Department of Pediatric Surgery, Fujian Maternity and Child Health Hospital, Fuzhou 350001, China

Publisher

NLM (Medline)

Year of Publication

2020

28.

The male genital system.

Wu W.J., Gitlin J.S.

Embase

Pediatrics in Review. 41 (3) (pp 101-111), 2020. Date of Publication: 01 Mar 2020.

[Article]

AN: 2005208079

* Male differentiation relies on the presence of the Y chromosome and the action of testosterone on bipotent genital structures. * Hypospadias is a commonly encountered disorder of the penis. Further evaluation is prudent in cases associated with undescended testis based on an observational study. (12) * By consensus, micropenis is defined as 2.5 SD or more below the mean stretched length for a patient's age. Endocrinologic evaluation should be instituted once this diagnosis is made. More benign entities, including webbed penis and buried penis, should be ruled out by proper measurement of the penis. Treatment with testosterone supplementation is effective based on observational studies. (17)(18)(19) * Physiologic phimosis resolves over time, with the prepuce becoming retractile in most patients by 4 years of age. Paraphimosis is an emergency condition that requires immediate medical attention. Complications from the prepuce may be treated medically or surgically. * Male circumcision has medical benefits, such as decreased risk of urinary tract infection, penile cancer, and contracting human immunodeficiency virus based on strong data from meta-analysis of the present literature. (24) * Penile trauma is an uncommon clinical entity. A thorough history should be obtained to ensure that the mechanism of injury is consistent with the physical injury. * Cryptorchidism is a common condition that requires close followup. Referral to a surgical specialist is recommended should a testis fail to descend by 6 months of age. There is strong evidence based on a literature review that surgical correction helps lower the risk of testicular malignancy in cryptorchid testes. (35) * By consensus, testicular

torsion is an emergency condition that requires rapid diagnosis and management. This condition should be considered in all patients with acute scrotal pain. * Varicoceles are of clinical concern because of possible future infertility. Its management in the adolescent population is still a subject of great debate. * Epididymo-orchitis may mimic testicular torsion because patients present with similar symptoms of acute scrotal pain and swelling. Duplex ultrasonography is helpful in making the proper diagnosis. * Based on clinical studies, cancer of the testis is uncommon in the pediatric population. A slow-growing, nontender mass should raise concern for an intrascrotal malignancy. Referral to a urologist is indicated for surgical resection once the diagnosis is made. * Trauma to the testis may lead to testicular rupture. Early surgery is indicated to prevent testicular atrophy when rupture is suspected or when there is penetrating trauma. Conservative management is acceptable in select populations based on an observational study. (68).

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PMC Identifier

32123021 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32123021>]

Status

Embase

Institution

(Wu) Smith Institute for Urology, Zucker School of Medicine, Lake Success, NY, United States

(Gitlin) Cohen Children's Medical Center, Division of Pediatric Urology, Zucker School of

Medicine, New Hyde Park, NY, United States

Publisher

American Academy of Pediatrics (141 Northwest Point Blvd, P.O. Box 927, Elk Grove Village IL 60007-1098, United States)

Year of Publication

2020

29.

Cryptorchidism in Children with Zika-Related Microcephaly.

de Vasconcelos R.A.L., Ximenes R.A.A., Calado A.A., Martelli C.M.T., Goncalves A.V., Brickley

E.B., de Araujo T.V.B., Wanderley Rocha M.A., Miranda-Filho D.B.

Embase

The American journal of tropical medicine and hygiene. (no pagination), 2020. Date of Publication: 09 Mar 2020.

[Article]

AN: 631227978

The genitourinary tract was recently identified as a potential site of complications related to the congenital Zika syndrome (CZS). We provide the first report of a series of cryptorchidism cases in 3-year-old children with Zika-related microcephaly who underwent consultations between October 2018 and April 2019 as part of the follow-up of the children cohort of the Microcephaly Epidemic Research Group, Pernambuco, Brazil. Of the 22 males examined, eight (36.4%) presented with cryptorchidism. Among 14 undescended testis cases, 11 (78.6%) could be palpated in the inguinal region. Seven of the eight children had severe microcephaly. Conventional risk factors for cryptorchidism were relatively infrequent in these children. We hypothesize that cryptorchidism is an additional manifestation of CZS present in children with severe microcephaly. As in our cases, for most of the children, the testes were located in the inguinal region, and the possible mechanisms for cryptorchidism were gubernaculum disturbance or cremasteric abnormality.

PMC Identifier

32157994 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32157994>]

Institution

(de Vasconcelos, Ximenes, Calado, Goncalves, Wanderley Rocha, Miranda-Filho) Universidade de Pernambuco, Recife, Brazil (Ximenes, Goncalves, de Araujo) Universidade Federal de Pernambuco, Recife, Brazil

(Martelli) Instituto de Pesquisa Aggeu Magalhaes, Fiocruz, Recife, Brazil

(Brickley) London School of Hygiene and Tropical Medicine, London, United Kingdom

Publisher

NLM (Medline)

Year of Publication

2020

30.

Detection of monorchidism in boys with unilateral undescended testes: clinical benefits and limitations of contralateral testicular size.

Huang Y., Liu P., Sun N., Zhang W., Song H.

Embase

Journal of Pediatric Urology. (no pagination), 2020. Date of Publication: 2020.

[Article]

AN: 2005190186

Introduction: Contralateral testicular size was recommended as an effective measurement in prediction of monorchidism in some previous studies but a few argued it as invalid. Further investigation was demanded.

Objective(s): To investigate the effectiveness of contralateral testicular size in prediction of monorchidism in patients with unilateral non-palpable undescended testes (NPT) aged between 9 and 48 months.

Material(s) and Method(s): Total of 707 patients aged between 9 and 48 months and diagnosed with unilateral undescended testes (UDT) between January 2016 and December 2018 at the study department were enrolled. In accordance with physical examinations and surgical findings, patients were divided into three groups: palpable UDT (group A, n = 609), non-palpable but viable testes (group B, n = 57) and monorchidism (group C, n = 41). Contralateral testicular length and volume were evaluated with ultrasonography. Comparison of contralateral testicular size between three groups and calculation of optimal cut-off value and diagnostic performance of it among NPT were performed.

Result(s): The length and volume of contralateral testes of group C were larger than of group A ($P < 0.01$) and group B ($P < 0.01$), whereas these differences between groups were small. Among patients with NPT, a receiver operating characteristic curve was used to determine the optimal cut-off value. It revealed that both a testicular length of 17.5 mm and a volume of 1.05 ml provided the highest Youden's index for prediction of monorchidism. The sensitivity and specificity for testicular length were 34.1% and 94.7%, and volume were 34.1% and 93%, respectively. The predictive accuracy for testicular length and volume were 69.4% and 65.7%, respectively. Even though the negative predictive value was merely 66.6% (54/81) and 66.2% (53/80), the positive predictive value (PPV) reaches to 82.3% (14/17) and 77.7% (14/18) for testicular length and volume.

Discussion(s): Several factors including choosing of measurement tools, age range, ethnicity, and selection bias of cohorts may be accounted for the huge differences among cut-off values and predictive accuracy. The diagnostic performance of contralateral testicular size in prediction of monorchidism in patients with NPT was poor. But the PPV was relatively promising. Contralateral testicular hypertrophy can provide information for surgical planning.

Conclusion(s): The overall diagnostic performance of contralateral testicular size in prediction of monorchidism in patients with UDT aged between 9 and 48 months was poor. But the efficiency of cut-off value predicting absence of viable testes was relatively higher. This value should be

objectively applied but only as a reference which would not be a complete replacement of laparoscopy exploration.[Formula presented]

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PMC Identifier

32165086 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32165086>]

Status

Article-in-Press

Institution

(Huang, Liu, Sun, Zhang, Song) Department of Pediatric Urology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing 100045, China

Publisher

Elsevier Ltd

Year of Publication

2020

31.

'Testis-epididymis dissociation' in cryptorchidism and hydrocele: the tip of the iceberg of a persistent genital mesentery.

Mentessidou A., Mirilas P.

Embase

Surgical and Radiologic Anatomy. (no pagination), 2020. Date of Publication: 2020.

[Article]

AN: 2004382991

Purpose: To investigate whether testis-epididymis dissociation encountered in boys with cryptorchidism/hydrocele is related with an abnormal persistence of the fetal mesentery of testis and associated ducts.

Method(s): We examined the morphology of peritoneal folds of the testis, epididymis, and vas deferens in 25 boys operated for unilateral cryptorchidism [inguinal (n = 20), intrabdominal (n = 5)] and 20 boys operated for unilateral communicating hydrocele. Findings were compared with the normally persisting genital mesentery of rats (n = 30, both sides), a known animal model of the genital mesentery of human fetuses, as well as with the normal mature pattern of genital peritoneal folds in adult male cadavers (n = 12, both sides). Rats before testis descent [aged 18

days (n = 15)] served for comparison with boys with cryptorchidism, while rats after testis descent [aged 50 (n = 15)] known to retain patent processi vaginales for comparison with boys with hydrocele.

Result(s): A well-developed genital mesentery, identical to the fetal-type genital mesentery in the rat, was documented in cryptorchidism and hydrocele. The peritoneum enveloped the testis, epididymis, and vas deferens, and formed wide ligaments between testis-epididymis, epididymis-vas deferens, and vas-posterior wall; processus vaginalis was patent in all cases. The testis-epididymis ligament was related with testis-epididymis distancing, the so-called testis-epididymis dissociation. On the contrary, genital mesentery had involuted in the adult male cadavers, except for a small portion of testis-epididymis ligament corresponding to the so-called sinus epididymis.

Conclusion(s): The testis-epididymis dissociation encountered in cryptorchidism/hydrocele is part of an anomalously persisting fetal-type genital mesentery.

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Status

Article-in-Press

Author NameID

Mentessidou, Anastasia; ORCID: <http://orcid.org/0000-0003-1428-2312>

Institution

(Mentessidou, Mirilas) Department of Pediatric Surgery, Aghia Sophia Children's Hospital, Thivon St. and Papadiamontopoulou St., Athens 11527, Greece (Mentessidou) Department of Pediatric Surgery, Chelsea Children's Hospital, Chelsea and Westminster Hospital NHS Foundation Trust, London, United Kingdom

(Mirilas) Centers for Surgical Anatomy and Technique, Emory University School of Medicine, Atlanta, GA, United States

Publisher

Springer (E-mail: springer@springer.it)

Year of Publication

2020

Prune-belly syndrome in Africa: An analysis and systematic review of cases, etiology, treatment, and outcomes.

Keet K., Henry B.M., Tubbs R.S.

Embase

Journal of Clinical Urology. (no pagination), 2020. Date of Publication: 2020.

[Review]

AN: 2004411794

Background: Prune-belly syndrome is a rare congenital disorder characterized by a spectrum of three anomalies: bilateral undescended testes, dilated urinary tract, and anterior abdominal muscle deficiency.

Objective(s): In developing countries, inadequate access to health care may affect treatment and outcomes of prune-belly syndrome. This study's goal was to review the anatomical features, etiology, genetics, management, and outcomes of cases in Africa.

Method(s): PubMed was searched to identify case reports and case studies describing prune-belly syndrome in Africa. Data collected from each study included the number of cases, age at diagnosis, sex, description of the abdominal muscles, testes, and urinary tract, as well as associated anomalies, management, and long-term outcomes.

Result(s): A total of 16 publications that reported 58 cases in African countries were included. The prevalence of female patients (15.5%) was higher than in developed countries (3%). The abdominal muscles were deficient in all cases, and bilateral cryptorchidism was present in nearly all males (96%). Distension of the bladder was common, with normal anatomy reported in only one case. Bilateral hydronephrosis and hydronephrosis also were present in the majority of cases. Only six cases (10.3%) had no associated anomalies, such as musculoskeletal or cardiovascular. Karyotyping was performed in only three cases (5.2%) because of limited hospital facilities. Six parents (10.3%) declined treatment for their children, 12 cases (20.7%) were managed conservatively, and 25 (43.1%) received surgical intervention. Patients' mortality rate was higher than in developed countries.

Conclusion(s): Diagnosis and treatment of prune-belly syndrome remains a challenge in Africa, in which multiple factors, such as access to health care and cultural beliefs, affect mortality rates and outcomes. Patient education and support groups may improve compliance with treatment.

Level of Evidence: Not applicable for this multicenter audit.

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Status

Article-in-Press

Author NameID

Keet, Kerri; ORCID: <http://orcid.org/0000-0003-3513-9232>

Institution

(Keet) Division of Clinical Anatomy, Department of Biomedical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University, South Africa (Henry) Cardiac Intensive Care Unit, The Heart Institute, Cincinnati Children's Hospital Medical Center, United States

(Tubbs) Departments of Neurosurgery and Structural and Cellular Biology, Tulane University School of Medicine, United States

(Tubbs) Department of Anatomical Sciences, St. George's University, Grenada

Publisher

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Year of Publication

2020

33.

Fertility in adult men born with hypospadias: A nationwide register-based cohort study on birthrates, the use of assisted reproductive technologies and infertility.

Skarin Nordenvall A., Chen Q., Norrby C., Lundholm C., Frisen L., Nordenstrom A., Almqvist C., Nordenskjold A.

Embase

Andrology. 8 (2) (pp 372-380), 2020. Date of Publication: 01 Mar 2020.

[Article]

AN: 2003653100

Background: Fertility in men with hypospadias may be affected due to anatomical, surgical, or etiological factors and associated conditions. Fertility is further influenced by psychosocial and genetic factors, often shared within families.

Objective(s): To evaluate fertility in men born with hypospadias and assess confounding by familial factors.

Material(s) and Method(s): A population-based cohort of 1.2 million men born in Sweden 1964-1998, identified through national demographic and healthcare registers. Associations between hypospadias and (a) being a biological father, (b) conceiving through ART, and (c) diagnosis of male infertility were investigated in the full cohort with logistic regression models and Cox proportional hazard models, expressed as odds ratios (ORs) and hazard ratios (HRs), respectively, with 95% confidence intervals (CIs). A stratified proportional hazard model, conditional on sibling group, was used to control for shared familial confounding.

Result(s): Men with hypospadias, as a whole group, had a lower probability of having biological children (adjusted HR 0.87, 95% CI 0.83-0.92). A significant association was present in both distal (adjusted HR 0.90, 95% CI 0.85-0.96) and proximal hypospadias (HR 0.59, 95% CI 0.42-0.81). Men with hypospadias more often became fathers through ART, regardless of concomitant cryptorchidism. The initial association between hypospadias and the diagnosis of infertility disappeared in sensitivity analyses excluding cryptorchidism.

Discussion(s): Men with hypospadias displayed lower birthrates as compared to their brothers and the general population. Mere birthrates may, however, be a questionable measure of fertility in a population using family planning. However, men with hypospadias were also at higher risk of reproducing through ART and did more often receive a diagnosis of male infertility. Altogether, these findings indicate impaired fertility in men with hypospadias.

Conclusion(s): Fertility in men with hypospadias is impaired, as shown by lower birthrates, increased use of ART and higher risk of receiving a diagnosis of male infertility.

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Status

Embase

Author NameID

Skarin Nordenvall, Anna; ORCID: <http://orcid.org/0000-0003-3034-9222>

Institution

(Skarin Nordenvall, Nordenstrom, Nordenskjold) Department of Women's and Children's Health, Karolinska Institutet, Stockholm, Sweden (Skarin Nordenvall, Chen, Norrby, Lundholm, Almqvist) Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Stockholm, Sweden

(Frisen) Department of Clinical Neuroscience, Karolinska Institutet, Stockholm, Sweden

(Frisen) Child and Adolescent Psychiatry Research Center, Stockholm, Sweden

(Nordenstrom) Department of Pediatric Endocrinology, Astrid Lindgren Children's Hospital, Karolinska University Hospital, Stockholm, Sweden

(Almqvist) Pediatric Allergy and Respiratory Medicine, Astrid Lindgren Children's Hospital, Karolinska University Hospital, Stockholm, Sweden

(Nordenskjold) Center for Molecular Medicine, Karolinska Institutet, Stockholm, Sweden

(Nordenskjold) Department of Pediatric Surgery, Astrid Lindgren Children's Hospital, Karolinska University Hospital, Stockholm, Sweden

Publisher

Blackwell Publishing Ltd

Year of Publication

34.

Epidemiology, Diagnosis and Therapeutic Approaches of Cryptorchidism at the Panzi General Hospital, DR Congo: A 5-year Retrospective Study.

Desire A.M., Buhendwa C., Cesaire T.M.A., Prisca K.I.S., Levi L.N., David N.B., Longombe A.O., Pierre W.

Embase

Ethiopian journal of health sciences. 30 (1) (pp 107-114), 2020. Date of Publication: 01 Jan 2020.

[Article]

AN: 631130665

Background: Cryptorchidism is a common congenital malformation characterized by unilateral or bilateral undescended testis in the scrotum. It is a common disease in pediatric urology although the prevalence is yet to be determined in the East DR Congo. The aim of this study was to describe the prevalence, clinical and therapeutic features of cryptorchidism at the Panzi General Hospital.

Method(s): Medical records of patients who underwent pediatric surgery between January 2011 and December 2016 were reviewed. The prevalence of cryptorchidism, associated malformations, sociodemographic features of patients and therapeutic protocols were examined. Collected data were analyzed using SPSS software.

Result(s): A total of 5066 children were received during the study period in the service, of which 76 suffered from cryptorchidism giving a prevalence of 1.50% and an annual incidence of 12.70. Unilateral cases were found in 53(69.74%) patients among which 66.04% and 33.96% had unilateral right and left cryptorchidism respectively. Cryptorchidism was associated with other malformations in 71.1% of patients. The age of patients ranged from 0 to 15 years and 78.9% were older than two years. Surgery was the only therapeutic approach and fixation of cryptorchid testis in dartos through inguinal incision was the preferred therapeutic used method.

Conclusion(s): This study indicates that cryptorchidism is a relatively common and neglected disease in the Est-DR Congo, due to late consultation. Therefore, the local population and pediatricians should be sensitized on scrotal palpation techniques in order to shorten the consultation delay and prevent testis damages.

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PMC Identifier

32116439 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32116439>]

Institution

(Desire, Cesaire, Pierre) Research Unit of Animal Physiology and Phytopharmacology (URPAP), Faculty of Science, University of Dschang, Cameroon (Desire, Buhendwa, Prisca, Levi, David, Longombe) School of Medicine and Community Health, Evangelical University in Africa, DR Congo

Publisher

NLM (Medline)

Year of Publication

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35.

Proximal Hypospadias: Isolated Genital Condition or Marker of More?.

Johnson E.K., Jacobson D.L., Finlayson C., Yerkes E.B., Goetsch A.L., Leeth E.A., Cheng E.Y.

Embase

The Journal of urology. (pp 101097JU0000000000000810), 2020. Date of Publication: 24 Feb 2020.

[Article]

AN: 631044348

PURPOSE: The prevalence of endocrine/genetic anomalies among boys with proximal hypospadias is unknown. This study aims to describe the endocrine/genetic evaluation for boys with proximal hypospadias to inform who may have a difference/disorder of sex development and/or benefit from additional testing.

METHOD(S): Boys with hypospadias seen at our hospital from 1/2013-10/2018 were retrospectively reviewed. Those with proximal (penoscrotal, scrotal, perineal) hypospadias who presented at <6 months old and underwent endocrine/genetic testing were included.

Demographics, test results, testicular exam at presentation, comorbidities, and diagnoses were abstracted.

RESULT(S): 1789 boys with hypospadias were identified. Of 131 boys with proximal hypospadias, all 60 who underwent endocrine/genetic evaluation were included. Most had bilateral palpable testes (52/60, 86%) that were fully descended (41/60, 68%). Associated

anatomic anomalies were found in 53%. All boys underwent endocrine testing, which was completely typical for an infant male in most (41/60, 68%). Common genetic tests included karyotyping (100%), Microarray (38%), and multi-gene panel (13%). Genetic anomalies were found in 17 boys (28%): 7/41 (17%) with bilateral descended testes and 10/19 (53%) with ≥ 1 undescended testis ($p=0.01$). Most boys (6/8) with ≥ 1 non-palpable testis had a genetic anomaly (vs. 11/52 with bilateral palpable testes; $p=0.005$). Differences/disorders of sex development were found in 9 (15%).

CONCLUSION(S): Among 60 boys with proximal hypospadias, 53% had non-genital anomalies, 28% had genetic anomalies, and 15% had a difference/disorder of sex development. Although endocrine testing was clinically useful, genetic testing was most diagnostically revealing. Endocrine/genetic evaluation should be considered for boys with proximal hypospadias.

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Institution

(Johnson, Jacobson, Yerkes, Cheng) Department of Urology, Northwestern University, Evanston, IL, United States (Johnson, Jacobson, Yerkes, Cheng) Division of Pediatric Urology, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, Mexico

(Finlayson, Goetsch, Leeth) Department of Pediatrics, Northwestern University, Evanston, IL, United States

(Finlayson) Division of Pediatric Endocrinology, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, Mexico

(Goetsch) Division of Genetics, Birth Defects & Metabolism, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, Mexico

(Leeth) Department of Pathology and Laboratory Medicine, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, Mexico

Publisher

NLM (Medline)

Year of Publication

2020

Unsatisfactory testicular position after inguinal orchidopexy: Is there a role for upfront laparoscopy?.

Youssef A.A., Marei M.M., Abouelfadl M.H., Mahmoud W.M., Elbarawy A.S.A., Yassin T.Y.M.

Embase

Arab Journal of Urology. 18 (1) (pp 48-53), 2020. Date of Publication: 02 Jan 2020.

[Article]

AN: 2003725211

Objectives: To examine the role of laparoscopy in managing unsatisfactory testicular position after an open inguinal orchidopexy. We hypothesised that testes that were originally peeping, where short vessels represented a difficulty and testes that only reached a high scrotal position under tension, especially after an initial surgery performed with the appropriate expertise, are candidates for initial laparoscopic dissection.

Patients and Methods: Nineteen boys with an initial open inguinal orchidopexy, with a mean age of 31 months, were considered. Twelve were then treated by a laparoscopic-assisted orchidopexy technique. Standard laparoscopy was established and utilised to mobilise the spermatic cord from above, then completed by an open inguinal mobilisation.

Result(s): The mean age at surgery was 26 months. The laparoscopic redo surgery took place at a mean interval of 11.9 months after the initial operation. The mean operative time was 72 min. A good position and size of the testis were achieved in all cases, evidenced by ultrasonography at 6 months postoperatively and clinically thereafter.

Conclusion(s): An upfront combined laparoscopic and inguinal approach to redo orchidopexy for recurrent palpable undescended testes is suitable in selected patients. This study identifies the selection criteria and outlines the operative considerations. This laparoscopic-assisted approach is a safe and feasible way to correct unsatisfactory position of the testis, with diminished risk of injury to the vas and vessels, while gaining the maximum possible length by high retroperitoneal dissection. Abbreviation: UDT: undescended testis/testes.

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Embase

Author NameID

Youssef, Ahmed Abdelhaseeb; ORCID: <http://orcid.org/0000-0001-7288-1255> Elbarawy, Atef

Salaheldin Abdulaziz; ORCID: <http://orcid.org/0000-0001-5062-9315>

Yassin, Tamer Yassin Mohamed; ORCID: <http://orcid.org/0000-0002-8010-2466>

Marei, Mahmoud Marei; ORCID: <http://orcid.org/0000-0002-8670-1830>

Institution

(Youssef) Department of Pediatric Surgery, El-Demerdash Hospital, Faculty of Medicine, Ain Shams University, Cairo, Egypt (Youssef, Elbarawy, Yassin) Department of Pediatric Surgery, King Abdulaziz Medical City, Al-Hasa, Saudi Arabia
(Marei, Abouelfadl, Mahmoud, Yassin) Department of Surgery, Pediatric and Neonatal Surgery Unit, Cairo University Specialized Pediatric Hospital (CUSPH), Faculty of Medicine, Cairo University, Cairo, Egypt
(Marei) The Royal Manchester Children's Hospital and Manchester Royal Infirmary, Manchester University NHS Foundation Trust, Manchester, United Kingdom
(Elbarawy) Department of General Surgery, Faculty of Medicine, Beni-Suef University, Beni-Suef, Egypt
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Taylor and Francis Ltd. (E-mail: michael.wagreich@univie.ac.at)
Year of Publication
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37.

General surgery of childhood in the UK: a general surgeon's perspective.

Gordon A.C., Davenport M.

Embase

Journal of Pediatric Surgery. 55 (2) (pp 213-217), 2020. Date of Publication: February 2020.

[Review]

AN: 2003844104

The future of general surgery of children as practiced in District General (DGHs) and Rural General Hospitals (RGHs) by adult general surgeons and urologists is uncertain. It is likely that this is because of a combination of the overall trend towards specialization, concerns about clinical risk; uncertainty within the profession about the behavior of the regulator and criminal justice system when considering cases of alleged incompetence; reduced and more targeted training time, curriculum changes, and perhaps a concern by other specialties regarding the ability of DGH and RGH surgeons to provide a safe service. The impact of this on regional pediatric surgical units (RPSUs) is however considerable. While transfer of some conditions such as infantile hypertrophic pyloric stenosis and intussusception is justifiable, transfer of others such as undescended testis and suspected torsion is not. Close communication between regional

specialists and local generalists, preferably in the setting of a formal network, together with a change in the priorities of local medical and nonmedical managers and cooperation between competing Trusts is required. Strategies for dealing with the problem are available but require a change in management and National Health Service (NHS) ethos to enact effectively. Adherence to evidence-based best practice with the help of the "Getting It Right First Time (GIRFT)" initiative is vital and, together with targeted publicity and encouragement, the trend may not be irreversible. Level of Evidence: Level V.

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Status

Embase

Institution

(Gordon) Royal Berkshire Hospital, London Road, Reading RG1 5AN, United Kingdom

(Davenport) Kings College Hospital, London SE5 9RS, United Kingdom

Publisher

W.B. Saunders

Year of Publication

2020

38.

Retrospective analysis of testicular outcomes following laparoscopic two-stage Fowler Stephens orchidopexy.

Roy C., Cullis P.S., Clark C., Munro F.D.

Embase

Journal of Pediatric Surgery. 55 (2) (pp 300-303), 2020. Date of Publication: February 2020.

[Article]

AN: 2003843569

Aim of the Study: Few large series report outcomes for laparoscopic two-stage Fowler Stephens orchidopexy (LFSO). The aims were to evaluate testicular outcomes and to identify factors predictive of successful outcome.

Method(s): A retrospective case series of children undergoing laparoscopic surgery for impalpable testes between May 1996 and March 2018 was reviewed. Data were collected from case and operative records. The primary outcomes of interest were testicular atrophy or re-ascent. Regression analysis was conducted to identify factors predictive of successful outcome. Data was expressed as median (IQR).

Result(s): Of 279 patients (300 testes) undergoing laparoscopy for impalpable testis, 114 patients (128 testes) underwent LFSO. Eighty-five patients (96 testes) had adequate follow-up available (53 left; 43 right). Age at first stage was 19 (IQR 13-36) months. Fifteen children had relevant co-morbidities. Time between procedures was 7 (IQR 6-8) months. Longest follow-up available was 12.5 months (IQR 6.8-19.8). Atrophy occurred in 8 testes (8.3%), and ascent occurred in 6 (6.3%). No factors were significantly predictive of success, although a trend towards atrophy was seen amongst testes undergoing gubernacular division compared with a gubernaculum-sparing technique ($p = 0.06$; OR 3; 95% CI 0.97-9.3).

Conclusion(s): A successful outcome was seen amongst 82 of 96 testes (85%) undergoing LFSO, similar to previous reports. No factors were identified that significantly predicted outcome. Number of adverse outcomes was limited (hence possibility of type II error), and therefore preservation of the gubernaculum may reduce risk of testicular atrophy.

Type of Study: Prognosis Study.

Level of Evidence: Level IV.

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PMC Identifier

31753613 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31753613>]

Status

Embase

Institution

(Roy, Cullis, Clark, Munro) Department of Paediatric Surgery, Royal Hospital for Sick Children
Edinburgh, Edinburgh, United Kingdom

Publisher

W.B. Saunders

Year of Publication

2020

The spectrum of testicular-epididymal fusion anomalies in children with cryptorchidism: Personal experience, systematic review and meta-analysis.

Qin K.R., Morley C., Nataraja R.M., Pacilli M.

Embase

Journal of Pediatric Urology. (no pagination), 2020. Date of Publication: 2020.

[Review]

AN: 2004781134

Background: Testicular-epididymal fusion anomalies (TEFA) have been documented in the literature. The pathological significance of TEFA and their relationship to testicular maldescent is unclear. We aimed to clarify the real incidence of TEFA in children with undescended testes and their impact on testicular development after surgery.

Method(s): We conducted a retrospective review (2010-2018) of all patients who underwent orchidopexy. Cases with TEFA confirmed intra-operatively were matched against controls with normal fusion for age at the time of surgery. Records from follow-up visits were assessed to compare testicular size at six-months. A systematic review and meta-analysis of the literature (1980-2019) was also performed.

Result(s): In our retrospective review, 54 (21.4%) of 252 cryptorchid testes were found to have TEFA (Table). Intra-abdominal testes were more likely to exhibit TEFA than inguinal testes (20.4% vs. 9.6%, RR 1.8 [1.0-3.1], P = 0.03). There were no differences in testicular size at the time of surgery (P = 0.29) or the six-month followup (P = 0.18). The systematic review identified eight studies with 4871 children (5240 orchidopexies). The overall rate of TEFA was 25.8% [95% CI 15.2-38.0]. Tail nonfusion (NF) (10.7% [95% CI 5.4-17.4]) was the most common followed by head NF (7.2% [95% CI 3.2-12.5]) and complete NF (6.3% [95% CI 3.7-9.5]). Intra-abdominal testes were more likely to exhibit TEFA than inguinal testes RR 2.6 [95% CI 1.9-3.5]; P < 0.001.

Conclusion(s): Data from our retrospective review and the literature indicate that TEFA are present in approximately one-quarter of cryptorchid testes and are more commonly associated with intra-abdominal cryptorchidism. There appears to be no impact on testicular size at short-term followup. The clinical significance of TEFA remains unclear; long-term followup studies are necessary to better understand their impact on testicular development and function. [Table presented]

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Status

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Institution

(Qin, Morley, Nataraja, Pacilli) Department of Paediatric Surgery, Monash Children's Hospital, Melbourne, Victoria, Australia (Qin, Nataraja, Pacilli) Department of Paediatrics, School of Clinical Sciences at Monash Health, Medicine, Nursing and Health Sciences, Monash University, Melbourne, Victoria, Australia

(Nataraja, Pacilli) Department of Surgery, School of Clinical Sciences at Monash Health, Medicine, Nursing and Health Sciences, Monash University, Melbourne, Victoria, Australia

Publisher

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Year of Publication

2020

40.

Inguinal hernias in children.

Yeap E., Nataraja R.M., Pacilli M.

Embase

Australian journal of general practice. 49 (1-2) (pp 38-43), 2020. Date of Publication: 01 Jan 2020.

[Article]

AN: 630801363

BACKGROUND: An inguinal hernia is one of the most common paediatric surgical presentations in a primary care setting. Hernias can present in multiple ways, ranging from an emergency such as a strangulated hernia to a less urgent reducible hernia.

OBJECTIVE(S): The aim of this article is to aid in appropriate diagnosis and management of hernias in children. The article also provides useful tips for hernia reduction that are especially beneficial in the primary care setting and assist with the identification of hernias that require urgent referral. **DISCUSSION:** Recognising the signs of a hernia containing compromised contents is essential to prevent serious complications such as intestinal perforation, testicular atrophy and ovarian damage. Other common conditions such as hydrocoele and undescended testis are sometimes confused with an inguinal hernia. Young patients under the age of three months and patients with concern for compromised contents require urgent referral. Recent evidence regarding controversial issues in inguinal hernia repair such as the role of laparoscopy and the relevance of a contralateral patent internal inguinal ring will be discussed.

PMC Identifier

32008266 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32008266>]

Institution

(Yeap) Surgical Resident, Department of Paediatric Surgery, Monash Children@s Hospital, (Hons), Vic, Slovenia (Nataraja) MBBS, BSc (Hons), GCCS (Hons), FRCSEd (Paed Surg), FFSTEd, FRACS (Paeds), Consultant Paediatric and Neonatal Surgeon,@ Department of Paediatric Surgery, Monash Children@s Hospital, Vic; Senior Lecturer, Department of Paediatrics, School of Clinical Sciences at Monash Health, Medicine, Nursing and Health Sciences, Monash University, Vic

(Pacilli) MBBS (Hons), MD (Res), FRCS (Paed Surg), Consultant Paediatric and Neonatal Surgeon, Department of Paediatric Surgery, Monash Children@s Hospital, Vic; Senior Research Fellow, Department of Paediatrics, School of Clinical Sciences at Monash Health, Medicine, Nursing and Health Sciences, Monash University, Vic

Publisher

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2020

41.

Plasma level of laminin 5 and collagen IV in cryptorchidism.

Komarowska M., Szymanska B., Oldak, Sankiewicz A., Matuszczak E., Gorodkiewicz E., Debek W., Milewski R., Hermanowicz A.

Embase

Advances in Medical Sciences. 65 (1) (pp 176-181), 2020. Date of Publication: March 2020.

[Article]

AN: 2004671343

Purpose: Laminin 5 and collagen IV are the main compounds of the extracellular matrix of the germinal epithelium. The purpose of this study was to evaluate the concentration of these two markers of fibrosis in the plasma of boys with congenital unilateral cryptorchidism.

Material(s) and Method(s): The study group comprised 43 boys aged 1-3 years with congenital unilateral cryptorchidism. The control group included 54 healthy, age matched boys, admitted for planned hernioplasty. To assess laminin 5 and collagen IV in the plasma of boys with unilateral

cryptorchidism, we used a new biosensor with Surface Plasmon Resonance Imaging technique detection.

Result(s): The median concentration of laminin 5 and collagen IV in the serum of boys with congenital, unilateral cryptorchidism was higher than in boys with normal scrotal testis. The difference was statistically significant ($p < 0.0001$). We did not notice a correlation between a higher position of the testicles in the inguinal and/or their condition and levels of laminin 5 and collagen IV in the plasma.

Conclusion(s): Laminin 5 and collagen IV concentrations in the plasma were higher in patients with congenital unilateral cryptorchidism. We believe that in the future, our results could be compared with fertility level in adulthood.

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Embase

Institution

(Komarowska, Matuszczak, Debek, Hermanowicz) Department of Pediatric Surgery and Urology, Medical University of Bialystok, Bialystok, Poland (Szymanska, Oldak, Sankiewicz, Gorodkiewicz) Electrochemistry Department, University of Bialystok, Bialystok, Poland (Milewski) Department of Statistics and Medical Informatics, Medical University of Bialystok, Bialystok, Poland

Publisher

Medical University of Bialystok (Kilinskiego 1, Bialystok 15-089, Poland)

Year of Publication

2020

42.

Prognosis of testicular torsion orchiopexy.

He M., Li M., Zhang W.

Embase

Andrologia. 52 (1) (no pagination), 2020. Article Number: e13477. Date of Publication: 01 Feb 2020.

[Article]

AN: 2003607493

The purpose of this study was to follow up patients who underwent testicular torsion orchiopexies in order to observe whether testicular atrophy had occurred and to identify any influencing factors regarding atrophy. Patient data collected in this study included age, symptom duration, pre-operative preparation time, cryptorchidism testicular torsion, spermatic cord torsion degree, ultrasound findings at least 6 months after orchiopexy, testicular atrophy, mean platelet volume (MPV), address and medical insurance. Twenty-nine patients with a mean age of 147 (126.5-163) months involved in our study. The duration of follow-up ranged from 6 to 33 months with a median follow-up duration of 12 (8.5-21) months. Only MPV was significantly different between the atrophy group and nonatrophic group ($p = .022$) and the receiver operating characteristic (ROC) curve revealed that the cut-off value for MPV atrophy was 9.9 fl, with a sensitivity of 81.8% and a specificity of 70.6%. In conclusion, we found that 41.4% patients eventually experienced testicular atrophy after performing the testicular salvage procedure. MPV might be used as an indicator of testicular atrophy after an operation; however, the accuracy of MPV needs to be confirmed using significant follow-up prospective studies.

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Status

Embase

Author NameID

He, Meng; ORCID: <http://orcid.org/0000-0001-6925-2925>

Institution

(He, Li, Zhang) Urology Department, Beijing Children's Hospital, Capital Medical University, Beijing, China

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Urologic Conditions in Infants and Children: Inguinal Hernia, Hydrocele, and Cryptorchidism.
Simpson M., Sundaram V.

Embase

FP essentials. 488 (pp 16-20), 2020. Date of Publication: 01 Jan 2020.

[Article]

AN: 630491717

In children, inguinal hernias, hydroceles, and cryptorchidism typically are associated with a patent processus vaginalis. Inguinal hernias occur in 3.5%-5% of full-term newborns and 9%-11% of premature newborns. Inguinal hernias are characterized by an intermittent mass in the groin that may be reducible or incarcerated. Incarcerated hernias usually are painful, can cause vomiting, and require prompt intervention. The definitive treatment is surgery, and urgency depends on symptoms and ability to reduce the hernia. Hydrocele is an accumulation of serous fluid in the tunica vaginalis around the testicle that presents as a painless, fluctuant mass. Most hydroceles resolve spontaneously by age 1 year. Cryptorchidism occurs when one or both testes do not migrate to the scrotum. The diagnosis is made via history and physical examination.

Spontaneous descent of the testis may occur before age 6 months but referral to a surgical subspecialist is indicated if descent does not occur.

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Institution

(Simpson, Sundaram) LewisGale Medical Center Family Medicine Residency Program, 1900
Electric Rd Salem

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Year of Publication

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44.

Ambiguous Genitalia And Disorders of Sexual Differentiation. [Review]

Mehmood KT; Rentea RM.

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StatPearls Publishing. 2020 01.

[Review]

UI: 32491367

The birth of an infant with ambiguous genitalia generates difficult multiple medical, surgical, ethical, psychosocial, and physical issues for patients and their parents. Phenotypic sex results from the differentiation of internal ducts and external genitalia under the influence of hormones and other additional factors. When discordance occurs among three processes (chromosomal, gonadal, phenotypic sex determination), a DSD is the result. Terminology such as hermaphrodite, pseudo-hermaphrodite, and intersex, are considered to be pejorative and dated. These terms have been replaced by the term disorders of sexual development (DSD) by the consensus statement on management of intersex disorders.[1][2] Disorders of sexual development are defined as congenital conditions characterized by atypical development of chromosomal, gonadal, or anatomic sex.[3] Normal sexual development in utero is dependent upon a precise and coordinated spatiotemporal sequence of various activating and repressing factors.[4] Any deviations from the usual pattern of differentiation can present as DSDs. Two distinct processes occur in normal sexual development. The first of which is sex determination in which the bi-potential gonads are induced to form either the male testes or the female ovaries. Secondly, the newly formed gonads secrete hormones to modulate the formation of internal and external genitalia.[5] The phenotypic manifestation of DSDs are diverse and can include; bilateral undescended testes, severe hypospadias (scrotal or perineal), clitoromegaly, a fusion of posterior labial folds, female external genitalia with palpable gonad, discordant genitalia and sex chromosomes. The inclusion of disorders in which there is no genital/gonadal discordance like Turner syndrome, Klinefelter syndrome, simple hypospadias remains controversial. Regardless of presentation or severity, individuals require a multidisciplinary approach that is warranted to improve the quality of life and achieve the best possible outcomes.

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Book Title

StatPearls

Version ID

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Authors Full Name

Mehmood, Khawar T.; Rentea, Rebecca M..

Institution

Mehmood, Khawar T.. Hameed Latif Hospital Rentea, Rebecca M.. Children's Mercy

Publisher

StatPearls Publishing

Year of Publication

2020

45.

Comparison of diagnostic and treatment guidelines for undescended testis.

Shin J; Jeon GW.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Clinical and Experimental Pediatrics. 2020 Mar 23.

[Journal Article]

UI: 32252147

Cryptorchidism or undescended testis is the single most common genitourinary disease in male neonates. In most cases, the testes will descend spontaneously by three months of age. If the testes do not descend by six months of age, the probability of spontaneous descent thereafter is low. About 1-2% of boys older than six months have undescended testes after their early postnatal descent. In some cases, a testis vanishes into the abdomen or re-ascends after birth after being present in the scrotum at birth. An inguinal undescended testis is sometimes mistaken for an inguinal hernia. A surgical specialist referral is recommended if descent does not occur by six months, undescended testis is newly diagnosed after six months of age, or testicular torsion is suspected. International guidelines do not recommend ultrasonography or other diagnostic imaging because they cannot add diagnostic accuracy or change treatment. Routine hormonal therapy is not recommended for undescended testis due to a lack of evidence. Orchiopexy is recommended between 6 and 18 months at the latest to protect the fertility potential and decrease the risk of malignant changes. Patients with unilateral undescended testis have an infertility rate of up to 10%. This rate is even higher in patients with bilateral undescended testes, with intra-abdominal undescended testis, or who underwent delayed orchiopexy. Patients with undescended testis have a threefold increased risk of testicular cancer later in life compared to the general population. Self-examination after puberty is recommended to facilitate early cancer detection. A timely referral to a surgical specialist and timely surgical correction are the most important factors for decreasing infertility and testicular cancer rates.

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1

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Publisher

Authors Full Name

Shin, Jaeho; Jeon, Ga Won.

Institution

Shin, Jaeho. Division of Pediatric Surgery, Department of Surgery, Inje University Busan Paik Hospital, Busan, Korea. Jeon, Ga Won. Department of Pediatrics, Inje University Busan Paik Hospital, Busan, Korea.

Jeon, Ga Won. Inje University College of Medicine, Busan, Korea.

Year of Publication

2020

46.

Testicular function and sexuality in adult patients with anorectal malformation.

Trovalusci E; Rossato M; Gamba P; Midrio P.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Pediatric Surgery. 2020 Jan 29.

[Journal Article]

UI: 32057441

PURPOSE: To collect data on sexual and fertility issues in adult male patients with history of anorectal malformations (ARM).

MATERIALS AND METHODS: Thirty adult males born with ARM, cared for at the Pediatric Surgery of Treviso and Padua Hospitals, were enrolled and interviewed about sexual habits and relationships. Testicular ultrasound, evaluation of male sex hormones and semen analysis were performed to assess testicular function and compare data with 15 fertile controls. Presence of erectile dysfunction was evaluated with IIEF-5 questionnaire.

RESULTS: Cryptorchidism and recurrent orchiepididymitis were reported in 33% and 40% of patients, respectively. Average testicular volume resulted significantly lower than fertile controls (11.1 vs 14.3mL, $p=0.002$) and 53.5% presented testicular hypotrophy (<10mL). Erectile dysfunction was reported by a single patient and ejaculatory anomalies by 46.5%. Thirteen

patients were azoospermic/cryptozoospermic; 6 of them presented a reduced peripheral sensitivity to androgens (ASI>139). Coital debut resulted delayed at 18years old (vs 15years in the control group). Overall 63.5% reported their condition did not affect their sexual sphere.
CONCLUSIONS: Evaluation of testicular function is recommended in ARM patients to detect and treat possible infertility disorders, to recognize the clinical conditions which could affect the spermatogenesis since childhood, and to guarantee psychological support.
LEVEL OF EVIDENCE RATING: Prognosis study. Level III (case-control study).
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Authors Full Name

Trovalusci, Emanuele; Rossato, Marco; Gamba, Piergiorgio; Midrio, Paola.

Institution

Trovalusci, Emanuele. Pediatric Surgery, Ca' Foncello Hospital, Treviso, Italy; Pediatric Surgery, University of Padova, Padova, Italy. Rossato, Marco. Department of Medicine-DIMED, Clinica Medica 3degree, University of Padova, Padova, Italy.

Gamba, Piergiorgio. Pediatric Surgery, University of Padova, Padova, Italy.

Midrio, Paola. Pediatric Surgery, Ca' Foncello Hospital, Treviso, Italy. Electronic address: paola.midrio@aulss2.veneto.it.

Year of Publication

2020

47.

A novel method of bilateral patent processus vaginalis ligation in transumbilical single-site multiport laparoscopic orchiopexy.

Wang X; Guan Y; Wu Y; Meng Q; Dong M.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

International Journal of Medical Sciences. 17(8):1043-1047, 2020.

[Journal Article]

UI: 32410833

Objective: To evaluate safety and efficacy of a novel method of bilateral patent processus vaginalis ligation in transumbilical single-site multiport laparoscopic orchiopexy for children.

Methods: A retrospective study was carried out comparing the novel ligation and conventional ligation performed by a single surgeon between July, 2017-July, 2018. The patients were divided into the novel group (42 cases) and the conventional group (59 cases). In the novel group, transumbilical single-site multiport laparoscopic orchiopexy was performed and the bilateral internal rings was stitched with "8" pattern suture. In the conventional group, the conventional TriPort laparoscopic orchiopexy was performed and purse string suture was used to fix the internal rings. The parameters of operative duration time, postoperative hospital stay; postoperative complications were compared between 2 groups.

Results: All operations were successful. No Perioperative period complications were found and all patients were discharged within 4-6 days after operation. There is no statistic difference in the surgery time and hospitalization day. However, there is significant difference in the Pain face scale scores after day 2(1.60+/-0.73 VS 2.02+/-0.86). And there is no scar and the satisfactory cosmetic could be seen in scrotum and inguinal area in the novel group. **Conclusion:** The novel ligation was safety and efficacy. It is relatively easy to perform with smaller scar and less pain.

We propose the novel ligation as a more viable treatment option for pediatric cryptorchidism with bilateral patent processus vaginalis.

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1

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Authors Full Name

Wang, Xin; Guan, Yong; Wu, Yong; Meng, QingYa; Dong, Ming.

Institution

Wang, Xin. Department of Pediatric Surgery, Tianjin Children's Hospital, No.238 LongYan Road, Tianjin 300134, PR China. Guan, Yong. Department of Pediatric Surgery, Tianjin Children's Hospital, No.238 LongYan Road, Tianjin 300134, PR China.

Wu, Yong. Department of Pediatric Surgery, Tianjin Children's Hospital, No.238 LongYan Road, Tianjin 300134, PR China.

Meng, QingYa. Department of Pediatric Surgery, Tianjin Children's Hospital, No.238 LongYan Road, Tianjin 300134, PR China.

Dong, Ming. Department of lung cancer surgery, Tianjin medical university general hospital, No.154 Anshan Road, Tianjin 300052, PR China.

PMC Identifier

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48.

Torsion of an undescended testis - A surgical pediatric emergency.

Kargl S; Haid B.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Pediatric Surgery. 55(4):660-664, 2020 Apr.

[Journal Article]

UI: 31272681

BACKGROUND/PURPOSE: Torsion of an undescended testis is a surgical emergency whose frequency may be underestimated in the pediatric population. We describe this entity and focus on diagnostic challenges and optimal treatment of torsion of an undescended testis.

METHODS: We present a two-center retrospective chart review of patients with torsion of an undescended testis treated between 2013 and 2018. Two instructive cases are used to depict characteristics of this rare entity.

RESULTS: We identified 11 patients with previously diagnosed cryptorchidism undergoing surgery for torsion of an undescended testis, accounting for 9.7% (11/107) of all testicular torsions in the period. Mean age at diagnosis was 9.4 months (1-22 months). Mean duration from onset of symptoms to presentation was 19.3 h (8-48 h). At admission to hospital 10 patients presented with groin lump (10/11, 90.9%) with or without pain leading to a suspected diagnosis of inguinal testicular torsion (5/11, 45.5%), incarcerated inguinal hernia (4/11, 36.4%) and epididymitis (1/11, 9.1%). Ten patients had an ultrasound examination before surgery leading to the correct diagnosis in six patients. Ultrasound findings were misinterpreted as incarcerated inguinal hernia in three patients. In eight patients the testis had to be removed at time of surgery; one of the three salvaged testes atrophied, resulting in a salvage rate of 18%.

CONCLUSION: Torsion of an inguinal testis is not as rare as it might be presumed. Presentation of these patients is often deferred owing to equivocal signs and symptoms. In addition age at presentation differs from typical testicular torsion. As this might negatively influence testicular salvage rate, we advocate for special attention to this differential diagnosis in children with groin

pathologies. Even if the child is not in pain, a tender groin in boys with undescended testes must prompt a quick and thorough examination to rule out torsion of an undescended testis. Ultrasound examination is of limited value and must not delay acute surgical treatment.

LEVEL OF EVIDENCE: IV.

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Authors Full Name

Kargl, Simon; Haid, Bernhard.

Institution

Kargl, Simon. Department of Pediatric Surgery, MedCampus IV, Kepler University Hospital Linz.

Electronic address: kargl.simon@gmail.com. Haid, Bernhard. Department of Pediatric Urology, Ordensklinikum Linz, Hospital of the Sisters of Charity.

Year of Publication

2020

49.

Phenotype and variations associated with the deletion of the 1q44 cytoband and the pathogenic duplication in the 9q32q34.3 cytobands.

Gomez-Carpintero Garcia A; Vidal Esteban A; Bermejo Gomez A; Pua Torrejon RC.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

BMJ Case Reports. 13(3), 2020 Mar 08.

[Journal Article]

UI: 32152069

The advance in the human genetic field has permitted to identify small chromosome alterations and associate them to a specific phenotype. However, there are many mutations that have not yet been described in the literature. We describe the clinical case of a term newborn with appropriate weight to its gestational age, without perinatal background of interest that, at birth, presented: macrocephaly, hypertelorism, low-set ears, prominent forehead, micrognathia,

camptodactyly, bilateral cryptorchidism, inspiratory stridor with the cry, multifocal systolic murmur, wide anterior fontanel and hypotonia of mixed characteristics and in whom a deletion of the 1q44 cytoband and a pathogenic duplication in the 9q32q34.3 cytoband were detected. We perform a review of the literature.

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Authors Full Name

Gomez-Carpintero Garcia, Ana; Vidal Esteban, Ana; Bermejo Gomez, Amanda; Pua Torrejon, Ruth Camila.

Institution

Gomez-Carpintero Garcia, Ana. Pediatria, Hospital Universitario de Mostoles, Mostoles, Madrid, Spain gmezcarpintero@gmail.com. Vidal Esteban, Ana. Pediatria, Hospital Universitario de Mostoles, Mostoles, Madrid, Spain.

Bermejo Gomez, Amanda. Pediatria, Hospital Universitario de Mostoles, Mostoles, Madrid, Spain.

Pua Torrejon, Ruth Camila. Pediatria, Hospital Universitario de Mostoles, Mostoles, Madrid, Spain.

Year of Publication

2020

50.

Five new cases of syndromic intellectual disability due to KAT6A mutations: widening the molecular and clinical spectrum.

Urreizti R; Lopez-Martin E; Martinez-Monseny A; Pujadas M; Castilla-Vallmanya L; Perez-Jurado LA; Serrano M; Natera-de Benito D; Martinez-Delgado B; Posada-de-la-Paz M; Alonso J; Marin-Reina P; O'Callaghan M; Grinberg D; Bermejo-Sanchez E; Balcells S.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Orphanet Journal Of Rare Diseases. 15(1):44, 2020 Feb 10.

[Journal Article]

UI: 32041641

BACKGROUND: Pathogenic variants of the lysine acetyltransferase 6A or KAT6A gene are associated with a newly identified neurodevelopmental disorder characterized mainly by intellectual disability of variable severity and speech delay, hypotonia, and heart and eye malformations. Although loss of function (LoF) mutations were initially reported as causing this disorder, missense mutations, to date always involving serine residues, have recently been associated with a form of the disorder without cardiac involvement.

RESULTS: In this study we present five new patients, four with truncating mutations and one with a missense change and the only one not presenting with cardiac anomalies. The missense change [p.(Gly359Ser)], also predicted to affect splicing by in silico tools, was functionally tested in the patient's lymphocyte RNA revealing a splicing effect for this allele that would lead to a frameshift and premature truncation.

CONCLUSIONS: An extensive revision of the clinical features of these five patients revealed high concordance with the 80 cases previously reported, including developmental delay with speech delay, feeding difficulties, hypotonia, a high bulbous nose, and recurrent infections. Other features present in some of these five patients, such as cryptorchidism in males, syndactyly, and trigonocephaly, expand the clinical spectrum of this syndrome.

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1

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Author NameID

Urreizti, Roser; ORCID: <http://orcid.org/0000-0003-3617-7134>

Authors Full Name

Urreizti, Roser; Lopez-Martin, Estrella; Martinez-Monseny, Antonio; Pujadas, Montse; Castilla-Vallmanya, Laura; Perez-Jurado, Luis Alberto; Serrano, Mercedes; Natera-de Benito, Daniel; Martinez-Delgado, Beatriz; Posada-de-la-Paz, Manuel; Alonso, Javier; Marin-Reina, Purificacion; O'Callaghan, Mar; Grinberg, Daniel; Bermejo-Sanchez, Eva; Balcells, Susanna.

Institution

Urreizti, Roser. Department of Genetics, Microbiology and Statistics, Faculty of Biology, University of Barcelona, IBUB, IRSJD, Barcelona, Spain. roseruf@yahoo.es. Urreizti, Roser. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain. roseruf@yahoo.es.

Urreizti, Roser. Present address: Neurometabolic Unit, Hospital Sant Joan de Deu, Barcelona, Spain. roseruf@yahoo.es.

Lopez-Martin, Estrella. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Lopez-Martin, Estrella. Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Martinez-Monseny, Antonio. Department of Genetic and Molecular Medicine and Pediatric Rare Diseases Institute (IPER), Institut de Recerca Sant Joan de Deu (IRSJD), Hospital Sant Joan de Deu, Barcelona, Spain.

Pujadas, Montse. Genetics Unit, University Pompeu Fabra, Hospital del Mar Research Institute IMIM, Barcelona, Spain.

Castilla-Vallmanya, Laura. Department of Genetics, Microbiology and Statistics, Faculty of Biology, University of Barcelona, IBUB, IRSJD, Barcelona, Spain.

Castilla-Vallmanya, Laura. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Perez-Jurado, Luis Alberto. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Perez-Jurado, Luis Alberto. Genetics Unit, University Pompeu Fabra, Hospital del Mar Research Institute IMIM, Barcelona, Spain.

Perez-Jurado, Luis Alberto. Women's and Children's Hospital, South Australian Health and Medical Research Institute and The University of Adelaide, Adelaide, Australia.

Serrano, Mercedes. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Serrano, Mercedes. Department of Neurology, Hospital Sant Joan de Deu, Barcelona, Spain.

Natera-de Benito, Daniel. Department of Neurology, Hospital Sant Joan de Deu, Barcelona, Spain.

Martinez-Delgado, Beatriz. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Martinez-Delgado, Beatriz. Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Posada-de-la-Paz, Manuel. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Posada-de-la-Paz, Manuel. Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Alonso, Javier. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Alonso, Javier. Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Marin-Reina, Purificacion. Dysmorphology and Clinical Genetics, Division of Neonatology, Neonatal Research Unit, Hospital Universitario y Politecnico La Fe, Valencia, Spain.

O'Callaghan, Mar. Department of Neurology, Hospital Sant Joan de Deu, Barcelona, Spain.

Grinberg, Daniel. Department of Genetics, Microbiology and Statistics, Faculty of Biology, University of Barcelona, IBUB, IRSJD, Barcelona, Spain.

Grinberg, Daniel. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Bermejo-Sanchez, Eva. Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

Balcells, Susanna. Department of Genetics, Microbiology and Statistics, Faculty of Biology, University of Barcelona, IBUB, IRSJD, Barcelona, Spain.

Balcells, Susanna. Centro de Investigaciones Biomedicas en Red de Enfermedades Raras (CIBERER), Instituto de Salud Carlos III (ISCIII), Madrid, Spain.

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51.

Modified stepwise mini-incision microdissection testicular sperm extraction: a useful technique for patients with a history of orchidopexy affected by non-obstructive azoospermia.

Li P; Yao CC; Zhi EL; Xu Y; Wan Z; Jiang YC; Huang YH; Gong YH; Chen HX; Tian RH; Yang C; Zhao LY; Li Z.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Zhejiang University SCIENCE B. 21(1):87-92, 2020 Jan..

[Journal Article]

UI: 31898445

Non-obstructive azoospermia (NOA), which is defined as the absence of spermatozoa in the ejaculate secondary to impaired spermatogenesis within the testis, may be caused by a variety of etiologies, including varicocele-induced testicular damage, cryptorchidism, prior testicular torsion, post-pubertal mumps orchitis, gonadotoxic effects from medications, genetic abnormalities,

chemotherapy/radiation, and other unknown causes currently classified as idiopathic (Cocuzza et al., 2013). The microdissection testicular sperm extraction (micro-TESE) technique involves a meticulous microsurgical exploration of the testicular parenchyma to identify and selectively extract larger seminiferous tubules that carry a higher probability of complete spermatogenesis (Schlegel, 1999). The Cornell group evaluated the efficacy of micro-TESE in 152 NOA patients with an associated history of cryptorchidism. In their series, spermatozoa were successfully retrieved in 116/181 attempts (64%), and the resulting pregnancy rate was 50% with a delivery rate of 38% (Dabaja and Schlegel, 2013). Franco et al. (2016) described a stepwise micro-TESE approach in NOA patients, which was considered to reduce the cost, time, and effort associated with the surgery. Alrabeeah et al. (2016) further reported that a mini-incision micro-TESE, carried through a 1-cm equatorial testicular incision, can be useful for micro-TESE candidates, particularly in patients with cryptozoospermia. We conducted a retrospective study of 20 consecutive NOA patients with a history of orchidopexy from May 2015 to March 2017.

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1

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Authors Full Name

Li, Peng; Yao, Chen-Cheng; Zhi, Er-Lei; Xu, Yuan; Wan, Zhong; Jiang, Ying-Chuan; Huang, Yu-Hua; Gong, Yue-Hua; Chen, Hui-Xing; Tian, Ru-Hui; Yang, Chao; Zhao, Liang-Yu; Li, Zheng.

Institution

Li, Peng. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China. Li, Peng.

Department of Urology, Shanghai General Hospital of Nanjing Medical University, Shanghai 200080, China.

Yao, Chen-Cheng. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.

Zhi, Er-Lei. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.

Xu, Yuan. Reproductive Medicine Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.

Wan, Zhong. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.

Jiang, Ying-Chuan. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.

Jiang, Ying-Chuan. Department of Urology, Zhoushan Hospital, Zhoushan 316021, China.

Huang, Yu-Hua. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Gong, Yue-Hua. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Chen, Hui-Xing. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Tian, Ru-Hui. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Yang, Chao. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Zhao, Liang-Yu. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Li, Zheng. Department of Andrology, Center for Men's Health, Urologic Medical Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.
Li, Zheng. Reproductive Medicine Center, Shanghai General Hospital, Shanghai Jiao Tong University, Shanghai 200080, China.

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Year of Publication

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52.

Molecular and clinical studies in 107 Noonan syndrome affected individuals with PTPN11 mutations.

Athota JP; Bhat M; Nampoothiri S; Gowrishankar K; Narayanachar SG; Puttamalles V; Farooque MO; Shetty S.

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BMC Medical Genetics. 21(1):50, 2020 03 12.

[Journal Article. Research Support, Non-U.S. Gov't]

UI: 32164556

BACKGROUND: Noonan syndrome (NS), an autosomal dominant developmental genetic disorder, is caused by germline mutations in genes associated with the RAS / mitogen-activated protein kinase (MAPK) pathway. In several studies PTPN11 is one of the genes with a significant number of pathogenic variants in NS-affected patients. Therefore, clinically diagnosed NS individuals are initially tested for pathogenic variants in PTPN11 gene to confirm the relationship before studying genotype-phenotype correlation.

METHODS: Individuals (363) with clinically diagnosed NS from four hospitals in South India were recruited and the exons of PTPN11 gene were sequenced.

RESULTS: Thirty-two previously described pathogenic variants in eight different exons in PTPN11 gene were detected in 107 patients, of whom 10 were familial cases. Exons 3, 8 and 13 had the highest number of pathogenic variants. The most commonly identified pathogenic variants in this series were in exon 8 (c.922A > G, c.923A > G), observed in 22 of the affected. Congenital cardiac anomalies were present in 84% of the mutation-positive cohort, the majority being defects in the right side of the heart. The most common facial features were downward-slanting palpebral fissures, hypertelorism and low-set posteriorly rotated ears. Other clinical features included short stature (40%), pectus excavatum (54%) and, in males, unilateral or bilateral cryptorchidism (44%).

CONCLUSION: The clinical features and mutational spectrum observed in our cohort are similar to those reported in other large studies done worldwide. This is the largest case series of NS-affected individuals with PTPN11 mutations described till date from India.

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1

Status

MEDLINE

Author NameID

Shetty, Swathi; ORCID: <https://orcid.org/0000-0002-2290-6204>

Authors Full Name

Athota, Jeevana Praharsa; Bhat, Meenakshi; Nampoothiri, Sheela; Gowrishankar, Kalpana; Narayanachar, Sanjeeva Ghanti; Puttamalles, Vinuth; Farooque, Mohammed Oomer; Shetty, Swathi.

Institution

Athota, Jeevana Praharsa. Molecular Genetics, Centre for Human Genetics, Bengaluru, 560100, India. Bhat, Meenakshi. Molecular Genetics, Centre for Human Genetics, Bengaluru, 560100, India.

Bhat, Meenakshi. Pediatric Genetics, Indira Gandhi Institute of Child Health, Bengaluru, 560029, India.

Nampoothiri, Sheela. Pediatric Genetics, Amrita Institute of Medical Sciences & Research Centre (AIMS), Kochi, 682041, India.

Gowrishankar, Kalpana. Medical Genetics, Kanchi Kamakoti CHILDS Trust Hospital, Chennai, 600034, India.

Narayanachar, Sanjeeva Ghanti. Pediatric Genetics, Indira Gandhi Institute of Child Health, Bengaluru, 560029, India.

Puttamallesh, Vinuth. Molecular Genetics, Centre for Human Genetics, Bengaluru, 560100, India.

Farooque, Mohammed Oomer. Molecular Genetics, Centre for Human Genetics, Bengaluru, 560100, India.

Shetty, Swathi. Molecular Genetics, Centre for Human Genetics, Bengaluru, 560100, India.
swathi@chg.res.in.

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Year of Publication

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53.

Fertility Potential is Compromised in 20% to 25% of Boys with Nonsyndromic Cryptorchidism despite Orchiopexy within the First Year of Life.

Hildorf S; Clasen-Linde E; Cortes D; Fossum M; Thorup J.

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Journal of Urology. 203(4):832-840, 2020 04.

[Journal Article]

UI: 31642739

PURPOSE: One of the concerns surrounding cryptorchidism is the risk of impaired fertility. Current guidelines recommend orchiopexy at age 6 to 12 months to optimize fertility outcome. We evaluated the fertility potential of boys with nonsyndromic cryptorchidism who underwent orchiopexy within the recommended age range to clarify the need for eventual supplemental treatment modalities.

MATERIALS AND METHODS: We retrospectively evaluated mini-puberty hormones (follicle-stimulating hormone, luteinizing hormone and inhibin B) and testicular biopsies from boys with

cryptorchidism who underwent orchiopexy within the first year of life between 2010 and 2019. We histologically analyzed germ cell number and type A dark spermatogonia number per seminiferous tubule cross-section in relation to normal values.

RESULTS: Of the 333 boys with nonsyndromic cryptorchidism 83 (25%, 21% with bilateral cryptorchidism) had a reduced number of germ cells. A total of 70 boys (21%) had low serum inhibin B, of whom 32 (46%) had a decreased number of germ cells and 23 (33%) had a decreased number of type A dark spermatogonia ($p < 0.01$). Overall, 75 boys (23%) had no type A dark spermatogonia present.

CONCLUSIONS: Despite early and successful orchiopexy, 20% to 25% of boys with cryptorchidism may be at risk for infertility based on hormonal and histological data. Blood test and testicular biopsy are mandatory to identify boys at high risk for infertility, in whom additional treatment modalities and followup may be needed.

Version ID

1

Status

MEDLINE

Authors Full Name

Hildorf, Simone; Clasen-Linde, Erik; Cortes, Dina; Fossum, Magdalena; Thorup, Jorgen.

Institution

Hildorf, Simone. Department of Pediatric Surgery, Surgical Clinic C, Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark. Clasen-Linde, Erik. Department of Pathology, Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark.

Cortes, Dina. Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark.

Cortes, Dina. Section of Endocrinology, Department of Pediatrics, Copenhagen University Hospital Hvidovre, Hvidovre, Denmark.

Fossum, Magdalena. Department of Pediatric Surgery, Surgical Clinic C, Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark.

Fossum, Magdalena. Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark.

Thorup, Jorgen. Department of Pediatric Surgery, Surgical Clinic C, Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark.

Thorup, Jorgen. Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark.

Comments

Comment in (CIN) Comment in (CIN)

Year of Publication

54.

Early Experience with Laparoscopic Surgical Practice in Ondo State, South Western Nigeria.

Obonna GC; Etonyeaku AC; Katung IA; Bamigbola KT; Okereke CE.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

West African Journal of Medicine. 37(2):183-188, 2020 Apr-Jun.

[Journal Article]

UI: 32150638

BACKGROUND: Laparoscopic surgery is a relatively new and expanding field of surgical therapy in Ondo state. This is a multi-centre study cataloguing the work of the authors in Ondo State, Nigeria.

AIM: To determine the indications, operative findings, and interventions at Laparoscopy in our resource challenged settings.

PATIENTS AND METHODS: Medical records of all patients who had laparoscopic procedure at the Federal Medical Centre, Owo, Ondo State Specialist hospital, Okitipupa, University of Medical Sciences Teaching Hospital, Ondo, George and Martins Medical Centre, Ore and Mishmael Hospitals and Clinics, Akure from December, 2009 to December, 2018 were reviewed. Data on patient's age, gender, indications for surgery, duration of hospital stay, outcome of surgery were analyzed. Challenges and adaptations were also noted.

RESULTS: One hundred and eighty-one (181) laparoscopic procedures were done, but only 152 had complete records for review. The median age was 35.5 years (mean = 33.7+/-11.4years; age range of 8 month -72 years). There were more males 88(57.9%) than females, 64(42.1%).

Laparoscopy was purely diagnostic (n=28, 18.4%), therapeutic (n=118, 77.6%) or both (n=6, 3.9%). Cholecystectomy (n=76, 50%) and appendicectomy (n=37, 24.3%) were the two most common procedures done. In the paediatric patients, patent processus vaginalis (hernia), cryptorchidism and indeterminate sex (gender) were common indication. Challenges encountered were power failure (n=3, 2%), equipment failure (n=4, 2.6%) and difficult dissection (n=4, 2.6%).

The mean duration of surgery was 96.96(+/-25) minutes (diagnostic), 150 (+/-57.6) minutes (therapeutic); while the mean duration of hospital stay was one day (diagnostic) and 2.3(+/-1.7) days for therapeutic interventions.

CONCLUSION: Laparoscopic service is achievable with adequate motivation, males appear to benefit more in our setting, and the service transcends all aged groups.

Version ID

1

Status

MEDLINE

Authors Full Name

Obonna, G C; Etonyeaku, A C; Katung, I A; Bamigbola, K T; Okereke, C E.

Institution

Obonna, G C. Department of Surgery, University of Medical Sciences, Medical village Ondo City, Ondo State, Southwestern Nigeria. Etonyeaku, A C. Department of Surgery Obafemi Awolowo University/ Obafemi Awolowo University Teaching Hospitals Complex Ile-Ife Osun State, Southwestern, Nigeria.

Katung, I A. Department of Surgery, Federal Medical Centre Owo, Ondo State, Southwestern Nigeria.

Bamigbola, K T. Department of Surgery, Federal Medical Centre Owo, Ondo State, Southwestern Nigeria.

Okereke, C E. Department of Surgery, Federal Medical Centre Owo, Ondo State, Southwestern Nigeria.

Year of Publication

2020

55.

A novel nonsense mutation in the STS gene in a Pakistani family with X-linked recessive ichthyosis: including a very rare case of two homozygous female patients.

Afzal S; Ramzan K; Ullah S; Wakil SM; Jamal A; Basit S; Waqar AB.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

BMC Medical Genetics. 21(1):20, 2020 01 31.

[Journal Article]

UI: 32005174

BACKGROUND: X-linked ichthyosis (XLI; OMIM# 308100) is a recessive keratinization disorder characterized by the presence of dark brown, polygonal, adherent scales on different parts of the body surface. It almost exclusively affects males and the estimated prevalence ranges from 1:2000-6000 in males worldwide. Extracutaneous manifestations are frequent including corneal opacities, cryptorchidism, neuropsychiatric symptoms or others. Up to 90% of XLI cases are caused by recurrent hemizygous microdeletion encompassing entire STS gene on chromosome Xp22.3, while only a minority of patients shows partial deletions or loss of function point mutations in STS. Larger deletions also involving contiguous genes are identified in syndromic patients.

METHODS: Here, we report clinical and genetic findings of a large Pakistani family having 16 affected individuals including 2 females with XLI. Molecular karyotyping and direct DNA sequencing of coding region of the STS gene was performed.

RESULTS: The clinical manifestations in affected individuals involved generalized dryness and scaling of the skin with polygonal, dark scales of the skin on scalp, trunk, limbs, and neck while sparing face, palms and soles. There were no associated extra-cutaneous features such as short stature, cryptorchidism, photophobia, corneal opacities, male baldness, and behavioral, cognitive, or neurological phenotypes including intellectual disability, autism or attention deficit hyperactivity disorder. Molecular karyotyping was normal and no copy number variation was found. Sanger sequencing identified a novel hemizygous nonsense mutation (c.287G > A; p.W96*), in exon 4 of STS gene in all affected male individuals. In addition, two XLI affected females in the family were found to be homozygous for the identified variant.

CONCLUSIONS: This study is useful for understanding the genetic basis of XLI in the patients studied, for extending the known mutational spectrum of STS, diagnosis of female carriers and for further application of mutation screening in the genetic counseling of this family.

Version ID

1

Status

MEDLINE

Author NameID

Waqar, Ahmed Bilal; ORCID: <https://orcid.org/0000-0001-8255-3634>

Authors Full Name

Afzal, Sibtain; Ramzan, Khushnooda; Ullah, Sajjad; Wakil, Salma M; Jamal, Arshad; Basit, Sulman; Waqar, Ahmed Bilal.

Institution

Afzal, Sibtain. Faculty of Allied and Health Sciences, Imperial College of Business Studies, Lahore, Pakistan. Ramzan, Khushnooda. Department of Genetics, King Faisal Specialist Hospital and Research Centre, PO Box 3354, Riyadh, 11211, Saudi Arabia.

Ullah, Sajjad. Faculty of Allied and Health Sciences, Imperial College of Business Studies, Lahore, Pakistan.

Wakil, Salma M. Department of Genetics, King Faisal Specialist Hospital and Research Centre, PO Box 3354, Riyadh, 11211, Saudi Arabia.

Jamal, Arshad. Faculty of Allied and Health Sciences, Imperial College of Business Studies, Lahore, Pakistan.

Basit, Sulman. Center for Genetics and Inherited Diseases, Taibah University, Madinah Al-Munawarah, Medina, Saudi Arabia.

Waqar, Ahmed Bilal. Faculty of Allied and Health Sciences, Imperial College of Business Studies, Lahore, Pakistan. drabwaqar@yahoo.com.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6995215>

Year of Publication

2020

56.

Replacement of male mini-puberty.

Papadimitriou D.T., Chrysis D., Nyktari G., Zoupanos G., Liakou E., Papadimitriou A., Mastorakos G.

Embase

Journal of the Endocrine Society. 3 (7) (pp 1275-1282), 2019. Date of Publication: 2019.

[Article]

AN: 629116430

Context: Clinical management of congenital hypogonadotropic hypogonadism (CHH) remains a challenge in pediatric endocrinology.

Objective(s): To investigate whether daily subcutaneous injections of the recombinant human LH/FSH preparation could mimic the physiological male mini-puberty. Design and Setting: The REMAP (REplacement of MAle mini-Puberty) study with up to 10 years of follow-up. Patients and Intervention: Ten neonates or infants, all with bilateral cryptorchidism in intraabdominal/inguinal position and micropenis with the absence of neonatal male mini-puberty, received daily subcutaneous injections of Pergoveris (LH/FSH 75/150 IU) for 3 months.

Main Outcome Measure(s): Restoration of bilateral cryptorchidism/micropenis and the Leydig/Sertoli cells function.

Result(s): At the end of treatment, median LH and FSH, both undetectable before treatment, reached high normal levels of 4.45 IU/L and supranormal levels 83 IU/L, respectively; median inhibin-b and anti-Mullerian hormone levels increased from subnormal (27.8 and 1.54 ng/mL, respectively) to normal levels (365 and 150 ng/mL, respectively); median testosterone increased from just detectable (0.02 ng/mL) to normal levels (3.3 ng/mL). Stretched penile length increased from a median of 2 to 3.8 cm. During therapy, all testes descended to the scrotal position (by the end of the first month in three patients, the second month in four patients, and the third month in three patients), measuring 1.5 mL and appearing normal in ultrasonography. Three infants received additional treatment with testosterone enanthate. In two infants, one of two testes regressed in the low inguinal area; both infants were successfully treated surgically. After 1 to 10 years of follow-up, all testes are still in scrotal position and have slightly regressed in size.

Conclusion(s): The proposed regimen mimics neonatal male mini-puberty and successfully treats infants with micropenis and cryptorchidism in CHH.

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Status

Embase

Author NameID

Papadimitriou, Dimitrios T.; ORCID: <http://orcid.org/0000-0002-6083-3560>

Institution

(Papadimitriou) Department of Pediatric-Adolescent Endocrinology and Diabetes, Athens Medical Center, 58, av. Kifissias, Athens 15125, Greece (Chrysis) Department of Pediatrics, Division of Pediatric Endocrinology, Medical School, University of Patras, Rion 26504, Greece

(Nyktari) Neonatal Intensive Care Unit, Gaia Maternity, Athens Medical Center, Athens 15125, Greece

(Zoupanos) Pediatric Urology Clinic, Athens Medical Center, Athens 15125, Greece

(Liakou) Athens Medical Center, Athens 15125, Greece

(Papadimitriou) Pediatric Endocrinology Unit, Department of Pediatrics, Attikon University Hospital, Athens 12462, Greece

(Mastorakos) Endocrine Unit, Second Department of Obstetrics and Gynecology, Aretaieion Hospital, Medical School, University of Athens, Athens 11528, Greece

Publisher

Oxford University Press (E-mail: Chris.Payne@oup.com)

Year of Publication

2019

57.

Risk factors affecting post-pubertal high serum follicle-stimulating hormone in patients with hypospadias.

Moriya K., Nakamura M., Kon M., Nishimura Y., Kanno Y., Kitta T., Shinohara N.

Embase

World journal of urology. 37 (12) (pp 2795-2799), 2019. Date of Publication: 01 Dec 2019.

[Article]

AN: 629923193

PURPOSE: The factors affecting spermatogenesis in adulthood in patients with hypospadias (HS) are not clearly understood. In the present study, risk factors affecting post-pubertal high serum follicle-stimulating hormone (FSH) were evaluated in patients with HS. **MATERIALS AND METHODS:** Among those with a history of HS surgery, patients in whom endocrinological evaluation regarding pituitary-gonadal axis was performed at 15 years of age or older between March 2004 and April 2018 were enrolled in the present study. High serum FSH was defined as greater than 10 mIU/ml. The severity of HS was divided into mild and severe. Factors affecting the post-pubertal high serum FSH were estimated.

RESULT(S): Seventy-nine patients were included in the present study. The severity of HS was mild in 35 and severe in 44. History of undescended testis (UDT) was confirmed in 12. High serum FSH was detected in nine. On logistic regression model analysis, a history of UDT was the only significant factor for high serum FSH. The incidence of high serum FSH in patients with UDT was significantly higher than that in those without UDT (58.3% vs 7.5%, $p < 0.01$). When stratified by severity of HS and the presence of UDT, high serum FSH was detected in 70% in patients with severe HS and UDT, whereas less than 10% in other groups.

CONCLUSION(S): A history of UDT was a significant factor for post-pubertal high serum FSH in patients with HS. Accordingly, the presence of UDT may be a marker for impaired spermatogenesis in patients with HS, especially in severe cases.

PMC Identifier

30820650 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=30820650>]

Author NameID

Moriya, Kimihiko; ORCID: <http://orcid.org/0000-0001-6161-9403>

Institution

(Moriya, Nakamura, Kon, Nishimura, Kanno, Kitta, Shinohara) Department of Renal and Genitourinary Surgery, Hokkaido University Graduate School of Medicine, North-15, West-7, Kita-Ku, Sapporo, 060-8638, Japan

Publisher

NLM (Medline)

Year of Publication

2019

58.

Laparoscopic inguinal exploration for impalpable undescended testis: can we avoid the open inguinal exploration altogether?.

Pathak M., Suchiang B., Saxena R., Sinha A., Rathod K., Jadhav A.

Embase

Journal of Pediatric Endoscopic Surgery. 1 (4) (pp 167-170), 2019. Date of Publication: 01 Dec 2019.

[Article]

AN: 2003828399

Introduction: Diagnostic laparoscopy is the gold standard for evaluating a child with impalpable undescended testis (UDT). During the diagnostic laparoscopy, if the vas and vessels are seen coursing through the inguinal canal, the standard norm is to explore the inguinal canal via an inguinal crease incision. In this study, however, we explored the feasibility of laparoscopic inguinal exploration without any additional inguinal crease incision.

Material(s) and Method(s): The prospective study was done from 1.1.2019 to 30.6. 2019 to assess the feasibility of the laparoscopic inguinal exploration in cases of impalpable undescended testis, where testicular vessels are found to course into the inguinal canal during a diagnostic laparoscopy. The data including the descriptive characteristics, intraoperative findings, and surgical technique were collected and analysed.

Result(s): There were 17 cases of impalpable UDT operated in the Department of Pediatric Surgery from January 2019 to June 2019. Mean age of the patients was 8.5 years (1-16 years). Two patients had testicular vas and vessels coursing through the inguinal canal, both of which underwent laparoscopic inguinal exploration. In both of these cases, vas and vessels were found

to end in testicular nubbin, at the level of the neck of the scrotum, which were excised laparoscopically, thus avoiding the open inguinal incision.

Conclusion(s): Laparoscopic inguinal exploration is a feasible and attractive alternative in cases of impalpable UDT where testicular vas and vessels are found to course through the inguinal ring.

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Status

Embase

Author NameID

Pathak, Manish; ORCID: <http://orcid.org/0000-0001-6385-1812>

Institution

(Pathak, Suchiang, Saxena, Sinha, Rathod, Jadhav) Department of Pediatric Surgery, All India Institute of Medical Sciences, Jodhpur 342005, India

Publisher

Springer (Singapore)

Year of Publication

2019

59.

Gonadal function and testicular histology in males with Prader-Willi syndrome.

Matsuyama S., Matsui F., Matsuoka K., Iijima M., Takeuchi M., Ida S., Matsumoto F., Mizokami A.

Embase

Endocrinology, Diabetes and Metabolism. 2 (1) (no pagination), 2019. Article Number: e00049.

Date of Publication: 01 Jan 2019.

[Article]

AN: 624685774

Context: Cryptorchidism is common in Prader-Willi syndrome (PWS) males, but the testicular histology in childhood remains uncertain. The association between testicular histology and long-term gonadal function in PWS males is also unknown.

Objective(s): To evaluate the relationship between testicular histology in childhood and long-term gonadal function in PWS males.

Patients and Methods: Forty men with PWS were assessed longitudinally at our institute over the past 24 years. Clinical examinations and blood tests for LH, FSH and testosterone levels were compared with normal reference values. Tissue specimens were collected during orchiopexy and analyzed based on Nistal categories.

Result(s): Of nine testes available for pathological assessments, two showed favourable histology (Nistal I) and seven showed unfavourable histology (Nistal II or III). Of five postpubertal males with histology available, four reached puberty spontaneously, but only one reached Tanner stage 5. In a male with favourable histology, LH and FSH were high, but testosterone was normal, though below the average of the reference range. In three males with unfavourable histology, LH was normal, but FSH was highly elevated, and testosterone was at the lower limit of normal. One patient took hCG treatment to induce puberty; this patient showed favourable histology, but LH, FSH and testosterone were not elevated in adolescence.

Conclusion(s): Testicular histology of PWS men in childhood varies from normal to Sertoli Cell-Only Syndrome. Regardless of the testicular histology in childhood, hypogonadism in PWS adults arises as a consequence of primary testicular dysfunction with highly elevated FSH and insufficient testosterone levels.

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Status

Embase

Author NameID

Matsuyama, Satoko; ORCID: <http://orcid.org/0000-0002-2460-0313> Matsui, Futoshi; ORCID: <http://orcid.org/0000-0002-7102-6115>

Institution

(Matsuyama, Matsui, Matsumoto) Department of Urology, Osaka Women's and Children's Hospital, Osaka, Japan (Matsuyama, Iijima, Mizokami) Department of Integrative Cancer Therapy and Urology, Kanazawa University Graduate School of Medical Science, Kanazawa, Japan

(Matsuoka, Takeuchi) Department of Pathology, Osaka Women's and Children's Hospital, Osaka, Japan

(Ida) Department of Gastroenterology and Endocrinology, Osaka Women's and Children's Hospital, Osaka, Japan

Publisher

Wiley-Blackwell Publishing Ltd (E-mail: info@royensoc.co.uk)

Year of Publication

2019

60.

Innovative method of diagnostics abdominal forms of cryptorchism at the children for prevention of infertility.

Baibakov V.M.

Embase

Wiadomosci lekarskie (Warsaw, Poland : 1960). 72 (12 cz 1) (pp 2334-2338), 2019. Date of Publication: 2019.

[Article]

AN: 631145031

OBJECTIVE: Introduction: Topicability of this problem is caused by well-spread of cryptorchism among children: from 2-3% at newborn boys in UK to 10-12% of newborn boys in the Post Soviet Union countries. Degenerative processes, occurred in testicle could lead to the development of eunuchoidism, feminization, gynecomastia, infantilism. In the future, changes in seminiferous epithelium contribute to the development of male infertility, impotence and malignant tumors - seminoma, teratoblastoma.

PATIENTS AND METHODS: The aim: To improve diagnosis for abdominal forms of cryptorchism at the children through implementation innovative methods in practice (laparoscopic diagnostics). Material(s) and Method(s): For a period from 2014 to 2017 years were carried out 43 diagnostic laparoscopy of non palpated testicle syndrome. By age children were distributed into the following groups: up to 1 year -18 children, 1-2 years - 25 children.

RESULT(S): Results: Analyzing results of our research there is no doubt that laparoscopy is one of the most reliable methods of diagnosis abdominal cryptorchism in children. This method allows both to determine location of a damaged testicle in the abdominal cavity, but also to assess its condition, developed further tactics of treatment.

CONCLUSION(S):

Conclusion(s): Laparoscopic diagnosis abdominal forms of cryptorchism is the most reliable method in a comparison with computed tomography, ultrasound, radioisotope studies. Procedure of laparoscopic diagnosis could show not only location and condition of the testis, blood vessels and ductus deferens, but help to develop the further treatment tactics.

PMC Identifier

32124749 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=32124749>]

Institution

(Baibakov) Dnipro Medical Institute of Traditional and Non-Traditional Medicine, Dnipro, Ukraine
Publisher
NLM (Medline)
Year of Publication
2019

61.

Phthalate-induced fetal leydig cell dysfunction mediates male reproductive tract anomalies.

Wang Y., Ni C., Li X., Lin Z., Zhu Q., Li L., Ge R.-S.

Embase

Frontiers in Pharmacology. 10 (no pagination), 2019. Article Number: 01309. Date of Publication: 2019.

[Review]

AN: 629884409

Male fetal Leydig cells in the testis secrete androgen and insulin-like 3, determining the sexual differentiation. The abnormal development of fetal Leydig cells could lead to the reduction of androgen and insulin-like 3, thus causing the male reproductive tract anomalies in male neonates, including cryptorchidism and hypospadias. Environmental pollutants, such as phthalic acid esters (phthalates), can perturb the development and differentiated function of Leydig cells, thereby contributing to the reproductive toxicity in the male. Here, we review the epidemiological studies in humans and experimental investigations in rodents of various phthalates. Most of phthalates disturb the expression of various genes encoded for steroidogenesis-related proteins and insulin-like 3 in fetal Leydig cells and the dose-additive effects are exerted after exposure in a mixture.

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Status

In-Process

Institution

(Wang, Ni, Li, Ge) Department of Anesthesiology, Second Affiliated Hospital, Wenzhou Medical University, Wenzhou, China (Lin, Zhu, Li, Ge) Center of Scientific Research, Second Affiliated Hospital, Wenzhou Medical University, Wenzhou, China

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2019

62.

Reliability of thermocautery-assisted circumcision: retrospective analysis of circumcision performed voluntarily in countries of low socioeconomic status.

Cakiroglu B., Gozukucuk A., Arda E., Tas T.

Embase

Therapeutic Advances in Urology. 11 (no pagination), 2019. Date of Publication: 2019.

[Article]

AN: 2003340526

Objective: The objective of this study was to evaluate the reliability of thermocautery-assisted circumcision performed voluntarily in patients of poor countries.

Material(s) and Method(s): Between 2016 and 2019, 32,000 children aged 7 days to 17 years were circumcised in multiple countries. The patients' urological examinations were done before the administration of local anaesthesia. Patients revealed to have undescended testicle, inguinal hernia, hypospadias, varicocele, penile rotation anomaly, epispadias and infection were not circumcised. All procedures were performed under local anaesthesia by using thermocautery. Afterwards, mucosa and skin were sutured using absorbable suture and the circumcised penis was dressed. Patients were instructed to remove the dressing after 3 days.

Result(s): Bleeding, requiring surgical intervention and drug reactions were not observed. The most observed complication was mucosal oedema, which occurred in approximately one-quarter of patients (26%, 8320/32,000) and continued for 3-5 days after the surgery. The most serious complication was a trapped penis, which occurred in 25 patients (0.078%). In six (0.018%) cases, meatal stenosis developed. Wound infection developed in only 10 (0.03%) cases, through the formation of an aseptic environment. Penile adhesion was seen in 35 cases (0.1%) and improved with anti-inflammatory treatment without any surgical intervention.

Conclusion(s): Thermocautery-assisted circumcision can be used as an effective, safe and useful technique with few complications and rapid healing rates.

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Status

In-Process

Author NameID

Cakiroglu, Basri; ORCID: <http://orcid.org/0000-0001-5337-5226>

Institution

(Cakiroglu) Department of Urology, Hisar Intercontinental Hospital, Saray Mah, Siteyolu
Cad.No.7-9, Istanbul 34768, Turkey (Gozukucuk) Department of Pediatric Surgery, Hisar
Intercontinental Hospital, Istanbul, Turkey

(Arda) Medical Faculty, Department of Urology, Trakya University, Edirne, Turkey

(Tas) Istanbul Cerrahi Hospital, Department of Urology, Esenyurt University, Istanbul, Turkey

Publisher

SAGE Publications Inc. (E-mail: claims@sagepub.com)

Year of Publication

2019

63.

Does the Apoptosis Pathway Play a Critical Role in Gonocyte Transformation?.

Tien M.Y., Abeydeera S.A., Cho H.-J., Sarila G., Catubig A., Burton E., Hutson J., Li R.

Embase

Journal of Pediatric Surgery. (no pagination), 2019. Date of Publication: 2019.

[Article]

AN: 2003701574

Purpose: Undescended Testes (UDT) are prevalent in 2%-5% of male infants and cause malignancy and infertility. During germ cell development, abnormal gonocytes usually undergo apoptosis. This process is believed to involve BAX (Bcl-2 Associated X) protein in clearing abnormal gonocytes which may fail in UDT, resulting in persisting gonocytes causing seminomas later in life. We aim to investigate the role of BAX in gonocyte apoptosis.

Material(s) and Method(s): BAXKO (BAX-knockout) mice were back-crossed to OG2 mice (Oct4-promoter driving enhanced green fluorescent protein-eGFP) to produce BAXOG2 mice. Testes (wildtype-BAX+/+, heterozygous-BAX+/- and homozygous-BAX-/- mice, n = 6/group) on postnatal days 1, 3, 6, 9 were fixed and embedded in OCT for frozen sectioning. Sections were labeled with Anti-Mullerian Hormone (Sertoli cell marker), Mouse Vasa Homolog (germ cell marker) and DAPI (nucleus marker) and imaged using confocal microscopy. Oct4-GFP + ve germ cells, germ cells

on/off the basement membrane and Sertoli cells were counted using ImageJ followed by data analysis with GraphPad.

Result(s): BAX^{-/-} OG2 mice had significantly higher number of germ cells/tubule comparing to BAX^{+/+} OG2 on day 9. There were Oct4-GFP + ve gonocyte-like germ cells that persisted in the center of the tubules in BAX^{-/-} OG2 even after the completion of gonocyte transformation. This suggests that abnormal gonocytes in BAX^{-/-} OG2 mice failed to undergo apoptosis and are allowed to persist.

Conclusion(s): This study demonstrated that apoptosis is important in regulating germ cell migration and differentiation during gonocyte transformation in neonatal mice. In addition, inhibition of apoptosis results in persisting neonatal gonocytes which might become seminomas in patients with UDT.

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PMC Identifier

31711746 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31711746>]

Status

Article-in-Press

Institution

(Tien, Abeydeera, Sarila, Catubig, Burton, Hutson, Li) Douglas Stephens Surgical Research Unit, Murdoch Children's Research Institute, Melbourne, Australia (Tien, Burton, Hutson, Li)

Department of Paediatrics, University of Melbourne, Melbourne, Australia

(Cho) Biological Optical Microscopy Platform, University of Melbourne, Melbourne, Australia

(Hutson) Department of Urology, The Royal Children's Hospital, Melbourne, Australia

Publisher

W.B. Saunders

Year of Publication

2019

64.

Fetal Safety of Dydrogesterone Exposure in the First Trimester of Pregnancy.

Koren G., Gilboa D., Katz R.

Embase

Clinical Drug Investigation. (no pagination), 2019. Date of Publication: 2019.

[Article]

AN: 2003506744

Background: The progestin dydrogesterone (DYD) is widely used for threatened and recurrent miscarriages, as well as for dysfunctional bleeding, infertility and other obstetric and gynecological indications. While its apparent efficacy has been compared to other progestins, its fetal safety has been only sparsely investigated.

Objective(s): To follow up fetal outcome after gestational exposure to DYD and compare it to a non-exposed comparison group.

Objective(s): To follow up fetal outcome after gestational exposure to DYD and compare it to a non-exposed comparison group.

Patients and Methods: Using the 2.5 million patient database of Maccabi Health Services, we compared rates of congenital malformations among babies exposed in utero during the first trimester of pregnancy to DYD between Jan 1999 and December 2016, to a comparison group not receiving this medication. From the DYD group we excluded all cases with concomitant exposure to in vitro fertilization (IVF) and other forms of assisted reproductive technology (ART).

Result(s): There were 8508 children exposed to DYD during the first trimester of pregnancy (4417 males, 4091 females) out of 777,422 cases in the database. After excluding from the DYD group cases with concomitant exposure to IVF and other ART, DYD exposure was associated with increased risk for hypospadias [OR 1.28 (95% confidence interval 1.06-1.55)], for overall cardiovascular malformations [OR 1.18 (91.06-1.33)], spina bifida [OR 2.29 (1.32-3.97)] and hydrocephalus [OR 2.04 (1.28-3.25)]. In a sensitivity analysis, including also cases exposed to IVF and ART in addition to DYD, there were also increased risks for cryptorchidism [1.37 (1.19-1.58)] and congenital dislocation of the hip [OR 1.58 (1.42-1.78)].

Conclusion(s): DYD confers teratogenic effects after exposure to the recommended doses in pregnant women. The risks of hypospadias and cryptorchidism have biological plausibility by the known effects on male genitalia, as is the risk for spina bifida, by the proven decrease in folic acid levels. Some of these adverse fetal effects appear to be further augmented by concomitant use of IVF and ART.

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PMC Identifier

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Article-in-Press

Author NameID

Koren, Gideon; ORCID: <http://orcid.org/0000-0002-9234-0875>

Institution

(Koren) Adelson Faculty of Medicine, Ariel University, Ariel 40700, Israel (Koren) Motherisk
Israel Program, Zerifin, Israel
(Koren, Gilboa, Katz) Kahn-Sagol-Maccabi Institute for Research and Innovation, Tel Aviv, Israel
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65.

Variability among Canadian pediatric surgeons and pediatric urologists in the management of cryptorchidism in boys before the publication of major guidelines: a retrospective review of a single tertiary centre.

Kim J.K., Chua M.E., Ming J.M., Lee M.J., Kesavan A., Kahn N., Langer J.C., Lorenzo A., Bagli D., Farhat W.A., Papanikolaou F., Koyle M.A.

Embase

Canadian journal of surgery. Journal canadien de chirurgie. 62 (3) (pp 1-6), 2019. Date of Publication: 22 Mar 2019.

[Article]

AN: 626998294

Background: Before 2014, there was a lack of recommendations on managing cryptorchidism, or undescended testis (UDT), from a large pediatric urological or surgical organization. We assessed the variability in management of UDT among pediatric urologists and pediatric surgeons at a single tertiary pediatric referral centre before publication of major guidelines.

Method(s): We performed a retrospective review of the electronic records of patients who underwent primary unilateral or bilateral orchidopexy at our centre between January 2012 and January 2014.

Result(s): A total of 488 patients (616 testes) were identified, of whom 405 (83.0%) and 83 (17.0%) were managed by pediatric urologists and pediatric surgeons, respectively. There was no difference in baseline characteristics, including age seen in clinic or at surgery, testis location/palpability and availability of preoperative ultrasonograms, of patients seen by the 2 groups. Pediatric surgeons ordered preoperative ultrasonography more often than pediatric urologists (25.3% v. 3.7%, $p < 0.001$). With palpable UDTs, although both groups used open

approaches, pediatric urologists preferred a scrotal approach (56.9%), and pediatric surgeons approached most testes inguinally (98.8%). With nonpalpable UDTs, laparoscopic approaches were preferred by both groups; however, pediatric urologists used a 2-stage Fowler-Stephens approach more often than pediatric surgeons (48.4% v. 15.8%, $p < 0.001$).

Conclusion(s): There was wide variation in the management of primary UDT between pediatric urologists and pediatric surgeons before the publication of guidelines. The most prominent difference between the 2 groups was in the ordering of preoperative ultrasonography. Future assessment of change in practice patterns may elucidate whether guidelines are an effective tool for standardization of practice.

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Institution

(Kim, Chua, Ming, Lee, Kesavan, Kahn, Langer, Lorenzo, Bagli, Farhat, Papanikolaou, Koyle)
From the Faculty of Medicine, University of Toronto, Toronto, Ont. (Kim, Lee); the Division of Urology, The Hospital for Sick Children, Toronto, Ont. (Kim, Chua, Ming, Lee, Kesavan, Kahn, Lorenzo, Bagli, Farhat, Papanikolaou, Koyle); the School of Medicine, Royal College of Surgeons in Ireland, Dublin, Republic of Ireland (Kesavan); the Division of General and Thoracic Surgery, The Hospital for Sick Children, Toronto, Ont., (Langer); and the Department of Surgery, University of Toronto, Toronto, Ont. (Langer, Lorenzo, Bagli, Farhat, Papanikolaou, Koyle)

Publisher

NLM (Medline)

Year of Publication

2019

66.

The importance of surgical timing in inguinoscrotal surgical pathologies. Inguinoskrotal cerrahi patolojilerde cerrahi zamanlamanin onemi <Inguinoskrotal cerrahi patolojilerde cerrahi zamanlamanin onemi.>

Kaya M., Ozkan A., Kabaklioglu M.

Embase

Duzce Medical Journal. 21 (3) (pp 181-185), 2019. Date of Publication: 2019.

[Article]

AN: 2003446135

Aim: Various inguinal pathologies can occurred if the processus vaginalis cannot closed fully. The aim of this study was to evaluate all patients who underwent inguinoscrotal surgery operations between 2011 and 2018 in our clinic, in terms of age, gender and accompanying with another operation, retrospectively.

Material(s) and Method(s): In this study, records of 807 patients who were performed inguinal surgery operations including 558 inguinal hernia repair, 184 orchiopexy and 65 hydroselectomy between 2011 and 2018 at Duzce University, Faculty of Medicine, Pediatric Surgery Department were evaluated retrospectively.

Result(s): Mean age of the 558 patients who underwent inguinal hernia operation was 3.0+/-3.6 years, and 288 (51.6%) patients were older than 2 years of age. Of the patients who performed inguinal hernia operation, 411 (73.7%) were male and 147 (26.3%) were female. There was a statistically significant difference in terms of inguinal hernia repair side according to gender ($p=0.038$), and left inguinal hernia repair rate in females was detected higher than in males. Mean age of the 184 patients who performed orchiopexy operation was 4.0+/-3.4 years old, and only 46 patients (25.0%) who underwent orchiopexy were younger than 2 years of age. Mean age of the 65 patients who performed hydrocele operation was 4.6+/-4.1 years old.

Conclusion(s): According to the results of this study, it is seen that the community does not have enough information about the right operation time of inguinal hernia and undescended testis, and that the society should be informed about this issue.

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Status

Embase

Author NameID

Kaya, Murat; ORCID: <http://orcid.org/0000-0001-6650-0145> Ozkan, Aybars; ORCID: <http://orcid.org/0000-0003-0214-4203>

Kabaklioglu, Murat; ORCID: <http://orcid.org/0000-0002-6642-9294>

Institution

(Kaya, Ozkan, Kabaklioglu) Duzce University, Medical Faculty, Department of Pediatric Surgery, Duzce, Turkey

Publisher

Duzce University Medical School (E-mail: duzcetipdergisi@gmail.com)

Year of Publication

2019

67.

A low cost and non-complicated circumcision; when, how, where, who should be made by?.

Dusuk maliyetli ve komplikasyonsuz bir sunnet; ne zaman, nasıl, nerede, kim tarafından yapılmalı? <Dusuk maliyetli ve komplikasyonsuz bir sunnet; ne zaman, nasıl, nerede, kim tarafından yapılmalı?.>

Kaya M., Ozkan A., Kabaklıoğlu M.

Embase

Konuralp Tıp Dergisi. 11 (3) (pp 440-443), 2019. Date of Publication: 2019.

[Article]

AN: 2004765994

Objective: Circumcision, in Turkey as a Muslim country is one of the most commonly performed operation. The aim of this study is to retrospectively evaluated circumcision datas and discuss about 'a low cost and non-complicated circumcision; when, how, where, who should be made by'.

Method(s): 4059 patients who were circumcised between 2011-2018 were included in the study. The age of the patient, time of operation, presence of accompanying surgical pathology (inguinal hernia, undescended testis, hydrocele, etc.) and anesthesia type (local only, sedoanalgesia and general) were evaluated.

Result(s): The mean age of 4059 patients was 4.1 years (+/- 3.29). The number of patients who had circumcised <2year was 1315 (32.40%), aged 2-6 who no recommended by child psychiatrists was 1154 (28.43%) and >6 year was 1590 (39.17%). 422 (10.40%) of patients who underwent circumcision had additional surgical pathology. The most common additional surgical pathologies were inguinal hernia (n=212, 50.24%), undescended testes (n=100, 23.70%) and hydrocele (n=32, 7.58%).

Conclusion(s): Since the circulation of complicated surgical operations is intense, especially in the 3rd level public university hospitals are more appropriate to perform circumcision in patients who require additional operation to reduce the cost and hospitalization. If additional operation is not required, circumcision should be performed by private health institutions or certified 1st level family physicians. In terms of childhood psychology, it is important to take measures to increase the level of knowledge of families at this issue, since a significant number of families still circumcised their children aged 2-6 years.

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Status

Embase

Institution

(Kaya, Ozkan, Kabaklioglu) Department of Pediatric Surgery, Duzce University, Medical Faculty,
Duzce, Turkey

Publisher

Duzce University Medical School (E-mail: duzcetipdergisi@gmail.com)

Year of Publication

2019

68.

The structure-activity relationship (SAR) for phthalate-mediated developmental and reproductive toxicity in males.

Li X., Mo J., Zhu Q., Ni C., Wang Y., Li H., Lin Z.-K., Ge R.-S.

Embase

Chemosphere. 223 (pp 504-513), 2019. Date of Publication: May 2019.

[Review]

AN: 2001620406

Testicular dysgenesis syndrome includes the hypospadias, cryptorchidism and abnormal fetal testis in male neonate. This is possibly caused by the environmental phthalates, which down-regulate the expression of androgen synthetic genes and *Insl3* or directly inhibits steroidogenic enzymes. There are distinct structure-activity relationships (SARs) for phthalate-mediated developmental and reproductive toxicity. Here, we review the SAR for phthalate-mediated testicular dysgenesis syndrome. Of phthalates of straight side chains, C5-C6 ones are the most potent, C4 or C7 are moderate, C3 is weakest, and C1-2 or C8-13 are ineffective. The branching and unsaturation of side chains increases the toxicity. The cycling of side chains does not increase the toxicity.

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Embase

Institution

(Li, Mo, Zhu, Ni, Wang, Li, Lin, Ge) Department of Anesthesiology, The Second Affiliated Hospital and Yuying Children's Hospital of Wenzhou Medical University, Wenzhou, Zhejiang, China

Publisher

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Year of Publication

2019

69.

Long-term outcome in a case series of Denys-Drash syndrome.

Roca N., Munoz M., Cruz A., Vilalta R., Lara E., Ariceta G.

Embase

Clinical Kidney Journal. 12 (6) (pp 836-839), 2019. Date of Publication: 25 Feb 2019.

[Article]

AN: 630562014

Background: Denys-Drash syndrome (DDS) is a rare disease caused by mutations in exons 8 and 9 of the WT1 gene. It is characterized by the association of early onset steroid-resistant nephrotic syndrome (SRNS), Wilms' tumour and, in some patients, intersex disorders, with increasing risk of gonadoblastoma. There are few published data concerning the long-term outcome of patients with DDS. The aim of this study was to report our experience.

Method(s): Data were collected from five children (three boys) with confirmed DDS diagnosed from 1996 to 2017. The mean follow-up of these patients was 16 years.

Result(s): The patients presented with SRNS and diffuse mesangial sclerosis at renal biopsy. All patients were hypertensive and progressed to end-stage kidney disease, initiating dialysis at a mean age of 28 months. Three patients developed Wilms' tumour 9 months after the SRNS was identified, which was treated by nephrectomy and chemotherapy. All five patients received kidney transplantation. SRNS did not recur after transplantation in any of the patients and graft survival was similar to that of other kidney transplant recipients in our programme. All three boys had ambiguous genitalia and cryptorchidism but a confirmed male karyotype (46, XY). One girl presented with gonadal agenesis, whereas the other one had normal female ovarian tissue and external genitalia. Both girls had a female karyotype (46, XX). Gonadoblastoma was not observed at any case.

Conclusion(s): Early DDS recognition in patients with SRNS is crucial due to its low prevalence, the specific treatment approach required and early detection of Wilms' tumour. Few data are available regarding long-term outcomes.

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Status

Embase

Institution

(Roca) Pediatric Nephrology, Consorci Hospitalari de Vic Ringgold Standard Institution, Francesc Pla el Vigata ndegree1, Vic 08500, Spain (Munoz, Cruz, Vilalta) Pediatric Nephrology, Hospital Vall d'Hebron Ringgold Standard Institution, Barcelona, Catalunya, Spain

(Lara) Pediatric Nephrology, Hospital Vall d'Hebron, Barcelona, Spain

(Ariceta) Pediatric Nephrology, Hospital Universitario Vall d'Hebron, Passeig de la Vall d'Hebron, Barcelona, Spain

Publisher

Oxford University Press

Year of Publication

2019

70.

Evaluation and Management of the Undescended Testis in Puerto Rico: A Single Surgeon's 10 Years of Experience.

Soto-Palou F.G., Escudero-Chu K., Pineyro-Ruiz C., Jorge J.C., Perez-Brayfield M.

Embase

Puerto Rico health sciences journal. 38 (4) (pp 268-270), 2019. Date of Publication: 01 Dec 2019.

[Article]

AN: 630622057

OBJECTIVE: Cryptorchidism is an abnormality of the male genitourinary tract in which one or both testes fail to descend into the scrotum. The American Urological Association (AUA) clinical guidelines for the evaluation and treatment of cryptorchidism were recently published. We reviewed our experience with the evaluation and management of our patients and examined our findings with respect to the AUA and European Association of Urology (EAU) guidelines.

METHOD(S): Data were obtained from pediatric patients who underwent a surgical intervention for an undescended testis from 2007 through 2017 at HIMA Hospital and the University Pediatric Hospital (both in Puerto Rico); all the surgeries were performed by the same surgeon. A total of 754 patients were identified; 142 patients were excluded due to lack of follow-up data (N = 612). The data obtained included age, testes locations, radiologic and surgical findings, and postoperative results.

RESULT(S): At their initial evaluations, a large proportion of the patients (46.4%) came accompanied with radiographic imaging. These findings were consistent with those of the physical examination in 58.5% of the patients and with the surgical findings in 63.1% (sensitivity 77.9%, specificity 45.8%). Our data showed that the median referral age was 24 months, which suggests that there was a significant delay in diagnosis. At the time of surgery, the average age of the patients who required an orchiectomy was 3.93 years, while those who underwent an orchiopexy had an average age of 3.28 years.

CONCLUSION(S): Our data reveal that, despite its lack of usefulness, radiologic imaging continues to be included in the diagnostic workups of children newly identified with cryptorchidism in Puerto Rico. In addition, and contrary to the guidelines, there tends to be a significant delay in treatment with surgical intervention. It is important to continue to educate our referring physicians on the AUA and EUA guidelines in order to create awareness and encourage the proper diagnostic and treatment approach for cryptorchidism.

PMC Identifier

31935315 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31935315>]

Institution

(Soto-Palou, Escudero-Chu, Perez-Brayfield) Urology Section, Department of Surgery, School of Medicine, University of Puerto Rico Medical Sciences Campus, PR, San Juan, Argentina

(Pineyro-Ruiz, Jorge) Department of Anatomy and Neurobiology, School of Medicine, University of Puerto Rico Medical Sciences Campus, PR, San Juan, Argentina

Publisher

NLM (Medline)

Year of Publication

2019

Cryptorchidism after the Fukushima Daiichi Nuclear Power Plant accident: causation or coincidence?.

Kojima Y., Yokoya S., Kurita N., Idaka T., Ishikawa T., Tanaka H., Ezawa Y., Ohto H.

Embase

Fukushima journal of medical science. 65 (3) (pp 76-98), 2019. Date of Publication: 2019.

[Article]

AN: 630572940

Cryptorchidism (undescended testes) is among the most common congenital diseases in male children. Although many factors have been linked to the incidence of cryptorchidism, and testicular androgen plays a key role in its pathogenesis, the cause remains unknown in most cases. Recently, a Japanese group published a speculative paper entitled, "Nationwide increase in cryptorchidism after the Fukushima nuclear accident." Although the authors implicated radionuclides emitted from the Fukushima accident as contributing to an increased incidence of cryptorchidism, they failed to establish biological plausibility for their hypothesis, and glossed over an abundance of evidence and expert opinion to the contrary. We assessed the adequacy of their study in terms of design setting, data analysis, and its conclusion from various perspectives. Numerous factors must be considered, including genetic, environmental, maternal/fetal, and social factors associated with the reporting of cryptorchidism. Other investigators have established that the doses of external and internal radiation exposure in both Fukushima prefecture and the whole of Japan after the accident are too low to affect testicular descent during fetal periods; thus, a putative association can be theoretically and empirically rejected. Alternative explanations exist for the reported estimates of increased cryptorchidism surgeries in the years following Japan's 2011 earthquake, tsunami, and nuclear crisis. Data from independent sources cast doubt on the extent to which cryptorchidism increased, if at all. In any case, evidence that radionuclides from the Fukushima Daiichi Nuclear Power Plant could cause cryptorchidism is lacking.

PMC Identifier

31915325 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31915325>]

Institution

(Kojima) Department of Urology, Fukushima Medical University School of Medicine (Yokoya)

Thyroid and Endocrine Center, Fukushima Medical University School of Medicine

(Kurita) Department of Clinical Epidemiology, Graduate School of Medicine, Fukushima Medical University

(Kurita) Department of Innovative Research and Education for Clinicians and Trainees (DiRECT), Fukushima Medical University Hospital

(Kurita) Center for Innovative Research for Communities and Clinical Excellence (CiRC2LE),

Fukushima Medical University

(Idaka) Medical Research Center, Fukushima Medical University School of Medicine
(Ishikawa) Department of Radiation Physics and Chemistry, Fukushima Medical University
(Tanaka) Department of Pediatric Surgery, Fukushima Medical University Hospital
(Ezawa) Medical Affairs Division, Fukushima Medical University Hospital
(Ohto) Radiation Medical Science Center for the Fukushima Health Management Survey,
Fukushima Medical University

Publisher

NLM (Medline)

Year of Publication

2019

72.

Gonadotropin-Induced Spermatogenesis in CHH Patients with Cryptorchidism.

Liu Z., Mao J., Xu H., Wang X., Huang B., Zheng J., Nie M., Zhang H., Wu X.

Embase

International Journal of Endocrinology. 2019 (no pagination), 2019. Article Number: 6743489.

Date of Publication: 2019.

[Article]

AN: 2004514010

Congenital hypogonadotropic hypogonadism (CHH) patients with cryptorchidism history usually have poor spermatogenesis outcome, while researches focusing on this population are rare. This study retrospectively evaluated gonadotropin-induced spermatogenesis outcome in CHH patients with cryptorchidism (n = 40). One hundred and eighty-three CHH patients without cryptorchidism were served as control. All patients received combined gonadotropins therapy (HCG and HMG) and were followed up for at least 6 months. The median follow-up period was 24 (15, 33) months (totally 960 person-months). Sperm (>0/ml) initially appeared in semen at a median of estimated 24 months (95% confidence interval (CI) 17.8-30.2). Twenty (20/40, 50%) patients succeeded in producing sperms, and the average time to produce first sperm was 19 +/- 8 months. Five pregnancies were achieved in 9 (5/9, 56%) couples who desired for children. Compared with CHH patients without cryptorchidism (n = 183), cryptorchid patients had longer median time for sperm appearance in semen (24 months vs. 15 months, P<0.001), lower rate of spermatogenesis (50% vs. 67%, P=0.032), and lower mean sperm concentration (1.9 (0.5, 8.6) million/ml vs.

11.1(1.0, 25.0) million/ml, P=0.006) at the last visit. In conclusion, CHH patients with cryptorchidism require a longer period for gonadotropin-induced spermatogenesis. The successful rate and sperm concentration were lower than patients without cryptorchidism.

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Status

Embase

Author NameID

Mao, Jiangfeng; ORCID: <http://orcid.org/0000-0003-0157-1545> Wu, Xueyan; ORCID:

<http://orcid.org/0000-0003-2540-8590>

Liu, Zhaoxiang; ORCID: <http://orcid.org/0000-0001-7016-5917>

Institution

(Liu, Mao, Xu, Wang, Huang, Zheng, Nie, Wu) Key Laboratory of Endocrinology, Department of Endocrinology, Peking Union Medical College Hospital, National Health Commission of People's Republic of China, Beijing, China (Liu) Beijing Tsinghua Changgung Hospital, School of Clinical Medicine, Tsinghua University, Beijing, China

(Zhang) Department of Physiology, State Key Laboratory of Medical Molecular Biology, Institute of Basic Medical Sciences, School of Basic Medicine, Peking Union Medical College, Chinese Academy of Medical Sciences, Beijing, China

Publisher

Hindawi Limited (410 Park Avenue, 15th Floor, 287 pmb, New York NY 10022, United States)

Year of Publication

2019

73.

Congenital Spigelian hernia and ipsilateral cryptorchidism: a new syndrome?.

Patoulas I., Rahmani E., Patoulas D.

Embase

Folia medica Cracoviensia. 59 (4) (pp 71-78), 2019. Date of Publication: 2019.

[Article]

AN: 630517768

Spigelian hernia (SH) is a rare ventral interstitial hernia occurring through a defect in the transversus abdominis aponeurosis (Spigelian fascia). Spigelian fascia is found between the

lateral border of the rectus abdominis muscle and the semilunar line, which extends from the costal cartilage to the pubic tubercle. In other words, Spigelian line is where the transversus abdominis muscle ends in an aponeurosis characterized by a congenital or acquired defect in the Spigelian aponeurosis. Pediatric cases of SH are either congenital or acquired due to trauma, previous surgery or increased intra-abdominal pressure. SH in combination with ipsilateral cryptorchidism may constitute a new syndrome, as such cases are extremely rare in the literature is new syndrome is characterized by the following congenital, ipsilateral disturbances: SH, absence of inguinal canal and gubernaculum and the homolateral testis found within the Spigelian hernia sac (a hernia sac containing undescended testis). The aim of this study is to emphasize some typical findings of this specific entity, and, hence, the necessity for a thorough investigation of the origin of the SH.

PMC Identifier

31904751 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31904751>]

Institution

(Patoulias, Rahmani) 1st Department of Pediatric Surgery, Aristotle University of Thessaloniki General Hospital 'G. Gennimatas', Thessaloniki, Greece (Patoulias) Department of Internal Medicine, General Hospital of Veria, Veria, Greece

Publisher

NLM (Medline)

Year of Publication

2019

74.

Evaluating the distance travelled for urological pediatric appointments.

Otis-Chapados S., Coderre K., Bolduc S., Moore K.

Embase

Canadian Urological Association Journal. 13 (12) (pp 391-394), 2019. Date of Publication: 2019. [Article]

AN: 2004089070

Introduction: In Quebec, eight pediatric urologists practice in three tertiary centers covering large territories. To improve the availability of pediatric urology to distant families and to reduce the economic burden on them, we examined the charts of all patients attending the pediatric

urological outpatient clinic. Our objectives were to evaluate the distance travelled by each urological pediatric outpatient and to report the most frequent urological referral complaints.

Method(s): From July 2016 to June 2017, we retrospectively reviewed the charts of all the 3604 pediatric patients seen in the outpatient urological clinic of the CHU de Quebec. We specifically focused on travel distance covered by families and the reason for referral.

Result(s): Most patients were boys (78%) and the mean age was 7.2 years. The average one-way distance travelled by each family was 69 km. The patients came more frequently from Capitale-Nationale (63.7%) and Chaudiere-Appalaches (21.9%), the closest regions. The most common reasons for consultations were postoperative followups (15%), phimosis and adhesions (14%), enuresias (14%), hydronephrosis (13%), micturition disorder (11%), and cryptorchidism and retractile testicles (8%). Of all patients seen for phimosis or cryptorchidism, only 24% and 36% of them, respectively, were scheduled for surgery.

Conclusion(s): Phimosis, cryptorchidism, and voiding disorders are the most frequent pediatric urological reasons for consultation; primary care continuing medical education seems worthwhile. It would, perhaps, be more beneficial for all to have the pediatric urologists travelling to perform clinics and surgeries in distant regions to save more than 300 km round trip to several families.

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Status

Embase

Institution

(Otis-Chapados, Coderre, Bolduc, Moore) Division of Urology, Department of Surgery, CHU de Quebec, Universite Laval, Quebec City, QC, Canada

Publisher

Canadian Urological Association (E-mail: josephine.sciortino@cua.org)

Year of Publication

2019

75.

Null variants and deletions in BRWD3 cause an X-linked syndrome of mild-moderate intellectual disability, macrocephaly, and obesity: A series of 17 patients.

Ostrowski P.J., Zachariou A., Loveday C., Baralle D., Blair E., Douzgou S., Field M., Foster A., Kyle C., Lachlan K., Mansour S., Naik S., Rea G., Smithson S., Sznajder Y., Thompson E., Cole T., Tatton-Brown K.

Embase

American Journal of Medical Genetics, Part C: Seminars in Medical Genetics. 181 (4) (pp 638-643), 2019. Date of Publication: 01 Dec 2019.

[Article]

AN: 2003606229

BRWD3 has been described as a cause of X-linked intellectual disability, but relatively little is known about the specific phenotype. We report the largest BRWD3 patient series to date, comprising 17 males with 12 distinct null variants and 2 partial gene deletions. All patients presented with intellectual disability, which was classified as moderate (65%) or mild (35%). Behavioral issues were present in 75% of patients, including aggressive behavior, attention deficit/hyperactivity and/or autistic spectrum disorders. Mean head circumference was +2.8 SD (2.8 standard deviations above the mean), and mean BMI was +2.0 SD (in the context of a mean height of +1.3 SD), indicating a predominant macrocephaly/obesity phenotype. Shared facial features included a tall chin, prognathism, broad forehead, and prominent supraorbital ridge. Additional features, reported in a minority (<30%) of patients included cryptorchidism, neonatal hypotonia, and small joint hypermobility. This study delineates the clinical features associated with BRWD3 null variants and partial gene deletions, and suggests that BRWD3 should be included in the differential diagnosis of patients with an overgrowth-intellectual disability (OGID) phenotype, particularly in male patients with a mild or moderate intellectual disability associated with macrocephaly and/or obesity.

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Status

Embase

Author NameID

Ostrowski, Philip J.; ORCID: <http://orcid.org/0000-0001-9777-5029> Foster, Alison; ORCID: <http://orcid.org/0000-0002-0216-3909>

Institution

(Ostrowski, Mansour, Tatton-Brown) South West Thames Regional Genetics Service, St George's University NHS Foundation Trust, London, United Kingdom (Zachariou) Division of Clinical Studies, Institute of Cancer Research, London, United Kingdom (Loveday) Division of Genetics and Epidemiology, Institute of Cancer Research, London, United Kingdom

(Baralle, Lachlan) Wessex Clinical Genetics Service, Princess Anne Hospital, Southampton, United Kingdom

(Baralle) Faculty of Medicine, Human Development and Health, University of Southampton, Southampton, United Kingdom

(Blair) Oxford Centre for Genomic Medicine, ACE Building, Nuffield Orthopaedic Centre, Oxford, United Kingdom

(Douzgou) Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Sciences Centre, Manchester, United Kingdom

(Douzgou) Division of Evolution and Genomic Sciences, School of Biological Sciences, University of Manchester, Manchester, United Kingdom

(Field) Genetics of Learning Disability Service, Hunter Genetics, Waratah, NSW, Australia

(Foster, Cole) West Midlands Regional Genetics Service, Birmingham Women's NHS Foundation Trust, Birmingham, United Kingdom

(Kyle) Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University Hospitals NHS Foundation Trust, Manchester, United Kingdom

(Naik) Clinical Genetics, Birmingham Women's and Children's NHS Foundation Trust, Birmingham, United Kingdom

(Rea) Northern Ireland Regional Genetics Service, Belfast City Hospital, Belfast, United Kingdom

(Smithson) Department of Clinical Genetics, St Michael's Hospital, Bristol, United Kingdom

(Sznajer) Center for Human Genetics, Cliniques Universitaires St-Luc, Universite Catholique de Louvain, Brussels, Belgium

(Thompson) South Australian Clinical Genetics Service, Women's and Children's Hospital, Adelaide, SA, Australia

(Tatton-Brown) Institute of Molecular and Clinical Sciences, St George's University of London, London, United Kingdom

Publisher

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Year of Publication

2019

Human chorionic gonadotrophin hormone for treatment of congenital undescended testis:
Anatomical barriers to its success.

Elsherbeny M.S., Abdelhay S.

Embase

Journal of Pediatric Surgery. 54 (11) (pp 2413-2415), 2019. Date of Publication: November 2019.

[Article]

AN: 2001675267

Background/Purpose: Although the surgical treatment was proved to be the recommended line of management for congenital undescended testis, hormonal therapy with human chorionic gonadotrophin hormone has been started long years ago and is still used in some areas with variable degrees of success. The factors responsible for treatment failure are not well explored. In this study, we aimed to highlight the anatomical abnormalities in the congenital undescended testis that might contribute to treatment failure.

Method(s): During the period from January 2014 to December 2015, 75 boys with congenital undescended testes received treatment with human chorionic gonadotrophin, in pediatric surgery department, Faculty of medicine, Ain Shams University. Their age ranged between 6 months and 4 years (mean 1.6 years, median 2 years). In 70 boys, the testes were palpable and in the remaining 5 boys, the testes were impalpable. Fifty boys had unilateral and 25 had bilateral undescended testes. Seven of the palpable testes were high scrotal in position and the remaining 83 were palpated in the inguinal canal. The patients were followed up for 6 months to determine the position of the testis after the treatment and surgical intervention was done for those who did not respond to the hormonal treatment either partially or completely.

Result(s): Only 7 testes showed complete descent (7%) (2 bilateral and 3 unilateral) and they were initially high scrotal in position, 8 testes showed partial descent (8%) (2 bilateral and 4 unilateral) and they were inguinal in 6 which became high scrotal and impalpable in 2 which became peeping. The remaining 85 (85%) did not respond to the hormonal treatment. Upon surgical exploration, abnormal attachment of the gubernaculum was found in 83 testes (83%), 2 testes were peeping (2%), short testicular vessels were found in 4 testes (4%), 3 testes were vanishing (3%) and a closed internal ring was found in one testis (1%).

Conclusion(s): Treatment of congenital undescended testis with human chorionic gonadotrophin hormone had low success rates. Anatomical abnormalities in the congenital undescended testis might contribute to this treatment failure. Type of the study: Clinical research paper.

Level of Evidence: level III.

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Status

Embase

Institution

(Elsherbeny, Abdelhay) Department of Pediatric Surgery, Faculty of Medicine, Ain Shams University, Cairo, Egypt

Publisher

W.B. Saunders

Year of Publication

2019

77.

Residential sources of pesticide exposure during pregnancy and the risks of hypospadias and cryptorchidism: The French ELFE birth cohort.

Cognez N., Warembourg C., Zaros C., Metten M.-A., Bouvier G., Garlantezec R., Charles M.-A., Beranger R., Chevrier C.

Embase

Occupational and Environmental Medicine. 76 (9) (pp 672-679), 2019. Date of Publication: 01 Sep 2019.

[Article]

AN: 628983138

Objectives Prenatal occupational exposure to pesticides has been associated with male reproductive tract abnormalities. Little is known about the possible impact of non-occupational pesticide exposure on fetal and child development in the general population. Using data from a nationwide birth cohort, we aimed to assess the association between residential sources of prenatal pesticide exposure and the risks of hypospadias and cryptorchidism. Methods Of the 9281 boys in ELFE (French Longitudinal Study of Children), the national French birth cohort, 53 were diagnosed with hypospadias and 137 with cryptorchidism. We assessed residential exposure sources from self-reported domestic use of eight types of pesticide products and French spatial land use data with acreage within a 1000 m radius around each family's home for 21 crop types. We used logistic regression modelling, adjusted for possible confounders that included estimated dietary pesticide intake. Multiple imputations were used to handle missing data. Results An increased risk of hypospadias was associated with domestic pesticide use against fleas and ticks (OR=2.28, 95% CI 1.09 to 4.75); no associations were found between

cryptorchidism and any domestic pesticide use. Slightly increased risks of cryptorchidism were observed in association with all crop acreages near homes during pregnancy, especially for orchards, and no association was observed for hypospadias. Conclusions Our results suggest a possible increased risk of hypospadias associated with prenatal use of some domestic pesticide products, likely to contain insecticides, and of cryptorchidism with nearby orchard acreage (crops repeatedly sprayed with pesticides). This work is limited by its modest number of cases.

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Institution

(Cognez, Warembourg, Chevrier) UnivRennes, Inserm, EHESP, Irset-UMR-S 1085, Rennes 35042, France (Warembourg) ISGlobal, Barcelona, Spain

(Zaros) Ined, Inserm, EFS, ELFE Joint Unit, Rennes, France

(Metten, Garlandezec, Beranger) UnivRennes, CHU Rennes, Inserm, EHESP, Irset-UMR-S 1085, Rennes, France

(Bouvier) University of Bordeaux, Inserm UMR 1219 EPICENE Team, Bordeaux Population Health Research Center, Bordeaux, France

(Charles) Ined, Inserm, EFS, ELFE Joint Unit, Villejuif, France

Publisher

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Year of Publication

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78.

Oral contraceptive use and genital anomalies in sons. A Danish cohort study.

Buur L.E., Laurberg V.R., Ernst A., Arendt L.H., Nybo Andersen A.-M., Olsen J., Ramlau-Hansen C.H.

Embase

Reproductive Toxicology. 89 (pp 67-73), 2019. Date of Publication: October 2019.

[Article]

AN: 2002275832

Exposure to exogenous sex hormones with estrogenic or anti-androgen properties may influence intrauterine development of male genitals. This population-based cohort study based on data from 44,408 live-born singleton sons in the Danish National Birth Cohort (DNBC) aimed to investigate whether maternal use of oral contraceptives prior to or during early pregnancy increase the risk of cryptorchidism or hypospadias. We found no consistent association between use of oral contraceptives and cryptorchidism or hypospadias, neither in those exposed any time four months prior to conception [cryptorchidism: adjusted Odds Ratio (aOR): 1.06 (95% CI: 0.91; 1.23), hypospadias: 0.74 (95% CI: 0.53; 1.03)] nor in those exposed any time during the first trimester of pregnancy [cryptorchidism: aOR: 0.93 (95% CI: 0.53; 1.62), hypospadias: 1.02 (95% CI: 0.32; 3.23)]. Despite relatively strong exposure levels from oral contraceptive use in pregnancy, this study revealed no evidence of an increased risk of either two genital malformations.

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Status

Embase

Institution

(Buur, Laurberg, Ernst, Arendt, Olsen, Ramlau-Hansen) Department of Public Health, Aarhus University, Aarhus, Denmark (Nybo Andersen) Department of Public Health, Section of Epidemiology, University of Copenhagen, Copenhagen, Denmark

(Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

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Year of Publication

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79.

Postnatal germ cell development during first 18 months of life in testes from boys with non-syndromic cryptorchidism and complete or partial androgen insensitivity syndrome.

Li R., Azzollini D., Shen R., Thorup J., Clasen-Linde E., Cortes D., Hutson J.M.

Embase

Journal of Pediatric Surgery. 54 (8) (pp 1654-1659), 2019. Date of Publication: August 2019.

[Article]

AN: 2001546733

Introduction: Neonatal testicular germ cells/gonocytes, transform into stem cells for spermatogenesis during 'minipuberty', driving change in timing of surgery. This study examined gonocyte transformation in cryptorchid testes in children \leq 18 months of age with unilateral, bilateral undescended testes (UDT), complete or partial androgen insensitivity syndrome (CAIS, PAIS) [3,4].

Material(s) and Method(s): Testicular biopsies were taken from patients with unilateral or bilateral UDT, PAIS or CAIS, aged 10 days-18 months. These testicular sections underwent immunohistochemistry with antibodies (Oct4, Ki67, C-Kit, Sox9) followed by confocal imaging, cell counting and statistical analysis.

Result(s): Both Sertoli cells/tubule and germ cells (GC)/tubule decreased with age, and % empty tubules (no GC) increased with age but with no significant differences between patient groups. Oct4+ germ cells/tubule decreased with age. There are some GCs and Sertoli cells proliferating during the first year and most proliferating Oct4+ germ cells (Oct4+/Ki67+) were located off tubular basement membrane.

Conclusion(s): Our study showed that Oct4 expression gradually decreased after minipuberty and transformation into spermatogonia. Germ cells and Sertoli cells undergo mitosis during the first 12 months although not abundantly. We propose that Oct4+ gonocyte transformation into spermatogonia via proliferation and migration to the basement membrane may be delayed in UDT.

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Institution

(Li, Azzollini, Shen, Hutson) Douglas Stephens Surgical Research Group, Murdoch Children's Research Institute, Melbourne, Australia (Li, Hutson) Department of Paediatrics, University of Melbourne, Australia

(Thorup) Department of Paediatric Surgery, Rigshospitalet, Copenhagen, Denmark

(Thorup, Cortes) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Clasen-Linde) Department of Pathology, Rigshospitalet, Copenhagen, Denmark

(Cortes) Department of Pediatrics, Hvidovre University Hospital and University of Copenhagen, Denmark

(Hutson) Department of Urology, The Royal Children's Hospital, Melbourne, Australia

Publisher

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Year of Publication

2019

80.

A Canadian national survey: understanding the differences in management of cryptorchidism among pediatric surgeons and pediatric urologists.

Kim J.K., Chua M., Braga L., Langer J.C., Hancock B.J., Lorenzo A.J., Bagli D., Farhat W.A., Koyle M.A.

Embase

Journal of Pediatric Surgery. 54 (9) (pp 1820-1824), 2019. Date of Publication: September 2019.

[Article]

AN: 2001349977

Background: This investigation aims to assess the variability in practice patterns regarding management of children with cryptorchidism (UDT) among pediatric urologists (U) and pediatric surgeons (S) practicing in Canada.

Method(s): All active members of Pediatric Urologists of Canada (PUC) and Canadian Association of Pediatric Surgery (CAPS) were invited to participate in an online multiple-choice type questionnaire with clinical scenarios in management of UDT. Responses were compared between U and S using Fisher's exact test.

Result(s): The response rates were 74% and 79% among CAPS members (54/73) and PUC members (27/34) respectively. CAPS members were more likely to order diagnostic ultrasounds prior to surgery (44.4% vs 18.5%, $p = 0.027$). For palpable testis, most (80%) CAPS members favored the classic inguinal approach, while most PUC members did not demonstrate a clear preference, and were flexible with their approach depending on the position of the palpated testes (55%; $p < 0.001$). There was no statistically significant difference in preferred approach to unilateral or bilateral nonpalpable testis. However, for both palpable and nonpalpable bilateral

UDT, more CAPS members preferred metachronous correction, compared to PUC members who opted to approach them synchronously (p = 0.008, 0.002, respectively).

Conclusion(s): Preferences with regard to use of diagnostic tools such as US, surgical approach for palpable testes and bilateral UDTs were not consistent between the two surgical specialties who perform orchidopexy across Canada. Both groups were compliant with guideline recommendations, with the exception of utilizing preoperative ultrasounds, which is uniformly not recommended by the most recent guidelines.

Level of Evidence: This is a level II evidence study.

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Institution

(Kim) Faculty of Medicine, University of Toronto, Toronto, ON, Canada (Kim, Chua, Lorenzo,

Bagli, Farhat, Koyle) Division of Urology, The Hospital for Sick Children, Toronto, ON, Canada

(Chua) Institute of Urology, St. Luke's Medical Center, Quezon City, NCR, Philippines

(Braga) McMaster Children's Hospital and McMaster University, London, Canada

(Langer) Division of General and Thoracic Surgery, The Hospital for Sick Children, Toronto, ON, Canada

(Langer, Lorenzo, Bagli, Farhat, Koyle) Department of Surgery, University of Toronto, Toronto, ON, Canada

(Hancock) Departments of Surgery & Pediatrics and Child Health, Children's Hospital, Winnipeg, MB, Canada

Publisher

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Year of Publication

2019

81.

The Impact of Groin Surgery during Childhood on the Incidence of Inguinal Hernia Repair and Its Postoperative Complications in Adult Life.

Sokratous A., Osterberg J., Sandblom G.

Embase

European Journal of Pediatric Surgery. 29 (3) (pp 271-275), 2019. Date of Publication: 13 Apr 2019.

[Article]

AN: 621756588

Background Pediatric inguinal hernia, hydrocele, and cryptorchidism are common congenital anomalies affecting children, and require surgical intervention in some cases. The association between surgical treatment of these conditions and acquired inguinal hernia later in life is poorly understood. The aim of this cohort study was to examine the effect of groin surgery during childhood on the incidence and surgical outcome of inguinal hernia repair in adult life. Materials and Methods Data from the Swedish Inpatient Register and the Swedish Hernia Register were cross-linked using the patient personal identity numbers. The incidence of inguinal hernia repair in patients 15 years or older in the study cohort, as well as postoperative complication rates, were compared with the expected incidence and complication rates extrapolated from the general Swedish population in 2014, stratifying for age and gender. Results Note that 68,238 children aged 0 to 14 years were found to have undergone groin surgery between 1964 and 1998. The median follow-up time after an operation in the groin was 30.8 years (21.0-50.0). Of those, 1,118 were found to have undergone inguinal hernia repair as adults (> 15 years old) between 1992 and 2013. The incidence of inguinal hernia repair in the cohort was significantly higher than that expected (1.43 [1.33-1.53]), both for men (1.32 [1.25-1.41]) and women (4.30 [3.28-5.55]). The incidence was also increased in the subgroup of patients that had undergone more than one procedure during childhood. No significant impact on postoperative complication rate, reoperation rate, or operation time was identified. Conclusion Individuals undergoing surgery in the groin during childhood are at increased risk for acquired inguinal hernia surgery later in life. Inguinal surgery during childhood did not affect the outcome of hernia repair in adult age.

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Embase

Institution

(Sokratous) Surgical Department, Falu Lasarett, Lasarettsvagen 10, Falun 791 82, Sweden

(Osterberg) Surgical Department, Mora Lasarett, Mora, Sweden

(Sandblom) Division of Surgery, Department of Clinical Science and Education, Karolinska Institutet, Stockholm, Sweden

Publisher

Georg Thieme Verlag (E-mail: iaorl@iaorl.org)

Year of Publication

2019

82.

Prognostic values of shear wave elastography in adolescent boys with varicocele.

Jedrzejewski G., Osemlak P., Wieczorek A.P., Nachulewicz P.

Embase

Journal of Pediatric Urology. 15 (3) (pp 223.e1-223.e5), 2019. Date of Publication: May 2019.

[Article]

AN: 2001579774

Introduction: Shear wave elastography is an ultrasound technique for non-invasive quantification of tissue stiffness. It was used in assessing testis elasticity in some scrotal abnormalities, such as undescended pediatric testes or adult varicocele testes. In this study, its usefulness in adolescent patients with varicocele was examined.

Objective(s): The aim of this study was to quantify elasticity of testes with the use of elastography and comparison of the results with typical threshold values used in varicocele management in adolescent patients with varicoceles. Study design: In 30 patients with clinically diagnosed left varicoceles, quantitative 2D shear wave imaging of varicocele testes and contralateral ones were performed.

Result(s): The relationships between the grade of varicocele and elastography were calculated. The stiffness was 2.5 +/- 0.49 kPa in testes with grade I of varicocele, 2.59 +/- 0.81 in grade II and 2.80 +/- 0.72 kPa in grade III. In contralateral testes, it was respectively grade I 2.39 +/- 0.49 kPa, grade II 2.41 +/- 0.61 kPa, and grade III 2.42 +/- 0.85 kPa. The statistical significance was close to importance in grade III (P = 0.153). There was a statistically significant difference between elastography results in patients with volume difference over 20%. In testes with varicocele, it was 2.77 +/- 0.75 kPa and in contralateral testes, 2.37 +/- 0.65 kPa (P < 0.05). In patients with testis volume difference between 0 and 20%, elastography results were comparable, and it was 2.45 +/- 0.57 kPa in testes with varicocele and 2.44 +/- 0.61 kPa in contralateral testes (Table).

Discussion(s): Ultrasound is currently the most widely used imaging technique for the assessment of varicocele, but its role in the diagnostic algorithm is still controversial. Therefore,

many attempts are made to determine the manifestations of testicular damage that precede morphological deterioration, which could increase the importance of imaging techniques in treatment planning.

Conclusion(s): The changes of tissue elasticity due to varicocele seem to confirm the need of surgery in patients with testis volume difference more than 20% and in grade III of varicocele (Table). In case of validation of diffuse testis changes, they could indicate the need for surgery also in other stages. [Table presented]

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Author NameID

Jedrzejewski G.; ORCID: <http://orcid.org/0000-0001-7070-5477>

Institution

(Jedrzejewski, Wieczorek) Department of Pediatric Radiology Medical University of Lublin, Poland (Osemlak, Nachulewicz) Department of Pediatric Surgery Medical University of Lublin, Poland

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83.

The clinical significance of an incidentally detected open internal inguinal ring.

Valioulis I., Papageorgiou I., Ioannidou D.

Embase

Journal of Pediatric Urology. 15 (2) (pp 185.e1-185.e5), 2019. Date of Publication: April 2019.

[Article]

AN: 2001509904

Introduction and objective: An open internal inguinal ring (IIR) may be discovered incidentally either in the context of correcting pathology involving the contralateral side or at the time of

surgical exploration for reasons unrelated to a patent processus vaginalis (PPV). The aim of this study is to determine the evolution of an incidentally encountered open IIR in patients undergoing laparoscopy for reasons not associated with unilateral inguinal hernia or cryptorchidism.

Material(s) and Method(s): The authors conducted a prospective study of all patients who underwent laparoscopic surgery in the department of pediatric surgery at Agios Loukas hospital between 2004 and 2013 for various indications. Patients operated for inguinal hernia and cryptorchidism were excluded. During this period, 572 patients underwent laparoscopy for reasons not related to PPV. The median age at time of initial laparoscopy was 9,4 years (range 2 days-16 years). The IIRs were always inspected. No attempt was made to repair the open IIRs, as they were asymptomatic. Parents were informed after the operation, and instructions were given to inform us, in case that inguinal hernia symptoms manifested. The duration of the follow-up was 4 years.

Result(s): Among these 572 patients, 39 patients with 44 open IIRs were found (6,82%). From the 39 patients, 35 were male and four were female; 22 had a right open IIR, 12 had a left one, and five of them a bilateral open IIR. The median age was 7,82 years (3-14 years). Four patients were lost during follow-up. Of the remaining 35 patients with 40 open IIRs, four developed an inguinal hernia (11,43%) and were operated on with laparoscopically assisted (subcutaneous endoscopically assisted ligation [SEAL]) technique at the time of diagnosis. The study results are demonstrated on Fig. 1.

Discussion(s): The percentage of an incidentally discovered open IIR in this study is lower in comparison with studies including patients with PPV pathologies. There is a possibility, in those patients, of underlying pathology which can affect both sides. It is also lower in comparison with previous studies including younger patients. However, gender and side predominance is in accordance with most published studies. In this study group, the possibility of developing a symptomatic hernia from an asymptomatic open IIR is rather small.

Conclusion(s): An incidentally discovered open IIR in patients without symptoms, excluding those with contralateral inguinal hernias or cryptorchidism, has relatively low chance of developing an inguinal hernia. Thus, the authors support the strategy of close follow-up in these patients.[Figure presented]

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Institution

(Valioulis, Papageorgiou) 1st Department of Pediatric Surgery, G. Gennimatas Hospital, Aristotle University of Thessaloniki, Greece (Valioulis, Ioannidou) Department of Pediatric Surgery, Agios Loukas Hospital, Panorama, Thessaloniki, Greece

Publisher

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84.

Clinical and socioeconomic factors associated with delayed orchidopexy in cryptorchid boys in China: A retrospective study of 2423 cases.

Zhao T.-X., Liu B., Wei Y.-X., Wei Y., Tang X.-L., Shen L.-J., Long C.-L., Lin T., Wu S.-D., Wei G.-H.

Embase

Asian Journal of Andrology. 21 (3) (pp 304-308), 2019. Date of Publication: 01 May 2019.

[Article]

AN: 627396540

We investigated the associations of clinical and socioeconomic factors with delayed orchidopexy for cryptorchidism in China. A retrospective study was conducted on cryptorchid boys who underwent orchidopexy at Children's Hospital at Chongqing Medical University in China from January 2012 to December 2017. Of 2423 patients, 410 (16.9%) received timely repair by 18 months of age, beyond which surgery was considered delayed. Univariate analysis suggested that the laterality of cryptorchidism ($P = 0.001$), comorbidities including inguinal hernia/scrotal hydrocele ($P < 0.001$) or urinary tract disease ($P = 0.016$), and whether patients lived in a poverty county ($P < 0.001$) could influence whether orchidopexy was timely or delayed. Logistic regression analysis suggested that the following factors were associated with delayed repair: unilateral rather than bilateral cryptorchidism (odds ratio [OR] = 1.752, $P < 0.001$), absence of inguinal hernia or hydrocele (OR = 2.027, $P = 0.019$), absence of urinary tract disease (OR = 3.712, $P < 0.001$), and living in a poverty county (OR = 2.005, $P < 0.001$). The duration of postoperative hospital stay and hospital costs increased with the patient's age at the time of surgery.

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PMC Identifier

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Embase

Institution

(Zhao, Wei, Wei, Lin, Wu, Wei) Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing 400014, China (Zhao, Liu, Wei, Wei, Shen, Long, Lin, Wu, Wei) Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, Chongqing 400014, China

(Zhao, Liu, Wei, Wei, Tang, Shen, Long, Wu, Wei) Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing 400014, China

(Zhao, Liu, Wei, Wei, Tang, Shen, Long, Lin, Wu, Wei) China Intl. Science and Technology Cooperation Base of Child Development and Critical Disorders, Chongqing 400014, China

(Zhao, Liu, Wei, Tang, Shen, Long, Wu, Wei) Chongqing Key Laboratory of Pediatrics, Chongqing 400014, China

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Year of Publication

2019

85.

Early Clinical Outcome of Staged Laparoscopic Traction Orchidopexy for Abdominal Testes.

Abouheba M.A.S., Younis W., Elsokary A., Roshdy W., Waheeb S.

Embase

Journal of Laparoendoscopic and Advanced Surgical Techniques. 29 (4) (pp 531-537), 2019.

Date of Publication: 01 Apr 2019.

[Article]

AN: 627212408

Aims of the Study: To assess the short-term clinical outcome of the novel Shehata technique of laparoscopic traction - lengthening for abdominal testes in a single center over a 12-month period

(January-December 2014). An ethics approval of the study and appropriate consents were obtained for all patients before inclusion in our study.

Material(s) and Method(s): A total of 47 consecutive boys presented with impalpable testes in the ipsilateral hemiscrota, 3 of them were bilateral summing up to a total of 50 U of impalpable testes to a single center over 12 months (January-December 2014). Those boys underwent a preoperative ultrasound (US) Doppler scan for the ipsilateral and contralateral testes. They then proceeded to a first-stage laparoscopic exploration for the testes in which the cranial testicular artery and the caudal vas deferens were traced to their meeting point to locate the abdominal testes that were either found (peeping at the internal inguinal ring [IIR] or more cranially) or otherwise vanishing (intraabdominally blind-ending vas and vessels or extra-abdominally passing through the IIR). All 50 testes failed to stay at the contralateral IIR when brought there mandating a preliminary lengthening of the testicular vessels by lateral dissection, traction, and fixation to a point 1-2 cm superolateral to the contralateral anterior superior iliac spine (ASIS), essentially a mobile traction point. After 12 weeks, all underwent a second-stage laparoscopic-assisted ipsilateral subdartos orchidopexy for the testes under traction. Occasional slippage of the testis under traction mandated an otherwise second-stage retraction and a third-stage orchidopexy. All underwent US Doppler scan 3 and 6 months after orchidopexy.

Result(s): The 47 cryptorchid boys presented at a mean age of 3 years 2 months (range: 6 months-8 years). Out of the 50 impalpable testes, 9 were nonvisualized on preoperative US Doppler scan and 16 were vanishing on laparoscopic exploration: 5 abdominally (in utero vascular accident) and 11 scrotally (perinatal torsion). The remaining 34 testes were fixed loosely near the contralateral ASIS in the first-stage laparoscopic exploration. Out of which, 3 had slipped traction at the second stage. The 12-week traction interval went uneventfully and a predefinitive US Doppler scan confirmed viability of all testes under traction. All 34 elongated testes were mobilized and fixed in the ipsilateral hemiscrota inside a created subdartos pouch (of de Netto). All 34 fixed testes were confirmed viable on US Doppler scan 1, 3, and 6 months after orchidopexy.

Conclusion(s): The novel Shehata technique of staged laparoscopic traction-lengthening for abdominal testes is safe, easy, and convenient as evidenced by our limited early experience. Neither internal herniation complicated the traction period nor testicular atrophy (by undue tension) complicated the traction or follow-up periods. We believe it is a good alternative to the Fowler-Stephens staged orchidopexy that entails risky division of the testicular vessels.

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PMC Identifier

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Status

Embase

Institution

(Abouheba, Waheeb) Department of Pediatric Surgery, Children's Hospital, Alexandria University, El-Shatby, Alexandria 21131, Egypt (Younis, Elsokary, Roshdy) Department of Surgery, School Stud' Hospital, Health Insurance Authority, Sporting, Alexandria, Egypt

Publisher

Mary Ann Liebert Inc. (E-mail: info@liebertpub.com)

Year of Publication

2019

86.

AMH and INSL3 in testicular and extragonadal pathophysiology: what do we know?.

Sansone A., Kliesch S., Isidori A.M., Schlatt S.

Embase

Andrology. 7 (2) (pp 131-138), 2019. Date of Publication: March 2019.

[Review]

AN: 626824585

Background: It is commonly accepted that testicular function is prevalently regulated by the hypothalamic-pituitary-gonadal axis: The pulsatile secretion of GnRH by the hypothalamus induces pituitary expression of the two gonadotropins FSH and LH, which then stimulate Sertoli and Leydig cells, respectively, therefore regulating steroidogenesis and spermatogenesis.

However, a growing body of evidence has recently suggested that other hormones act on the reproductive tract since the early phases of fetal development. Anti-Mullerian hormone and INSL3 are still largely used only for research purposes despite being increasingly recognized as markers of Sertoli and Leydig cells function, respectively.

Objective(s): Provide an up-to-date review of the role of anti-Mullerian hormone and INSL3 in human pathophysiology according to current evidence.

Material(s) and Method(s): A thorough literature review was performed on PubMed, OVID MEDLINE/EMBASE and Google Scholar for papers concerning anti-Mullerian hormone and INSL3 in human males.

Result(s): INSL3 is not acutely regulated by the hypothalamic-pituitary axis but is constitutively secreted by Leydig cells, therefore representing a valid marker for their number and status. Anti-Mullerian hormone expression, on the other hand, is downregulated by androgens, therefore

occurring mostly at the early stages of testicular differentiation and before the onset of puberty. Several conditions affecting testicular development, such as male hypogonadotropic hypogonadism, and their treatment have been associated to specific pattern of INSL3 and anti-Mullerian hormone expression, proving a role for both hormones in the diagnostic and therapeutic management. Recent reports suggest a role for both anti-Mullerian hormone and INSL3 in extragonadal physiology, such as cardiovascular and bone health.

Conclusion(s): Anti-Mullerian hormone and INSL3 are markers of Sertoli and Leydig cells maturation, respectively, usually involved in the pathogenesis of disorders of sexual differentiation. However, their role in testicular pathology has only been hinted at in the last decades. Recent evidence supports an involvement of both anti-Mullerian hormone and INSL3 in extragonadal pathophysiology as well.

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Author NameID

Sansone A.; ORCID: <http://orcid.org/0000-0002-1210-2843> Schlatt S.; ORCID:

<http://orcid.org/0000-0002-7571-5313>

Isidori A.M.; ORCID: <http://orcid.org/0000-0002-9037-5417>

Institution

(Sansone, Kliesch, Schlatt) Center of Reproductive Medicine and Andrology, Department of Clinical and Surgical Andrology, Institute of Reproductive and Regenerative Biology, Munster, Germany (Sansone, Isidori) Department of Experimental Medicine, Section of Medical Pathophysiology, Food Science and Endocrinology - Sapienza University of Rome, Rome, Italy

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Year of Publication

2019

Associations between major dietary patterns and testicular function in a population-based cohort of young men: results from the Western Australian Pregnancy Cohort (Raine) Study.

Ognjenovic M., Ambrosini G.L., Malacova E., Doherty D.A., Oddy W.H., Handelsman D.J., McLachlan R., Dickinson J., Hart R.J.

Embase

Andrology. 7 (3) (pp 273-280), 2019. Date of Publication: May 2019.

[Review]

AN: 626735131

Background: Controversial speculation suggests that dietary intake may affect semen quality and testicular function, however, there are limited comprehensive studies observing dietary patterns.

Objective(s): To study associations between major dietary patterns and markers of testicular function in adulthood.

Material(s) and Method(s): Observational cross-sectional study of two hundred and ninety men with an average age of 20 years, from the Western Australian Pregnancy Cohort (Raine) Study. Usual dietary intake assessed using a semi-quantitative food frequency questionnaire at 20 years of age. Two dietary patterns previously identified using exploratory factor analysis ("Healthy" or "Western") and participants received z-scores for each dietary pattern. Primary endpoints were testicular volume, total sperm per ejaculate, morning serum testosterone concentration.

Secondary endpoints were semen sample parameters, inhibin B and sex steroids (DHT: 3alpha-diol, 3beta-diol; LH; FSH; DHEA; estradiol; estrone). Result(s): Participants were on average 20.0 +/- 0.4 years old, had a median of 2 days sexual abstinence and a body mass index of 24.1 +/- 3.9 kg/m², 13% were smokers, 52% were 'moderate' alcohol drinkers, 23% frequently used recreational drugs and 68% reported 'high' physical activity levels. Sperm concentration and DHT 3alpha-diol were negatively associated with a greater z-score for the "Western" dietary pattern (p = 0.007 and; p = 0.044, respectively), and serum estradiol concentration was positively associated with a "Western" dietary pattern (p = 0.007) after adjustment for BMI, varicocele, cryptorchidism and sexual abstinence.

Discussion(s): Despite associations between greater intake of the "Western" dietary pattern and a decreased male reproductive health markers, our lack of consistent associations of either a "Healthy" or a "Western" dietary pattern, limit clinical or biological significance in isolation.

Conclusion(s): A potential negative association of a "Western" dietary pattern with male reproductive health was detected and should be studied further in population-based studies.

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Status

Embase

Institution

(Ognjenovic, Ambrosini, Malacova) School of Population and Global Health, The University of Western Australia, Perth, WA, Australia (Ambrosini) Telethon Kids Institute, Perth, WA, Australia

(Malacova) School of Public Health, Curtin University, Perth, WA, Australia

(Doherty) Women and Infants Research Foundation, King Edward Memorial Hospital, Perth, WA, Australia

(Doherty, Dickinson, Hart) Division of Obstetrics & Gynaecology, The University of Western Australia, Perth, WA, Australia

(Oddy) Menzies Institute for Medical Research, The University of Tasmania, Hobart, TAS, Australia

(Handelsman) ANZAC Research Institute, Concord Hospital, University of Sydney, Concord, NSW, Australia

(McLachlan) Hudson Institute of Medical Research, Monash Medical Centre, Melbourne, VIC, Australia

(Hart) Fertility Specialists of Western Australia, Bethesda Hospital, Claremont, WA, Australia

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2019

88.

A comparative study of shear wave elastography in the evaluation of undescended and retractile testes in a pediatric population.

Turna O., Alis D.

Embase

Journal of Medical Ultrasonics. 46 (2) (pp 231-237), 2019. Date of Publication: 15 Apr 2019.

[Article]

AN: 625861144

Purpose: To evaluate the diagnostic value of shear wave elastography (SWE) in the evaluation of undescended and retractile testes (RT) in a pediatric population.

Method(s): We prospectively evaluated a total of 37 undescended testes (UDT), 15 RT, and 56 normal testes using SWE. The stiffness values were recorded for speed (m/s) and elasticity (kPa), and the mean stiffness values of groups were compared with each other.

Result(s): The mean stiffness values of the UDT (13.80 +/- 4.14 kPa, 2.14 +/- 0.29 m/s) were higher than the mean SWE values of the normal testes (7.44 +/- 2.11 kPa, 1.57 +/- 0.21 m/s) ($p < 0.0001$). The mean stiffness values of the RT (9.64 +/- 3.71 kPa, 1.75 +/- 0.35 m/s) exceeded those of the normal testes ($p = 0.004$ for elasticity and $p = 0.02$ for speed). The mean stiffness value of the UDT was higher than the retractile ones ($p < 0.0001$ for elasticity and speed).

Conclusion(s): The higher stiffness values of the UDT and the RT compared to normal testes are likely reflective of underlying pathological alterations; hence, we suggest that SWE might serve as a valuable adjunct for the management of UDT and RT by assessing and monitoring ultrastructural changes.

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Status

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Author NameID

Alis, Deniz; ORCID: <http://orcid.org/0000-0002-7045-1793>

Institution

(Turna, Alis) Department of Radiology, Istanbul Mehmet Akif Ersoy Thoracic and Cardiovascular Surgery Training and Research Hospital, Halkali, Istanbul, Turkey

Publisher

Springer Tokyo (E-mail: orders@springer.jp)

Year of Publication

2019

89.

Maternal smoking during pregnancy and risk of cryptorchidism: a systematic review and meta-analysis.

Yu C., Wei Y., Tang X., Liu B., Shen L., Long C., Lin T., He D., Wu S., Wei G.

Embase

European Journal of Pediatrics. 178 (3) (pp 287-297), 2019. Date of Publication: 12 Mar 2019.
[Article]

AN: 625080286

The risk factors for undescended testes in male infants and the underlying pathogenesis still remain unclear. The aim of this study is to identify the relationship between maternal smoking during pregnancy and risk of cryptorchidism. A systematic review was conducted using appropriate search terms to identify articles pertaining to maternal smoking during pregnancy and risk of cryptorchidism. Entries up to December 23, 2017 were taken into consideration, without any language or regional restriction. The crude ORs and their 95% CIs were computed by using the fixed-effect model. Twenty studies involving 111,712 infants were included in our meta-analysis. The risk of having a male infant with cryptorchidism was significantly different between mothers who smoked during pregnancy and those who did not (pooled crude OR 1.18, 95% confidence interval [CI] 1.12-1.24, $p < 0.00001$).

Conclusion(s): Our findings suggest that smoking during pregnancy increased the risk of cryptorchidism by 1.18 times. Further investigations that are well-designed, multicentric studies measuring variables, such as the number of cigarettes smoked in a day and the stage of pregnancy during which the mothers smoked, are necessary to precisely determine the relationship between maternal smoking and risk of cryptorchidism. What is Known: * Preterm and low birth weight have been definitively shown to be risk factors for cryptorchidism. * The relationship between with maternal smoking during pregnancy and risk of cryptorchidism remains controversial all the time. What is New: * Mothers who smoked during pregnancy had a 1.18 times higher risk of having a child with cryptorchidism as compared to those who did not smoke. * Evidence has been found that maternal smoking during pregnancy is a definitive risk factor for cryptorchidism.

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Embase

Author NameID

Wu, Shengde; ORCID: <http://orcid.org/0000-0002-0083-561X>

Institution

(Yu, Wei, Tang, Liu, Shen, Long, Lin, He, Wu, Wei) Department of Urology, Children's Hospital of Chongqing Medical University, Room 806, Kejiao Building (No. 6 Building), No. 136, 2nd Zhongshan Road, Yuzhong District, Chongqing City 400014, China (Yu, Wei, Tang, Wu, Wei) Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, Chongqing, China

(Liu, Lin, He, Wu, Wei) Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing, China
(Shen) China International Science and Technology Cooperation Base of Child Development and Critical Disorders, Chongqing, China
(Long) Chongqing Key Laboratory of Pediatrics Chongqing, Chongqing, China
Publisher
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Year of Publication
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90.

A tailored surgical approach to the palpable undescended testis.

Neheman A., Levitt M., Steiner Z.

Embase

Journal of Pediatric Urology. 15 (1) (pp 59.e1-59.e5), 2019. Date of Publication: February 2019.

[Article]

AN: 2001382779

Introduction: Orchiopexy for a palpable undescended testis can be approached through a traditional inguinal incision or trans-scrotally. Despite the possible advantages of the scrotal approach, including reduced postoperative pain and shorter recovery, it is not consistently advocated.

Objective(s): The objective of this study was to present the experience with a tailored approach to orchiopexy based on physical findings. Study design: This is an extended case series.

Material(s) and Method(s): The mobility of the testis as described at examination under anesthesia informs the choice of surgical approach. If a 'low' palpable testis (defined as testis that can be manipulated to the scrotum) was found, a scrotal approach was used. In cases of 'high' palpable testis (testis that cannot be manipulated to scrotum), the inguinal approach was used. Success was defined by location and size of the testis 3 months after surgery.

Result(s): A total of 259 orchiopexies were performed in 181 boys (78 bilateral). Scrotal approach was used in 125 (48%) and inguinal in 134 (52%) orchiopexies. Operative time was significantly shorter for the scrotal approach, 25 min vs. 40 min for inguinal orchiopexy ($P < 0.05$). The overall success rate was 98% with no statistical difference between the groups. Three children from the

inguinal group and two from the scrotal group required an additional procedure for persistent undescended testis. The rates of testicular atrophy and hypotrophic testis were higher in the inguinal group than the scrotal group (5/134 vs. 0/125; $P < 0.05$ and 17/134 vs. 6/126; $P < 0.05$, respectively).

Discussion(s): The substantial cohort of patients selected for trans-scrotal orchiopexy experienced success rates and rates of atrophic and hypotrophic testis comparable with those found in the published literature. Furthermore, trans-scrotal operative times were significantly lower than those of inguinal procedures, and less patients required re-operation in the trans-scrotal group. Limitations of this study include significantly higher age at operation in trans-scrotal patients and a difficulty accurately classifying hypotrophic testes. Furthermore, the higher atrophic rate in the inguinal group vs. the scrotal group likely reflects the vulnerability of a testis that is located higher and not the superiority of the scrotal approach.

Conclusion(s): This tailored approach to a palpable undescended testis appears simple, safe, and effective, providing high success rate with marginal complications. It is considered a preference in cases of low undescended testis, whereas the standard two-incision inguinal orchiopexy may better serve those with high undescended testis. [Table presented]

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Status

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Institution

(Neheman, Levitt) Department of Urology, Division of Pediatric Urology, Assaf Harofeh Medical Center, Zerifin, Affiliated to Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

(Steiner) Department of Urology, Meir Medical Center, Kefar Sava, Affiliated to Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

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2019

Late ascended testes: is non-orthotopic gubernacular insertion a confirmation of an alternative embryological etiology?.

Haid B., Silay M.S., Radford A., Rein P., Banuelos B., Oswald J., Spinoit A.-F.

Embase

Journal of Pediatric Urology. 15 (1) (pp 71.e1-71.e6), 2019. Date of Publication: February 2019.

[Article]

AN: 2001310222

Introduction: Re-ascended testes account for a proportion of all undescended testes (UDTs); one main hypothesis relating to their etiology relates to a patent processus vaginalis peritonei. The aim was to investigate gubernacular insertion points in boys with late ascended testis as a possible guide to an alternative embryological etiology.

Patients and Methods: Patients with proven ascended testes were recruited from four different pediatric urology centers between May 2016 and September 2017. All patients were evaluated regarding their gubernacular insertion during orchidopexy. The presence of accompanying patent processus vaginalis and the association between the epididymis and testis were also documented.

Result(s): Seventy-seven children (mean age = 73.1 +/- 41.2 months [range 18-176]) were enrolled into the study. A non-orthotopic gubernacular insertion point was found in 96.1% (n = 74); 34.2% (n = 26) of these were located in the groin and 63.2% (n = 48), high within the scrotum. Figure A. An open processus vaginalis peritonei was found in 35.1%. Twelve patients (15.6%) had small, dysplastic appearing testis with testis-epididymis dissociation. Boys with a higher insertion of the non-orthotopic gubernaculum (n = 48, groin) were operated earlier (mean age at surgery, 62.3 months) compared with those with a gubernacular insertion at a high scrotal site (mean age at surgery, 90.5 months; p = 0.004). Figure B.

Discussion(s): This study revealed that non-orthotopic gubernacular insertion is found in the vast majority of the ascending testis cases. Patent processus vaginalis was accompanying only 35.1% of all children and might be the cause of the ascending testis in this small subgroup of patients in line with the earlier reports [1]. In boys with ascending testes, in this population, the gubernaculum was very likely to insert non-orthotopically. In concordance with previous reports [2] and regarding the finding of an earlier age at surgery in boys with higher inserting gubernacula, this could provide a logical explanation as to how these testes are initially palpable in the scrotum and then, during body growth are retracted to the groin.

Conclusion(s): In 96.1% of the patients, a non-orthotopic gubernacular insertion was found. This points to embryologic etiology, complying well with earlier reports and further underlining the critical importance of timely diagnosis and treatment for this group of patients.[Figure presented]

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Institution

(Haid, Rein, Oswald) Department for Pediatric Urology, Hospital of the Sisters of Charity, Linz, Austria (Silay) Division of Paediatric Urology, Department of Urology, Istanbul Medeniyet University, Istanbul, Turkey

(Radford) Department of Pediatric Surgery, Leeds Children's Hospital NHS Trust, United Kingdom

(Banuelos) Department of Urology, Charite University Clinic, Berlin, Germany

(Spinoit) Department of Urology, Ghent University Hospital, Ghent, Belgium

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92.

A retrospective population-based cohort study to evaluate the impact of an older sibling with undescended testis and hypospadias on the known maternal and fetal risk factors for undescended testis and hypospadias in Ontario, Canada, 1997-2007.

Dave S., Liu K., Clark R., Garg A.X., Shariff S.Z.

Embase

Journal of Pediatric Urology. 15 (1) (pp 41.e1-41.e9), 2019. Date of Publication: February 2019.

[Article]

AN: 2001290928

Introduction: There are several reported risk factors for undescended testis (UDT) and hypospadias (HYP). Also, a family history of UDT or HYP has not been accounted for in prior studies, and doing so may influence these independent risk estimates. Study design: A population-based retrospective cohort study was conducted using linked administrative databases in Ontario, Canada, to identify all live male newborns born between 1997 and 2007, and it was determined whether they underwent an orchidopexy or HYP repair within 5 years of birth. Baseline maternal and fetal risk factors were obtained using appropriate ICD codes. A

statistical analysis using a generalized estimating equation with a logit link was performed, adjusting for clustering in mothers with a previous child born in the 5 years before the proband with UDT or HYP, to evaluate the adjusted risk factors of UDT and HYP.

Result(s): A total of 709,968 male infants were followed up from birth for 5 years, of which 5830 underwent an orchidopexy and 2722 had an HYP repair. On multivariable analysis, factors associated with a higher risk of UDT included prematurity, small for gestational age (SGA), associated HYP, gestational hypertension, use of assisted fertility techniques, increased maternal age, Cesarean section, previous sibling with UDT, and disorders of sexual differentiation (DSDs). After adjusting for clustering in mothers with a previous baby with UDT, DSD, associated HYP (odds ratio [OR], 2.0; 95% confidence interval [CI], 1.0-4.1), and a previous sibling with UDT (OR, 3.6; 95% CI, 2.5-5.2) remained significant risk factors. The risk factors on multivariable analysis predicting the risk of HYP included SGA, prematurity, higher income families, and associated anomalies such as UDT. After adjusting for clustering in mothers with a previous sibling with HYP, SGA (OR, 1.8; 95% CI, 1.0-3.1), higher income families (OR, 1.5-1.6), associated UDT (OR, 7.1; 95% CI, 4.9-10.0), and a previous sibling with HYP (OR, 12.8; 95% CI, 9.1-18.1) remained significant risk factors.

Discussion(s): Studies estimating risk factors for UDT and HYP have used variable methodologies to identify index cases and perform statistical analysis. This study suggests that having an older sibling with UDT or HYP is a significant independent risk factor. Performing an analysis adjusting for clustering in mothers with a previous child with UDT or HYP leads to loss of statistical significance for other described risk factors.

Conclusion(s): Underlying genetic or similar environmental exposures may be a key risk factor for UDT and HYP, which confounds known maternal and fetal risk factors for these anomalies.

[Table presented]

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Status

Embase

Author NameID

Dave S.; ORCID: <http://orcid.org/0000-0003-1462-647X>

Institution

(Dave, Clark) Department of Surgery and Paediatrics, Division of Urology, Western University, London, Ontario, Canada (Dave, Liu, Clark, Garg, Shariff) Institute for Clinical Evaluative Sciences, London, Ontario, Canada

(Garg, Shariff) Arthur Labatt Family School of Nursing, Western University, London, Ontario, Canada

(Garg, Shariff) Lawson Health Research Institute, London, Ontario, Canada
(Garg, Shariff) Department of Medicine, Division of Nephrology, Western University, London, Ontario, Canada
(Garg, Shariff) Department of Epidemiology and Biostatistics, Western University, London, Ontario, Canada
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Year of Publication
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93.

Delayed access to care and unmet burden of pediatric surgical disease in resource-constrained African countries.

Yousef Y., Lee A., Ayele F., Poenaru D.

Embase

Journal of Pediatric Surgery. 54 (4) (pp 845-853), 2019. Date of Publication: April 2019.

[Review]

AN: 2000948163

Background: The purpose of this study was to estimate the unmet burden of surgically correctable congenital anomalies in African low- and middle-income countries (LMICs).

Method(s): We conducted a chart review of children operated for cryptorchidism, isolated cleft lip, hypospadias, bladder exstrophy and anorectal malformation at an Ethiopian referral hospital between January 2012 and July 2016 and a scoping review of the literature describing the management of congenital anomalies in African LMICs. Procedure numbers and age at surgery were collected to estimate mean surgical delays by country and extrapolate surgical backlog. The unmet surgical need was derived from incidence-based disease estimates, established disability weights, and actual surgical volumes.

Result(s): The chart review yielded 210 procedures in 207 patients from Ethiopia. The scoping review generated 42 data sets, extracted from 36 publications, encompassing: Benin, Egypt, Ghana, Ivory Coast, Kenya, Nigeria, Madagascar, Malawi, Togo, Uganda, Zambia, and Zimbabwe. The largest national surgical backlog was noted in Nigeria for cryptorchidism (209,260

cases) and cleft lip (4154 cases), and Ethiopia for hypospadias (20,188 cases), bladder exstrophy (575 cases) and anorectal malformation (1349 cases).

Conclusion(s): These data support the need for upscaling pediatric surgical capacity in LMICs to address the significant surgical delay, surgical backlog, and unmet prevalent need.

Type of Study: Retrospective study and review article Level of evidence: III

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Embase

Institution

(Yousef, Lee, Poenaru) Division of Pediatric General and Thoracic Surgery, The Montreal Children's Hospital, McGill University Health Centre, 1001 Decarie Blvd, Room B04.2028, Montreal, H4A3J1, Quebec, Canada (Ayele) MyungSung Christian Medical Center, MyungSung Medical College, Box 15478, Addis Ababa, PO, Ethiopia

Publisher

W.B. Saunders

Year of Publication

2019

94.

Impaired serum inhibin-B and number of germ cells in boys with cryptorchidism following heavily gestational maternal smoking.

Hildorf S., Clasen-Linde E., Dong L., Cortes D., Thorup J.

Embase

Journal of Pediatric Surgery. 54 (4) (pp 809-814), 2019. Date of Publication: April 2019.

[Article]

AN: 2000708864

Purpose: A meta-analysis including 11,900 cases showed that maternal gestational smoking was associated with increased risk of cryptorchidism. The aim of study was to investigate whether a hormone profile of cryptorchid boys and a supplementing histopathological evaluation of testicular

biopsies could add detailed knowledge to the impact of maternal gestational smoking on pathogenesis of cryptorchidism.

Method(s): 601 cryptorchid boys aged 4 months to 14 years old were included. Because normal hormones have a pronounced age dependency, we compared results from boys whose mothers had smoked heavily (> 10 cigarettes/day) during pregnancy with age matched cryptorchid controls of nonsmoking mothers (1:6). We studied: birthweight, germ-cell number/tubular cross section, frequency of germ cells positive for placental-like alkaline phosphatase (PLAP), gonadotropins and inhibin-B.

Result(s): 501 boys were sons of nonsmokers, 72 boys of intermittent smokers and 28 boys of heavy smokers. 39%, 44% and 61% respectively had bilateral cryptorchidism. Compared to age-matched cryptorchid controls of nonsmoking mothers, sons of heavy smokers had lower birthweight ($p = 0.006$), germ-cell number/tubular cross section ($p = 0.009$), frequency of germ cells positive for PLAP ($p = 0.037$) and inhibin-B ($p = 0.042$).

Conclusion(s): All findings could be associated with placental dysfunction with altered human chorionic gonadotropin production well described in women smoking during pregnancy.

Type of Study: Prognosis study (prospective cohort study with > 80% follow-up).

Level of Evidence: Level 1.

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Status

Embase

Institution

(Hildorf, Thorup) The Department of Pediatric Surgery, Copenhagen University Hospital Rigshospitalet, Denmark (Clasen-Linde) Department of Pathology, Copenhagen University Hospital Rigshospitalet, Denmark

(Dong) Laboratory of Reproductive Biology, Juliane Marie Centre for Women, Children and Reproduction Rigshospitalet, Denmark

(Cortes) Section of Endocrinology, Department of Pediatrics, Copenhagen University Hospital Hvidovre, Denmark

(Cortes, Thorup) Faculty of Health and Medical Sciences, University of Copenhagen, Denmark

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95.

Histopathology of Unilateral Cryptorchidism.

Verkauskas G., Malcius D., Dasevicius D., Hadziselimovic F.

Embase

Pediatric and Developmental Pathology. 22 (1) (pp 53-58), 2019. Date of Publication: 01 Jan 2019.

[Article]

AN: 623160297

Defective mini-puberty inducing insufficient gonadotropin secretion is one of the most common causes of nonobstructive azoospermia in men suffering from congenital isolated unilateral or bilateral cryptorchidism. The aim of our study was to determine the risk for azoospermia by histologic criteria in a cohort of unilateral cryptorchid boys undergoing orchidopexy and bilateral testicular biopsy. We performed a retrospective analysis of data available in the library of the Cryptorchidism Research Institute, Liestal, Switzerland. Complete histological evaluations were available for 319 boys operated on for unilateral cryptorchidism with simultaneous biopsy of the contralateral descended testicle. The median age was 39 (5-192) months and 58 patients were <18 months of age. Forty-eight percent of undescended testis (UDT) and 21% of contralateral testis had no A dark (Ad) spermatogonia. Furthermore, in 11% of boys Ad spermatogonia were lacking in both testes. Positive correlation was found between the spermatogonia/tubule ratio of the UDT and contralateral testis (Spearman rank order correlation is 0.16, P =.003). The extent of alteration in the UDT correlated with the contralateral descended testis, indicating that unilateral cryptorchidism is a bilateral disease. Observed impaired transition from gonocytes into Ad spermatogonia indicates defective mini-puberty, providing one of explanations for azoospermia and infertility development in unilateral cryptorchid men.

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Status

Embase

Author NameID

Verkauskas, Gilyvdas; ORCID: <http://orcid.org/0000-0001-7152-0802>

Institution

(Verkauskas) Children's Surgery Centre, Vilnius University Faculty of Medicine, Vilnius, Lithuania
(Malcius) Department of Pediatric Surgery, Lithuanian University of Health Sciences, Kaunas,
Lithuania

(Dasevicius) National Centre of Pathology, Affiliate of Vilnius University Hospital Santaros
Klinikos, Vilnius, Lithuania

(Hadziselimovic) Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal,
Liestal, Switzerland

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Year of Publication

2019

96.

Oxidative DNA Damage and NOX4 Levels in Children with Undescended Testes.

Avci V., Ayengin K., Alp H.H.

Embase

European Journal of Pediatric Surgery. 29 (6) (pp 545-550), 2019. Date of Publication: 2019.

[Article]

AN: 629942176

Background Undescended testis (UDT) is a common urological disorder. Patients with UDT have a risk of malignancy and infertility. The development of these conditions may be due to oxidative stress mediated by reactive oxygen species. The aim of this study was to investigate the relationship between these parameters by detecting oxidative DNA damage (8-hydroxy 2 deoxyguanosine/10 6 deoxyguanosine), ischemia-modified albumin (IMA), malondialdehyde (MDA), and nicotinamide adenine dinucleotide phosphate oxidase 4 (NOX4) levels in children with UDT and healthy control group. Materials and Methods The blood samples were obtained from 30 patients with UDT and 40 healthy male subjects. The levels of oxidative DNA damage were detected by high-pressure liquid chromatography method. We used commercially available kits that use enzyme-linked immunosorbent assay method to measure IMA, MDA, and NOX4 levels. Results The levels of MDA, IMA, NOX4, and oxidative DNA damage in children with UDT were statistically significantly higher than control group. In addition, we found that the levels of NOX4, IMA, and oxidative DNA damage after 12 months of age was significantly higher than

before 12 months of age. Conclusion We identified increased lipid peroxidation, oxidative DNA damage, IMA, and NOX4 levels in children with UDT. Delay in the treatment of UDT may cause oxidative damage. That is why, according to us the antioxidant treatment may be beneficial in children with UDT.

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Status

Embase

Institution

(Avci, Ayengin) Department of Pediatric Surgery, Van Yuzuncu Yil University, Van, Turkey (Alp) Department of Biochemistry, Van Yuzuncu Yil University, Van, Turkey

Publisher

Georg Thieme Verlag (E-mail: iaorl@iaorl.org)

Year of Publication

2019

97.

Evaluation of gonadotropin-replacement therapy in male patients with hypogonadotropic hypogonadism.

Ortac M., Hidir M., Salabas E., Boyuk A., Bese C., Pazir Y., Kadioglu A.

Embase

Asian Journal of Andrology. 21 (6) (pp 623-627), 2019. Date of Publication: 01 Nov 2019.

[Article]

AN: 629754446

Hypogonadotropic hypogonadism (HH) is a rare disease in which medical treatment has a high success rate to achieve fertility. This study aimed to analyze the efficacy of hormone replacement therapy and determine predictive factors for successful spermatogenesis and spontaneous pregnancy in patients with idiopathic HH. A total of 112 patients with low testosterone (T), luteinizing hormone (LH) and follicle-stimulating hormone (FSH), and normal prolactin levels were diagnosed with HH and administered LH and FSH analogs as hormone replacement therapy. During treatment, 96 (85.7%) patients had sperm present in ejaculate samples. Among these

patients, 72 were married and wanted a child. Of these 72 patients, 48 (66.7%) of couples had pregnancies from natural conception. After initiation of treatment, the mean time for the appearance of sperm in semen was 9.48 months. There were no significant differences between baseline FSH, T, and LH levels; however, older age, larger testicular size, and low rate of undescended testes were favorable factors for successful spermatogenesis. Larger testicular size and older age were also the main predictive factors for natural conception. We found that patients with undescended testes had a younger age, smaller testes, and lower T levels compared with patients exhibiting descended testes. The rate of sperm found in the ejaculate was not significantly decreased in patients with undescended compared with descended testis (73.7% vs 87.6%, $P = 0.261$). The medical approach for males with HH and azoospermia provides a successful treatment modality in regard to successful spermatogenesis and achievement of pregnancy.

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PMC Identifier

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Status

Embase

Institution

(Ortac) Haseki Training and Research Hospital, Department of Urology, Istanbul 34096, Turkey

(Hidir, Boyuk, Bese, Pazir, Kadioglu) Department of Urology, Istanbul Faculty of Medicine,

Istanbul University, Istanbul 34104, Turkey

(Salabas) Department of Urology, Biruni University Hospital, Istanbul 34295, Turkey

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(E), Mumbai 400 075, India)

Year of Publication

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98.

Identification of three novel SRD5A2 mutations in Chinese patients with 5alpha-reductase 2 deficiency.

Cheng T., Wang H., Han B., Zhu H., Yao H.-J., Zhao S.-X., Zhu W.-J., Zhai H.-L., Chen F.-G., Song H.-D., Cheng K.-X., Liu Y., Qiao J.

Embase

Asian Journal of Andrology. 21 (6) (pp 577-581), 2019. Date of Publication: 01 Nov 2019.

[Article]

AN: 629754425

In this study, we investigated the genetics, clinical features, and therapeutic approach of 14 patients with 5alpha-reductase deficiency in China. Genotyping analysis was performed by direct sequencing of PCR products of the steroid 5alpha-reductase type 2 gene (SRD5A2). The 5alpha-reductase activities of three novel mutations were investigated by mutagenesis and an in vitro transfection assay. Most patients presented with a microphallus, variable degrees of hypospadias, and cryptorchidism. Eight of 14 patients (57.1%) were initially reared as females and changed their social gender from female to male after puberty. Nine mutations were identified in the 14 patients. p.G203S, p.Q6X, and p.R227Q were the most prevalent mutations. Three mutations (p.K35N, p.H162P, and p.Y136X) have not been reported previously. The nonsense mutation p.Y136X abolished enzymatic activity, whereas p.K35N and p.H162P retained partial enzymatic activity. Topical administration of dihydrotestosterone during infancy or early childhood combined with hypospadias repair surgery had good therapeutic results. In conclusion, we expand the mutation profile of SRD5A2 in the Chinese population. A rational clinical approach to this disorder requires early and accurate diagnosis, especially genetic diagnosis.

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Status

Embase

Institution

(Cheng, Wang, Han, Zhu, Zhu, Qiao) Department of Plastic Surgery, Research Center of Tissue Engineering, Shanghai Ninth People s Hospital, Shanghai Jiao Tong University, School of

Medicine, Shanghai 200011, China (Yao) Department of Urology, Shanghai Ninth People s Hospital, Shanghai Jiao Tong University, School of Medicine, Shanghai 200011, China

(Zhao, Chen) Central Laboratory, Clinical Research Center, Shanghai Ninth People s Hospital, Shanghai Jiao Tong University, School of Medicine, Shanghai 200011, China

(Zhai, Song, Cheng, Liu) Department of Plastic Surgery, Research Center of Tissue Engineering, Shanghai Ninth People s Hospital, Shanghai Jiao Tong University, School of Medicine, Shanghai 200011, China

Publisher

Wolters Kluwer Medknow Publications (B9, Kanara Business Centre, off Link Road, Ghatkopar (E), Mumbai 400 075, India)

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2019

99.

Timing of pubertal development in boys born with cryptorchidism and hypospadias: A nationwide cohort study.

Arendt L., Ernst A., Braskhoj Lauridsen L., Brix N., Olsen Jo., Ramlau-Hansen C.

Embase

Asian Journal of Andrology. 21 (6) (pp 551-556), 2019. Date of Publication: 01 Nov 2019.

[Article]

AN: 629754411

Pubertal development may be altered in boys with cryptorchidism and hypospadias, but existing knowledge is inconsistent. Therefore, we investigated the association between cryptorchidism and hypospadias and pubertal development in a large cohort study. Boys in the Puberty Cohort, a cohort nested within the Danish National Birth Cohort, were included in this study. Information on cryptorchidism and hypospadias was retrieved from the Danish National Patient Register. From 11 years until 18 years or full pubertal development, information on physical markers of pubertal development was provided biannually, including Tanner stages, axillary hair, acne, voice break, and first ejaculation. In multivariate regression models for interval censored data, the mean (95% confidence intervals [CIs]) differences in months in obtaining the pubertal markers between boys with and without the anomalies were estimated. Among 7698 boys, 196 (2.5%) had cryptorchidism and 60 (0.8%) had hypospadias. Boys with hypospadias experienced first ejaculation and voice break 7.7 (95% CI: 2.5-13.0) months and 4.5 (95% CI: 0.3-8.7) months later than boys without hypospadias. The age at attaining the Tanner stages for gonadal and pubic hair growth was also higher, though not statistically significant. Pubertal development seemed unaffected in boys with mild as well as severe cryptorchidism. In conclusion, hypospadias may be associated with delayed pubertal development, but pubertal development seems unaffected by cryptorchidism. The relation between hypospadias and later pubertal development may be due to the underlying shared in utero risk or genetic factors.

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Institution

(Arendt, Ernst, Braskhoj Lauridsen, Brix, Ramlau-Hansen) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus 8000, Denmark (Arendt) Perinatal Epidemiology Research Unit, Department of Paediatric and Adolescent Medicine, Aarhus University Hospital, Aarhus 8000, Denmark

(Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus 8000, Denmark

Publisher

Wolters Kluwer Medknow Publications (B9, Kanara Business Centre, off Link Road, Ghatkopar (E), Mumbai 400 075, India)

Year of Publication

2019

100.

Insulin-Like Peptide 3 (INSL3) Serum Concentration During Human Male Fetal Life.

Harrison S.M., Bush N.C., Wang Y., Mucher Z.R., Lorenzo A.J., Grimsby G.M., Schlomer B.J., Bullesbach E.E., Baker L.A.

Embase

Frontiers in Endocrinology. 10 (no pagination), 2019. Article Number: 596. Date of Publication: 04 Sep 2019.

[Article]

AN: 629509999

Context: Insulin-like peptide 3 (INSL3), a protein hormone produced by Leydig cells, may play a crucial role in testicular descent as male INSL3 knockout mice have bilateral cryptorchidism.

Previous studies have measured human fetal INSL3 levels in amniotic fluid only.

Objective(s): To measure INSL3 serum levels and mRNA in fetal umbilical cord blood and fetal testes, respectively.

Design(s): INSL3 concentrations were assayed on 50 µl of serum from male human fetal umbilical cord blood by a non-commercial highly sensitive and specific radioimmunoassay. For

secondary confirmation, quantitative real-time PCR was used to measure INSL3 relative mRNA expression in 7 age-matched human fetal testes.

Setting(s): UT Southwestern Medical Center, Dallas, TX and Medical University of South Carolina, Charleston, SC. Patients or other Participants: Twelve human male umbilical cord blood samples and 7 human male testes were obtained from fetuses 14-21 weeks gestation. Male sex was verified by leukocyte genomic DNA SRY PCR.

Intervention(s): None.

Main Outcome Measure(s): Human male fetal INSL3 cord blood serum concentrations and testicular relative mRNA expression.

Result(s): INSL3 serum concentrations during human male gestational weeks 15-20 were 2-4 times higher than published prepubertal male levels and were 5-100 times higher than previous reports of INSL3 concentrations obtained from amniotic fluid. Testicular fetal INSL3 mRNA relative expression was low from weeks 14-16, rose significantly weeks 17 and 18, and returned to low levels at week 21.

Conclusion(s): These findings further support the role of INSL3 in human testicular descent and could prove relevant in uncovering the pathophysiology of cryptorchidism.

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Status

Embase

Institution

(Harrison) Clinical R&D Sequencing Platform, Broad Institute, MIT and Harvard, Cambridge, MA, United States (Bush) PARC Urology, Frisco, TX, United States

(Wang) Endocrinology Division, Department of Internal Medicine, The First Affiliated Hospital of Xi'an Jiaotong University, Xi'an, China

(Mucher) Department of Urology, Memorial Hermann Health System, Houston, TX, United States

(Lorenzo) Department of Pediatric Urology, Hospital for Sick Children, Toronto, ON, Canada

(Grimsby) Phoenix Children's Hospital, Phoenix, AZ, United States

(Schlomer) Division of Pediatric Urology, Department of Urology, University of Texas Southwestern Medical Center, Dallas, TX, United States

(Bullesbach) Department of Biochemistry and Molecular Biology, Medical University of South Carolina, Charleston, SC, United States

(Baker) John W. Duckett MD, Laboratory in Pediatric Urology, Division of Pediatric Urology, Department of Urology, University of Texas Southwestern Medical Center, Dallas, TX, United States

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2019

101.

Propagation of Spermatogonial Stem Cell-Like Cells From Infant Boys.

Dong L., Kristensen S.G., Hildorf S., Gul M., Clasen-Linde E., Fedder J., Hoffmann E.R., Cortes D., Thorup J., Andersen C.Y.

Embase

Frontiers in Physiology. 10 (no pagination), 2019. Article Number: 1155. Date of Publication: 19 Sep 2019.

[Article]

AN: 629485629

Background: Gonadotoxic treatment of malignant diseases as well as some non-malignant conditions such as cryptorchidism in young boys may result in infertility and failure to father children later in life. As a fertility preserving strategy, several centers collect testicular biopsies to cryopreserve spermatogonial stem cells (SSCs) world-wide. One of the most promising therapeutic strategies is to transplant SSCs back into the seminiferous tubules to initiate endogenous spermatogenesis. However, to obtain sufficient numbers of SSC to warrant transplantation, in vitro propagation of cells is needed together with proper validation of their stem cell identity.

Material(s) and Method(s): A minute amount of testicular biopsies (between 5 mg and 10 mg) were processed by mechanical and enzymatic digestion. SSCs were enriched by differential plating method in StemPro-34 medium supplemented with several growth factors. SSC-like cell clusters (SSCLCs) were passaged five times. SSCLCs were identified by immunohistochemical and immunofluorescence staining, using protein expression patterns in testis biopsies as reference. Quantitative polymerase chain reaction analysis of SSC markers LIN-28 homolog A (LIN28A), G antigen 1 (GAGE1), promyelocytic leukemia zinc finger protein (PLZF), integrin alpha 6 (ITGA6), ubiquitin carboxy-terminal hydrolase L1 (UCHL1) and integrin beta 1 (ITGB1) were also used to validate the SSC-like cell identity.

Result(s): Proliferation of SSCLCs was achieved. The presence of SSCs in SSCLCs was confirmed by positive immunostaining of LIN28, UCHL1 and quantitative polymerase chain reaction for LIN28A, UCHL1, PLZF, ITGA6, and ITGB1, respectively.

Conclusion(s): This study has demonstrated that SSCs from infant boys possess the capacity for in vitro proliferation and advance a fertility preservation strategy for pre-pubertal boys who may otherwise lose their fertility.

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Status

Embase

Institution

(Dong, Kristensen, Gul, Andersen) Laboratory of Reproductive Biology, Copenhagen University Hospital, Copenhagen, Denmark (Hildorf, Thorup) Department of Pediatric Surgery, Copenhagen University Hospital, Copenhagen, Denmark

(Gul) Department of Urology, Aksaray University School of Medicine, Aksaray, Turkey

(Clasen-Linde) Department of Pathology, Copenhagen University Hospital, Copenhagen, Denmark

(Fedder) Centre of Andrology and Fertility Clinic, Department D, Odense University Hospital, Odense C, Denmark

(Fedder) Research Unit of Human Reproduction, Institute of Clinical Research, University of Southern Denmark, Odense, Denmark

(Hoffmann) Center for Chromosome Stability, Department of Molecular and Cellular Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Cortes) Department of Pediatrics, Copenhagen University Hospital Hvidovre, Copenhagen, Denmark

(Cortes, Thorup, Andersen) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

Publisher

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Year of Publication

2019

102.

Plasma concentration of MMP-1 and MMP-2 in boys with cryptorchidism and its lack of correlation with INSL3 and inhibin B.

Matuszczak E., Komarowska M.D., Sankiewicz A., Oldak L., Gorodkiewicz E., Debek W., Milewski R., Tylicka M., Hermanowicz A.

Embase

Scandinavian Journal of Clinical and Laboratory Investigation. 79 (6) (pp 412-418), 2019. Date of Publication: 18 Aug 2019.

[Article]

AN: 628491456

The matrix metalloproteinases are enzymes capable of remodeling of extracellular matrix, and modulate the behavior of cells. Maturation of gubernaculum and spermatogenesis demand proper equilibrium of metalloproteinases and their inhibitors. The aim of this survey was to investigate the levels of matrix metalloproteinase type 1 (MMP-1) and matrix metalloproteinase type 2 (MMP-2) in the plasma of children with unilateral cryptorchidism along with levels of Insulin-like Peptide 3 (INSL3) and inhibin B. INSL3 have a role in gubernaculum development. Inhibin B is produced by Sertoli cells, and its levels reflect the status of the testis germinative epithelium. Fifty boys with an undescended testicle, aged 1-4 years (median = 2.4 years) were enrolled into the study. Fifty boys with inguinal hernia aged 1-4 years, served as controls (median age = 2.1 years). Investigators assessed the MMP-1 and MMP-2 concentrations using Surface Plasmon Resonance Imaging. The levels of INSL-3 and inhibin B were assessed using commercial enzyme-linked immunosorbent assay ELISA. The median concentration of MMP-1 and MMP-2 in the blood plasma of patients with unilateral cryptorchidism, was nearly 2-folds higher than in controls. The great area under the ROC curve with the cut off value of 0.865 for MMP-1, and 0.819 for MMP-2, indicates the high clinical sensitivity and specificity of the test of plasma levels of MMP-1 and MMP-2 for boys with cryptorchidism. The increased plasma levels of MMP-1 and MMP-2, probably reflect the level of apoptosis of the germ cells in undescended testicles, in response to the heat stress during the period of prepubertal testis development. In the group of cryptorchid boys, we found slightly lower concentrations of INSL3, without statistical significance and without correlation with MMP-1 and MMP-2 levels. There were no significant differences in the levels of inhibin B in the group of boys with cryptorchidism and boys with inguinal hernia and it also did not correlate with MMP-1 and MMP-2 concentrations.

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Institution

(Matuszczak, Komarowska, Debek, Hermanowicz) Department of Pediatric Surgery, Medical University of Bialystok, Bialystok, Poland (Sankiewicz, Oldak, Gorodkiewicz) Department of Electrochemistry, University of Bialystok, Bialystok, Poland

(Milewski) Department of Medical Statistics, University of Bialystok, Bialystok, Poland

(Tylicka) Department of Biophysics, Medical University of Bialystok, Bialystok, Poland

Publisher

Taylor and Francis Ltd

Year of Publication

2019

103.

The present crisis in male reproductive health: an urgent need for a political, social, and research roadmap.

De Jonge C., Barratt C.L.R.

Embase

Andrology. 7 (6) (pp 762-768), 2019. Date of Publication: 01 Nov 2019.

[Article]

AN: 628375952

Background: There is a global crisis in male reproductive health. Evidence comes from globally declining sperm counts and increasing male reproductive system abnormalities, such as cryptorchidism, germ cell tumors, and onset of puberty. Male factor infertility occurs in ~40% of couples experiencing infertility. Data demonstrate an association between male infertility and overall health. Associated significant health conditions include diabetes mellitus, metabolic disorders, and cardiovascular disease. Adding to the complexity is that men typically do not seek health care unless there is acute medical need or, as in the case of the infertile couple, the male goes for a reproductive examination and semen analysis. However, 25% of the time a reproductive health examination does not occur. Couples are increasingly utilizing IVF at more advanced ages, and advanced paternal age is associated with increased risk for (i) adverse perinatal outcomes for both offspring and mother; (ii) early child mortality, cancer, and mental health issues. In addition to age, paternal lifestyle factors, such as obesity and smoking, impact not only the male fertility but also the offspring wellness.

Objective(s): The purpose of this paper was (i) to spotlight emerging and concerning data on male reproductive health, the relationship(s) between male reproductive and somatic health, and the heritable conditions father can pass to offspring, and (ii) to present a strategic roadmap with the goals of increasing (a) the awareness of men and society on the aforementioned, (b) the participation of men in healthcare seeking, and (c) advocacy to invigorate policy and funding agencies to support increased research into male reproductive biology.

Conclusion(s): The Male Reproductive Health Initiative (MRHI) is a newly established and rapidly growing global consortium of key opinion leaders in research, medicine, funding and policy agencies, and patient support groups that are moving forward the significant task of accomplishing the goals of the strategic roadmap.

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Status

Embase

Author NameID

De Jonge C.; ORCID: <http://orcid.org/0000-0002-4083-5833>

Institution

(De Jonge) Andrology Program, University of Minnesota Medical Center, Minneapolis, MN, United States (De Jonge) Department of Urology, University of Minnesota, Minneapolis, MN, United States

(Barratt) Division Systems Medicine, Reproductive Medicine, School of Medicine, University of Dundee, Dundee, United Kingdom

Publisher

Blackwell Publishing Ltd

Year of Publication

2019

104.

Narrowing down the region responsible for 1q23.3q24.1 microdeletion by identifying the smallest deletion.

Hoshina T., Seto T., Shimono T., Sakamoto H., Okuyama T., Hamazaki T., Yamamoto T.

Embase

Human Genome Variation. 6 (1) (no pagination), 2019. Article Number: 47. Date of Publication: 01 Dec 2019.

[Article]

AN: 2003362886

Interstitial deletions of 1q23.3q24.1 are rare. Here, chromosomal microarray testing identified a de novo microdeletion of arr[GRCh37]1q23.3q24.1(164816055_165696996) x 1 in a patient with moderate developmental delay, hearing loss, cryptorchidism, and other distinctive features. The clinical features were common to those previously reported in patients with overlapping deletions. The patient's deletion size was 881 kb-the smallest yet reported. This therefore narrowed down the deletion responsible for the common clinical features. The deleted region included seven genes; deletion of LMX1A, RXRG, and ALDH9A1 may have caused our patient's neurodevelopmental delay.

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Status

Embase

Author NameID

Yamamoto, Toshiyuki; ORCID: <http://orcid.org/0000-0002-9484-3505>

Institution

(Hoshina, Seto, Hamazaki) Department of Pediatrics, Osaka City University Graduate School of Medicine, Osaka, Japan (Seto) Department of Medical Genetics, Osaka City University Graduate School of Medicine, Osaka, Japan

(Shimono) Department of Radiology, Osaka City University Graduate School of Medicine, Osaka, Japan

(Sakamoto) Department of Neurosurgery, Osaka City University Graduate School of Medicine, Osaka, Japan

(Okuyama) Department of Clinical Laboratory Medicine/Center for Lysosomal Storage Diseases, National Center for Child Health and Development, Tokyo, Japan

(Yamamoto) Tokyo Women's Medical University Institute for Integrated Medical Sciences, Tokyo, Japan

(Yamamoto) Institute of Medical Genetics, Tokyo Women's Medical University, Tokyo, Japan

Publisher

Nature Publishing Group (Houndmills, Basingstoke, Hampshire RG21 6XS, United Kingdom)

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2019

105.

Prevalence of surgically correctable conditions among children in a mixed urban-rural community in Nigeria using the SOSAS survey tool: Implications for paediatric surgical capacity-building.

Ademuyiwa A.O., Odugbemi T.O., Bode C.O., Elebute O.A., Alakaloko F.M., Alabi E.O., Bankole O., Ladipo-Ajayi O., Seyi-Olajide J.O., Okusanya B., Abazie O., Ademuyiwa I.Y., Onwuka A., Tran T., Makanjuola A., Gupta S., Ots R., Harrison E.M., Poenaru D., Nwomeh B.C.

Embase

PLoS ONE. 14 (10) (no pagination), 2019. Article Number: e0223423. Date of Publication: 2019.

[Article]

AN: 2003321499

Background In many low- and middle-income countries, data on the prevalence of surgical diseases have been derived primarily from hospital-based studies, which may lead to an underestimation of disease burden within the community. Community-based prevalence studies may provide better estimates of surgical need to enable proper resource allocation and prioritization of needs. This study aims to assess the prevalence of common surgical conditions among children in a diverse rural and urban population in Nigeria. **Methods** Descriptive cross-sectional, community-based study to determine the prevalence of congenital and acquired surgical conditions among children in a diverse rural-urban area of Nigeria was conducted. Households, defined as one or more persons 'who eat from the same pot' or slept under the same roof the night before the interview, were randomized for inclusion in the study. Data was collected using an adapted and modified version of the interviewer-administered questionnaire-Surgeons Overseas Assessment of Surgical Need (SOSAS) survey tool and analysed using the REDCap web-based analytic application. **Main results** Eight-hundred-and-fifty-six households were surveyed, comprising 1,883 children. Eighty-one conditions were identified, the most common being umbilical hernias (20), inguinal hernias (13), and wound injuries to the extremities (9). The prevalence per 10,000 children was 85 for umbilical hernias (95% CI: 47, 123), and 61 for inguinal hernias (95% CI: 34, 88). The prevalence of hydroceles and undescended testes was comparable at 22 and 26 per 10,000 children, respectively. Children with surgical conditions had similar sociodemographic characteristics to healthy children in the study population. **Conclusion** The most common congenital surgical conditions in our setting were umbilical hernias, while injuries were the most common acquired conditions. From our study, it is estimated that there will be about 2.9 million children with surgically correctable conditions in the nation. This suggests an acute need for training more paediatric surgeons.

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Institution

(Ademuyiwa, Bode, Elebute, Alabi, Bankole) Department of Surgery, Faculty of Clinical Sciences, College of Medicine, University of Lagos, Lagos, Nigeria (Ademuyiwa, Bode, Elebute, Alakaloko, Ladipo-Ajayi, Seyi-Olajide, Makanjuola) Paediatric Surgery Unit, Department of Surgery, Lagos University Teaching Hospital, Idi Araba, Lagos, Nigeria

(Odugbemi) Department of Community Health and Primary Care, Faculty of Clinical Sciences, College of Medicine, University of Lagos, Lagos, Nigeria

(Alabi) Department of Orthopaedics and Trauma, Lagos University Teaching Hospital, Idi Araba, Lagos, Nigeria

(Bankole) Neurosurgery Unit, Department of Surgery, Lagos University Teaching Hospital, Idi Araba, Lagos, Nigeria

(Okusanya) Department of Obstetrics and Gynaecology, Faculty of Clinical Sciences, College of Medicine, University of Lagos, Lagos, Nigeria

(Okusanya) Department of Obstetrics and Gynaecology, Lagos University Teaching Hospital, Idi Araba, Lagos, Nigeria

(Abazie, Ademuyiwa) Department of Nursing Science, Faculty of Clinical Sciences, College of Medicine, University of Lagos, Lagos, Nigeria

(Onwuka) Centre for Surgical Outcomes Research, Centre for Innovation in Paediatric Practice, Research Institute at Nationwide Children's Hospital, Columbus, OH, United States

(Tran) SOSAS Uganda, Duke University Division of Global Neurosurgery, Durham, NC, United States

(Tran) University of Minnesota Medical School, Minneapolis, MN, United States

(Gupta) University of California San Francisco East Bay; Surgeons Overseas, San Francisco, CA, United States

(Ots, Harrison) Department of Surgery, University of Edinburgh, Edinburgh, United Kingdom

(Poenaru) McGill University Health Centre, Montreal Children's Hospital, Montreal, Canada

(Nwomeh) Nationwide Children's Hospital, Columbus, OH, United States

Publisher

Public Library of Science (E-mail: plos@plos.org)

Year of Publication

106.

Yield of modern genetic evaluation for patients with proximal hypospadias and descended gonads.

Rowe C.K., Adam M.P., Ahn J.J., Merguerian P.A., Shnorhavorian M.

Embase

Journal of Pediatric Urology. 15 (5) (pp 527.e1-527.e6), 2019. Date of Publication: October 2019.

[Article]

AN: 2002906197

Introduction and background: Although the pediatric urologic community has embraced a multidisciplinary genetic and endocrine evaluation for newborns with ambiguous genitalia, this approach has been reserved for the most severe cases of undervirilized 46,XY individuals despite growing evidence that genetic differences are found even in patients whose only genitourinary anomaly appears to be proximal hypospadias. Identifying these genetic differences is vital for counseling patients as they move through puberty to parenthood as well as parents on future pregnancies.

Objective(s): The primary objective was to evaluate genetic diagnosis in patients with proximal hypospadias. The authors hypothesized the more sensitive genetic evaluation available in the modern era will reveal a high rate of patients with proximal hypospadias and descended testicles who are found to have a genetic difference, supporting a thorough genetic evaluation in these patients. Study design: A retrospective review was performed of all patients who underwent surgical correction for proximal hypospadias at a single institution from January 1, 2010, to December 31, 2016. Those with midshaft hypospadias were excluded as were patients whose primary surgery was performed at an outside institution. Patient characteristics, including demographics, clinical presentation, genetic evaluation, and referral to a multidisciplinary difference of sex development (DSD) clinic, were collected. The chi-squared test and t-test were used for analysis.

Result(s): There were 112 patients with proximal hypospadias who met the inclusion criteria. Of these, 91 had bilaterally descended testicles, whereas 21 had one or more undescended testicles. Thirty-three percent of patients with isolated proximal hypospadias received genetic testing of some kind, with 24% seen in the multidisciplinary DSD clinic. Four patients had an

associated genetic syndrome identified, and 5 had a genetic difference of unknown clinical significance. Overall, 10% of patients with proximal hypospadias and descended testicles had an identifiable genetic difference vs 33% with associated cryptorchidism. Of these, one patient with proximal hypospadias and descended testicles had a genetic difference of known clinical significance that was likely to have been missed in the absence of an evaluation by a geneticist. Discussion and conclusion: There was a high rate of identifiable genetic differences in patients whose only genitourinary abnormality was proximal hypospadias, especially with the 1% risk of a likely missed diagnosis. These findings support the discussion of a genetic evaluation for all patients with proximal hypospadias, regardless of the testicular location.[Formula presented]

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Institution

(Rowe) Division of Pediatric Urology, Connecticut Children's Hospital, Hartford, CT 06106, United States (Adam, Ahn, Merguerian, Shnorhavorian) Division of Pediatric Urology, Seattle Children's Hospital, NE, Seattle, WA 98105, United States

Publisher

Elsevier Ltd

Year of Publication

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107.

Prune belly syndrome: Current perspectives.

Arlen A.M., Nawaf C., Kirsch A.J.

Embase

Pediatric Health, Medicine and Therapeutics. 10 (pp 75-81), 2019. Date of Publication: 2019.

[Review]

AN: 2002750848

Prune belly syndrome (PBS) is a rare but morbid congenital disease, classically defined by a triad of cardinal features that includes cryptorchidism, urinary tract dilation and laxity of the abdominal

wall musculature. Children often require numerous surgical interventions including bilateral orchidopexy as well as individually tailored urinary tract and abdominal wall reconstruction. Along with the classic features, patients with PBS often experience gastrointestinal, orthopedic, and cardiopulmonary comorbidities.

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Status

Embase

Institution

(Arlen, Nawaf) Yale University School of Medicine, Department of Urology, New Haven, CT 06520, United States (Kirsch) Emory University, Children's Healthcare of Atlanta, Atlanta, GA 30328, United States

Publisher

Dove Medical Press Ltd. (PO Box 300-008, Albany, Auckland, New Zealand)

Year of Publication

2019

108.

Bk virus associated nephropathy and severe pneumonia in a kidney transplanted adolescent with schimke immuneosseous-dysplasia.

Duzova L., Gulhan B., Topaloglu R., Ozaltin F., Bulent Cengiz A., Filiz Yetimakman A., Dogru D., Gucer S., Besbas N.

Embase

Turkish Journal of Pediatrics. 61 (1) (pp 111-116), 2019. Date of Publication: 2019.

[Article]

AN: 2002724126

Patients with juvenile onset Schimke immune-osseous-dysplasia (SIOD) have less severe symptoms and can survive in the second and third decade of life. We present an 18 year-old adolescent with juvenile onset SIOD who was diagnosed after renal transplantation and developed BK virus associated nephropathy (BKVAN) and severe pneumonia during follow-up. The patient developed nephrotic syndrome, unresponsive to immunosuppressives, at the age of 8 years. He had a history of meningitis, short stature, microcephaly, prominent ears, and bilateral cryptorchidism. A renal transplantation was performed at the age of 15 years. During follow-up,

he suffered from leucopenia, urinary tract infections, herpes labialis, and candida esophagitis. Sanger sequencing of SMARCAL1 revealed a missense mutation on exon 11 (R586W). A renal biopsy performed after a sharp increase in serum creatinine (without significant viremia) revealed BKVAN which responded to sirolimus monotherapy and cidofovir. Three months later, he suffered from productive cough and dyspnea with diffuse ground glass pulmonary infiltrates. His clinical situation deteriorated and non-invasive mechanical ventilation was started. Cidofovir (2 mg/kg) was re-started weekly for a possible BKV pneumonia with intravenous immunoglobulin. After 5 doses of cidofovir and intense antibiotic regime, his dyspnea resolved with stable graft functions. In our case; BKVAN, which developed without significant viremia, and possibly associated pneumonia were treated successfully with cidofovir and sirolimus monotherapy.

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Institution

(Duzova, Gulhan, Topaloglu, Ozaltin, Besbas) Department of Pediatrics Division of Pediatric Nephrology, Hacettepe University Faculty of Medicine, Ankara, Turkey (Ozaltin) Department of Pediatrics Nephrogenetics Laboratory, Hacettepe University Faculty of Medicine, Ankara, Turkey (Bulent Cengiz) Department of Pediatrics Division of Pediatric Infectious Diseases, Hacettepe University Faculty of Medicine, Ankara, Turkey

(Filiz Yetimakman) Department of Pediatrics Division of Pediatric Intensive Care, Hacettepe University Faculty of Medicine, Ankara, Turkey

(Dogru) Department of Pediatrics Division of Pediatric Pulmonary Medicine, Hacettepe University Faculty of Medicine, Ankara, Turkey

(Gucer) Department of Pediatrics Department of Pediatric and Perinatal Pathology Research, Hacettepe University Faculty of Medicine, Ankara, Turkey

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Turkish Journal of Pediatrics (E-mail: deryakaraduman@gmail.com)

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Gonocyte transformation in congenital undescended testes: what is the role of inhibin-B in cell death?.

Wilson V., Thorup J., Clasen-Linde E., Cortes D., Hutson J.M., Li R.

Embase

Pediatric Surgery International. 35 (11) (pp 1309-1316), 2019. Date of Publication: 01 Nov 2019.

[Article]

AN: 2002534580

Purpose: Undescended testes (UDT) are subjected to heat stress, which can disturb gonocyte transformation as well as apoptosis. This study aims to describe the apoptosis pathway occurring during minipuberty of children with unilateral (UDT), and to investigate the role of inhibin-B.

Method(s): Testicular biopsies at unilateral orchidopexy of 10 boys (6-9 months old) with normal inhibin-B (n = 5) or low inhibin-B (n = 5) were selected for immunohistochemistry and TUNEL (Terminal deoxynucleotidyl transferase dUTP nick end labelling) assay. Testicular tubules were labelled with antibodies against Anti-Mullerian hormone (AMH, Sertoli cell marker), mouse Vasa Homolog (MVH) and placental alkaline phosphatase (PLAP) (both germ cell markers), cleaved caspase3 (apoptotic marker), and followed by confocal imaging and cell counting with Fiji/ImageJ. Data were analyzed with GraphPad Prism.

Result(s): In males with low and normal inhibin-B, there was no statistical difference ($p > 0.05$) in the percentage of testicular tubules containing TUNEL + cells, number of cleaved caspase3 +/- germ cells/tubule, total number of germ cells/tubule, and the percentage of fibrotic tubules or number of Sertoli cells/tubule.

Conclusion(s): These results suggest that inhibin-B does not regulate cell death of gonocytes and further studies are required to uncover any role of inhibin-B in gonocyte transformation.

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Status

Embase

Institution

(Wilson, Hutson, Li) Douglas Stephens Surgical Research Group, Murdoch Children's Research Institute, Flemington Road, Parkville, VIC 3052, Australia (Wilson, Hutson, Li) Department of Paediatrics, University of Melbourne, Melbourne, Australia

(Thorup) Department of Paediatric Surgery, Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark

(Thorup, Cortes) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Clasen-Linde) Department of Pathology, Copenhagen University Hospital Rigshospitalet, Copenhagen, Denmark

(Cortes) Department of Pediatrics, Copenhagen University Hospital Hvidovre, Copenhagen, Denmark

(Hutson) Department of Urology, The Royal Children's Hospital, Melbourne, Australia

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110.

Occupational exposure to endocrine-disrupting chemicals and other parental risk factors in hypospadias and cryptorchidism development: a case-control study.

Estors Sastre B., Campillo Artero C., Gonzalez Ruiz Y., Fernandez Atuan R.L., Bragagnini Rodriguez P., Frontera Juan G., Gracia Romero J.

Embase

Journal of Pediatric Urology. 15 (5) (pp 520.e1-520.e8), 2019. Date of Publication: October 2019.

[Article]

AN: 2002527146

Aim of the study: Endocrine-disrupting chemicals (EDCs) are exogenous agents that are capable of altering the endocrine system functions, including the regulation of developmental processes.

The aim of this study was to investigate the association between EDC exposure and other parental factors in the etiology of hypospadias and cryptorchidism.

Method(s): A case-control study was conducted. Cases (n = 210) were infants aged between 6 months and 14 years diagnosed with hypospadias or cryptorchidism who attended the authors' hospital over a period of 18 months, and controls (n = 210) were infants within the same range of age and without any urological disorders who attended the outpatient clinic of the same hospital during the same time period. Their selection was independent of exposures. Data on parental occupational exposure to EDCs and other sociodemographic variables were collected through face-to-face interviews and systematically for both cases and controls. Crude and adjusted odds ratios (ORs) were estimated to control for confounding with their 95% confidence interval (CI) by means of logistic regressions. Specifically, three final models of a dichotomous outcome were

constructed: one for cryptorchidism, one for hypospadias, and the third considering both malformations together. The Hosmer-Lemeshow test was used to assess the goodness of fit of the models. Their discriminatory accuracy (DA) was ascertained by estimating their areas under the receiver operating characteristic curves area under the curve (AUC) along with their 95% CI. Result(s): Associations were found between advanced maternal age (OR adjusted = 1.82; 95% CI: 1.14-2.92), mother's consumption of anti-abortives (OR = 5.40; 95% CI: 1.40-38.5) and other drugs (OR = 2.02; 95% CI: 1.31-3.16) during pregnancy, maternal and paternal occupational exposure to EDCs (OR = 4.08; 95% CI: 2.03-8.96 and OR = 3.90; 95% CI: 2.41-6.48, respectively), fathers smoking (OR = 2.0; 95% CI: 1.33-2.99), and fathers with urological disorders (OR = 2.31; 95% CI: 1.15-4.90). Maternal and paternal high educational level could be protective of cryptorchidism (OR = 0.47; 95% CI: 0.28-0.76 and OR = 0.63; 95% CI: 0.42-0.93, respectively). The DA of the models for the whole sample (AUC = 0.75; 95% CI: 0.70-0.79) for cryptorchidism (AUC = 0.76; 95% CI: 0.71-0.82) and for hypospadias (AUC = 0.75; 95% CI: 0.69-0.81) was moderately high.

Conclusion(s): Advanced age, some parental occupational exposure to EDCs, some drug consumption, smoking, and the father's history of urological disorders may increase risk and predict the developments of these malformations. Studies with higher samples sizes are needed to assess associations between individual EDC occupational exposures and drugs and these malformations. [Table presented]

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Author NameID

Estors Sastre B.; ORCID: <http://orcid.org/0000-0001-9064-3496> Gonzalez Ruiz Y.; ORCID: <http://orcid.org/0000-0002-2536-3559>

Bragagnini Rodriguez P.; ORCID: <http://orcid.org/0000-0002-3053-3436>

Institution

(Estors Sastre) Department of Pediatric Surgery, Hospital Universitari Son Espases, Palma de Mallorca, Spain (Campillo Artero) Balearic Health Service, Palma de Mallorca, Center for Research in Health and Economics, Barcelona School of Management, Universitat Pompeu Fabra, Barcelona, Spain

(Gonzalez Ruiz, Fernandez Atuan, Bragagnini Rodriguez, Gracia Romero) Department of Pediatric Surgery, Hospital Universitario Miguel Servet, Zaragoza, Spain

(Frontera Juan) Clinical Trials and Methodology Support Platform-IDISBA, Clinical Pharmacology Department, Hospital Universitari Son Espases, Palma de Mallorca, Spain

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111.

HIST1H1E heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals.

Burkardt D.D., Zachariou A., Loveday C., Allen C.L., Amor D.J., Ardissonne A., Banka S., Bourgois A., Coubes C., Cytrynbaum C., Faivre L., Marion G., Horton R., Kotzot D., Lay-Son G., Lees M., Low K., Luk H.-M., Mark P., McConkie-Rosell A., McDonald M., Pappas J., Phillipe C., Shears D., Skotko B., Stewart F., Stewart H., Temple I.K., Mau-Them F.T., Verdugo R.A., Weksberg R., Zarate Y.A., Graham J.M., Tatton-Brown K.

Embase

American Journal of Medical Genetics, Part A. 179 (10) (pp 2049-2055), 2019. Date of Publication: 01 Oct 2019.

[Article]

AN: 2002413644

Histone Gene Cluster 1 Member E, HIST1H1E, encodes Histone H1.4, is one of a family of epigenetic regulator genes, acts as a linker histone protein, and is responsible for higher order chromatin structure. HIST1H1E syndrome (also known as Rahman syndrome, OMIM #617537) is a recently described intellectual disability (ID) syndrome. Since the initial description of five unrelated individuals with three different heterozygous protein-truncating variants (PTVs) in the HIST1H1E gene in 2017, we have recruited 30 patients, all with HIST1H1E PTVs that result in the same shift in frame and that cluster to a 94-base pair region in the HIST1H1E carboxy terminal domain. The identification of 30 patients with HIST1H1E variants has allowed the clarification of the HIST1H1E syndrome phenotype. Major findings include an ID and a recognizable facial appearance. ID was reported in all patients and is most frequently of moderate severity. The facial gestalt consists of a high frontal hairline and full lower cheeks in early childhood and, in later childhood and adulthood, affected individuals have a strikingly high frontal hairline, frontal bossing, and deep-set eyes. Other associated clinical features include

hypothyroidism, abnormal dentition, behavioral issues, cryptorchidism, skeletal anomalies, and cardiac anomalies. Brain magnetic resonance imaging (MRI) is frequently abnormal with a slender corpus callosum a frequent finding.

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Embase

Author NameID

Burkardt, Deepika D'Cunha; ORCID: <http://orcid.org/0000-0002-1743-7503> Tatton-Brown,

Katrina; ORCID: <http://orcid.org/0000-0001-8686-0973>

Weksberg, Rosanna; ORCID: <http://orcid.org/0000-0002-6501-4150>

Faivre, Laurence; ORCID: <http://orcid.org/0000-0001-9770-444X>

Lay-Son, Guillermo; ORCID: <http://orcid.org/0000-0001-9933-1253>

Low, Karen; ORCID: <http://orcid.org/0000-0002-4975-9363>

Luk, Ho-Ming; ORCID: <http://orcid.org/0000-0003-4066-4066>

Pappas, John; ORCID: <http://orcid.org/0000-0001-5625-2598>

Stewart, Helen; ORCID: <http://orcid.org/0000-0002-1196-3000>

Zarate, Yuri A.; ORCID: <http://orcid.org/0000-0001-8235-6200>

Institution

(Burkardt) Center for Human Genetics, University Hospitals Rainbow Babies and Children,

Department of genetics, Case Western Reserve University, Cleveland, OH, United States

(Zachariou, Loveday, Tatton-Brown) Institute of Cancer Research, London, United Kingdom

(Allen) Lowerbank Dental Practice, Leyland, United Kingdom

(Amor) Department of Paediatrics, The Royal Children's Hospital, Murdoch Children's Research

Institute, University of Melbourne, Parkville, VIC, Australia

(Ardissone) Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Lombardia, Italy

(Banka) Faculty of Biology, Medicine and Health, Division of Evolution and Genomic Sciences,

School of Biological Sciences, University of Manchester, Manchester, United Kingdom

(Banka) Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University

NHS Foundation Trust, Health Innovation Manchester, Manchester, United Kingdom

(Bourgeois) CHU Cote de Nacre, Service de Genetique, Caen, France

(Coubes) Hopital Arnaud de Villeneuve Montpellier, Montpellier, France

(Cytrynbaum, Weksberg) Division of Clinical and Metabolic Genetics, The Hospital for Sick

Children, Toronto, ON, Canada

(Faivre) Genetics Center, Hopital d'Enfants, Dijon, France

(Marion) Service de Genetique, Centre Hospitalier Universitaire de Caen Normandie, Caen, France

(Horton) University Hospital Southampton NHS Foundation Trust, Southampton, United Kingdom

(Kotzot) Division of Clinical Genetics, Department of Pediatrics, Paracelsus Medical University Salzburg, Salzburg, Austria

(Lay-Son) Division de Pediatria, Pontificia Universidad Catolica de Chile, Santiago, Chile

(Lees) Clinical Genetics Department, Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom

(Low) Clinical Genetics, St Michaels Hospital, University Hospitals Bristol, Bristol, United Kingdom

(Luk) Department of Health, Clinical Genetic Service, Hong Kong

(Mark) Spectrum Health Division of Medical Genetics, Grand Rapids, MI, United States

(McConkie-Rosell, McDonald) Division of Medical Genetics, Department of Pediatrics, Duke University Medical Genetics, Durham, NC, United States

(Pappas) Human Genetics Program, University School of Medicine, New York, NY, United States

(Phillipe) UF Innovation en Diagnostic Genomique des Maladies Rares, CHU Dijon Bourgogne, INSERM UMR1231 GAD, Dijon, France

(Shears) Clinical Genetics, Churchill Hospital, Oxford, United Kingdom

(Skotko) Division of Medical Genetics and Genomics, Department of Pediatrics, Massachusetts General Hospital, Harvard Medical School, Boston, MA, United States

(Stewart) Medical Genetics, Belfast City Hospital, Belfast, United Kingdom

(Stewart) Clinical Genetics, Oxford University Hospitals NHS Foundation Trust, Oxford Centre for Genomic Medicine, Nuffield Orthopaedic Centre, Oxford, United Kingdom

(Temple) Faculty of Medicine, Wessex Clinical Genetics Service, University Hospital Southampton, University of Southampton, Southampton, United Kingdom

(Mau-Them) UF D'innovation en Genetique Moleculaire, Plateau Technique de Biologie, Centre Hospitalier Universitaire de Dijon, FHU TRANSLAD, Dijon, France

(Verdugo) Programa de Genetica Humans, ICBM, Santiago, Chile

(Zarate) Section of Genetics and Metabolism, Arkansas Children's Hospital, Little Rock, AR, United States

(Graham) Medical Genetics, Cedars-Sinai Medical Center, Los Angeles, CA, United States

(Tatton-Brown) South West Thames Regional Genetics Service, St George's University Hospitals NHS Foundation Trust, London, United Kingdom

(Tatton-Brown) St George's University of London, London, United Kingdom

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112.

Primary gonadal failure.

Ladjouze A., Donaldson M.

Embase

Best Practice and Research: Clinical Endocrinology and Metabolism. 33 (3) (no pagination), 2019. Article Number: 101295. Date of Publication: June 2019.

[Review]

AN: 2002323955

The term primary gonadal failure encompasses not only testicular insufficiency in 46,XY males and ovarian insufficiency in 46,XX females, but also those disorders of sex development (DSD) which result in gender assignment that is at variance with the genotype and gonadal type. In boys, causes of gonadal failure include Klinefelter and other aneuploidy syndromes, bilateral cryptorchidism, testicular torsion, and forms of 46,XY DSD such as partial androgen insensitivity. Causes in girls include Turner syndrome and other aneuploidies, galactosemia, and autoimmune ovarian failure. Iatrogenic causes in both boys and girls include the late effects of childhood cancer treatment, total body irradiation prior to bone marrow transplantation, and iron overload in transfusion-dependent thalassaemia. In this paper, a brief description of the physiology of testicular and ovarian development is followed by a section on the causes and practical management of gonadal impairment in boys and girls. Protocols for pubertal induction and post-pubertal hormone replacement - intramuscular, oral and transdermal testosterone in boys; oral and transdermal oestrogen in girls - are then given. Finally, current and future strategies for assisted conception and fertility preservation are discussed.

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Status

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Institution

(Ladjouze) Faculte de Medecine d'Alger, Service de Pediatrie, Centre Hospitalo-Universitaire Bad El Oued, 1 Boulevard Said Touati, Algiers, Algeria (Donaldson) Section of Child Health, School

of Medicine, Queen Elizabeth University Hospital, Govan Road, Glasgow G51 4TF, United Kingdom

Publisher

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113.

Relationship between phenotype and genotype of 102 Chinese newborns with Prader-Willi syndrome.

Ge M.-M., Gao Y.-Y., Wu B.-B., Yan K., Qin Q., Wang H.J., Zhou W.H., Yang L.

Embase

Molecular Biology Reports. 46 (5) (pp 4717-4724), 2019. Date of Publication: 01 Oct 2019.

[Article]

AN: 2002266641

High rates of misdiagnosis and delayed intervention in neonatal PWS are leading to poor prognoses. To determine the clinical and image characteristics of newborns with Prader-Willi syndrome (PWS). A total of 102 cases of newborns definitively diagnosed with PWS at the Children's Hospital of Fudan University from 02/2014 to 12/2017 were retrospectively analyzed. We analyzed the modulated voxel-based morphology (VBM) of gray matter in PWS by T2 weighted imaging. Of 102 cases, 75 (73.5%) have paternal deletion of 15q11.2-q13, whereas 27 (26.5%) have maternal uniparental disomy (UPD). Of the 75 deletion cases, 75 (100%) week crying, 71 (94.7%) hypotonia, 70 (93.3%) poor feeding, 46 (61.3%) hypopigmentation, 43 (57.3%) male cryptorchidism, 10 (13.3%) female labia minora, 48 (64%) characteristic facial features. Of 27 UPD cases, 27 (100%) week crying and hypotonia, 25 (92.6%) hypophagia, 20 (74.1%) male cryptorchidism, 1 (3.7%) female labia minora, 19 (70.4%) characteristic facial features, 12 (44.4%) hypopigmentation. The modulated VBM analysis shows that the middle frontal gyrus, orbitofrontal cortex (middle), and inferior frontal gyrus are the most variable brain regions that determine the endo-phenotype difference between the two genotypes. Hypotonia, hypophagia, and maldevelopment of sexual organs are general characteristics of newborns with PWS in Chinese population. In UPD cases, the proportions of premature newborns, elderly parturient women and congenital malformations were higher than for paternal deletion cases. The

differences in the gray matter volume of these three regions between the two genotypes may explain the differences in maladaptive behaviors and emotions.

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Institution

(Ge, Yan, Zhou) Department of Neonates, Children's Hospital, Fudan University, 399 Wan Yuan Road, Shanghai 201102, China (Wang, Zhou) Birth Defect Laboratory, Children's Hospital of Fudan University, Shanghai, China

(Wu, Qin, Yang) Clinical Genetic Center, Children's Hospital of Fudan University, 399 Wan Yuan Road, Shanghai 201102, China

(Gao) Department of B Ultrasonography, Children's Hospital, Fudan University, Shanghai, China

Publisher

Springer Netherlands (E-mail: rbk@louisiana.edu)

Year of Publication

2019

114.

Bioinformatic identification of key genes and molecular pathways in the spermatogenic process of cryptorchidism.

Zhou Y., Zhang D., Liu B., Hu D., Shen L., Long C., Yu Y., Lin T., Liu X., He D., Wei G.

Embase

Genes and Diseases. 6 (4) (pp 431-440), 2019. Date of Publication: December 2019.

[Article]

AN: 2001817832

This study aims to determine key genes and pathways that could play important roles in the spermatogenic process of patients with cryptorchidism. The gene expression profile data of GSE25518 was obtained from the Gene Expression Omnibus (GEO) database. Microarray data were analyzed using BRB-Array Tools to identify differentially expressed genes (DEGs) between high azoospermia risk (HAZR) patients and controls. In addition, other analytical methods were

deployed, including hierarchical clustering analysis, class comparison between patients with HAZR and the normal control group, gene ontology (GO), Kyoto Encyclopedia of Genes and Genomes (KEGG) pathway enrichment analysis, and the construction of a protein-protein interaction (PPI) network. In total, 1015 upregulated genes and 1650 downregulated genes were identified. GO and KEGG analysis revealed enrichment in terms of changes in the endoplasmic reticulum cellular component and the endoplasmic reticulum protein synthetic process in the HAZR group. Furthermore, the arachidonic acid pathway and mTOR pathway were also identified as important pathways, while RICTOR and GPX8 were identified as key genes involved in the spermatogenic process of patients with cryptorchidism. In present study, we found that changes in the synthesis of endoplasmic reticulum proteins, arachidonic acid and the mTOR pathway are important in the incidence and spermatogenic process of cryptorchidism. GPX8 and RICTOR were also identified as key genes associated with cryptorchidism. Collectively, these data may provide novel clues with which to explore the precise etiology and mechanism underlying cryptorchidism and cryptorchidism-induced human infertility.

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Status

Embase

Institution

(Zhou, Zhang, Liu, Hu, Yu, Lin, Liu, He, Wei) Department of Urology, Children's Hospital of Chongqing Medical University, Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing 400014, China (Zhou, Zhang, Liu, Hu, Shen, Long, Yu, Lin, Liu, He, Wei) Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, China (Zhang, Liu, Shen, Long, Yu, Lin, He, Wei) China International Science and Technology Cooperation Base of Child Development and Critical Disorders, China (Zhou, Zhang, Shen, Long, He, Wei) Chongqing Key Laboratory of Pediatrics, China

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2019

Gubernaculum Testis and Cremasteric Vessel Preservation during Laparoscopic Orchiopexy for Intra-Abdominal Testes: Effect on Testicular Atrophy Rates.

Braga L.H., Farrokhyar F., McGrath M., Lorenzo A.J.

Embase

Journal of Urology. 201 (2) (pp 378-385), 2019. Date of Publication: 01 Feb 2019.

[Article]

AN: 628755001

Purpose:Gubernaculum sparing laparoscopic orchiopexy, which involves anatomical delivery of the testis through the internal inguinal ring, has been proposed as an alternative to conventional laparoscopic Fowler-Stephens orchiopexy, maximizing collateral blood supply and potentially decreasing atrophy rates. We compared the 2 techniques to test this hypothesis.
Materials and Methods:The primary (dependent) outcome of the study was rate of testicular atrophy, which was defined as palpation of a nubbin or inability to palpate a testis (complete atrophy) on postoperative physical examination at 3 and 12 months. Doppler ultrasound was obtained routinely to further confirm the diagnosis of testicular atrophy. Independent variables that were captured were age at surgery, type of procedure (conventional laparoscopic Fowler-Stephens orchiopexy vs gubernaculum sparing laparoscopic orchiopexy), surgical approach (single vs 2-stage), location of intra-abdominal testis (high vs low) and patency of the internal inguinal ring.
Result(s):Mean +/- SD age at surgery was 25.7 +/- 13.3 months (median 22). Laparoscopy was carried out for nonpalpable testes and revealed vanishing intra-abdominal testes in 120 cases (29%), peeping testes in 80 (19%) and intra-abdominal testes in 212 (51%), with 104 being low and 108 being high in the abdomen. A single stage procedure was performed in 44 cases (21%) and a 2-stage procedure in 168 (79%). Based on surgeon preference, conventional laparoscopic Fowler-Stephens orchiopexy was undertaken in 46 patients (22%) and gubernaculum sparing laparoscopic orchiopexy in 166 (78%). Overall testicular atrophy rate was 6.6% (14 of 212 cases). Atrophy was observed in 13 of 46 testes after conventional laparoscopic Fowler-Stephens orchiopexy and 1 of 166 following gubernaculum sparing laparoscopic orchiopexy (28.3% vs 0.6%, $p < 0.01$).

Conclusion(s):Gubernaculum sparing laparoscopic orchiopexy is a feasible alternative to conventional laparoscopic Fowler-Stephens orchiopexy. Our findings suggest that preservation of additional vascular supply to the testis (cremasteric vessels and deferential artery) may translate into improved testicular survival rates following laparoscopic orchiopexy.

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Institution

(Braga, McGrath) Division of Urology, McMaster Children's Hospital, McMaster University, 1200 Main St. West 4E19, Toronto, ON L8N3Z5, Canada (Braga) McMaster Pediatric Surgery

Research Collaborative, Toronto, ON, Canada

(Farrokhyar) Department of Surgery, Hamilton Health Sciences, Hamilton, ON, Canada

(Lorenzo) Division of Pediatric Urology, Hospital for Sick Children, Toronto, ON, Canada

(Lorenzo) Department of Surgery, University of Toronto, Toronto, ON, Canada

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

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116.

Phenotype evolution and health issues of adults with Beckwith-Wiedemann syndrome.

Gazzin A., Carli D., Sirchia F., Molinatto C., Cardaropoli S., Palumbo G., Zampino G., Ferrero G.B., Mussa A.

Embase

American Journal of Medical Genetics, Part A. 179 (9) (pp 1691-1702), 2019. Date of Publication: 2019.

[Article]

AN: 628719692

Background: Beckwith-Wiedemann syndrome (BWS) phenotype usually mitigates with age and data on adulthood are limited. Our study aims at reporting phenotype evolution and health issues in adulthood.

Method(s): 34 patients (16 males), aged 18-58 years (mean 28.5) with BWS were enrolled.

Result(s): 26 patients were molecularly confirmed, 5 tested negative, and 3 were not tested. Final tall stature was present in 44%. Four patients developed Wilms' Tumor (2, 3, 5, and 10 years, respectively); one hepatoblastoma (22 years); one acute lymphoblastic leukemia (21 years); one adrenal adenoma and testicular Sertoli cell tumor (22 and 24 years, respectively); and three benign tumors (hepatic haemangioma, uterine myoma, and mammary fibroepithelioma). Surgery for BWS-related features was required in 85%. Despite surgical correction several patients

presented morbidity and sequelae of BWS pediatric issues: pronunciation/swallow difficulties (n = 9) due to macroglossia, painful scoliosis (n = 4) consistent with lateralized overgrowth, recurrent urolithiasis (n = 4), azoospermia (n = 4) likely consequent to cryptorchidism, severe intellectual disability (n = 2) likely related to neonatal asphyxia and diabetes mellitus (n = 1) due to subtotal pancreatectomy for intractable hyperinsulinism. Four patients (two males) had healthy children (three physiologically conceived and one through assisted reproductive technology).

Conclusion(s): Adult health conditions in BWS are mostly consequent to pediatric issues, underlying the preventive role of follow-up strategies in childhood. Malignancy rate observed in early adulthood in this small cohort matches that observed in the first decade of life, cumulatively raising tumor rate in BWS to 20% during the observation period. Further studies are warranted in this direction.

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Status

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Author NameID

Gazzin, Andrea; ORCID: <http://orcid.org/0000-0002-5230-2831> Carli, Diana; ORCID:

<http://orcid.org/0000-0001-5690-6504>

Mussa, Alessandro; ORCID: <http://orcid.org/0000-0003-2795-6013>

Institution

(Gazzin, Carli, Molinatto, Cardaropoli, Ferrero, Mussa) Department of Public Health and Pediatrics, University of Torino, Torino, Italy (Sirchia) Institute for Maternal Child Health IRCCS "Burlo Garofolo", Trieste, Italy

(Palumbo) Department of Haematology, Bambino Gesù Hospital, Rome, Italy

(Zampino) Department of Woman and Child Health, Center for Rare Diseases and Birth Defects, Institute of Pediatrics, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Università Cattolica del Sacro Cuore, Rome, Italy

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117.

Can haematologic parameters be used to predict testicular viability in testicular torsion?.

He M., Zhang W., Sun N.

Embase

Andrologia. 51 (9) (no pagination), 2019. Article Number: e13357. Date of Publication: 01 Oct 2019.

[Article]

AN: 628408466

The purpose of this study was to evaluate the predictive value of haematologic parameters for testicular survival in torsion. Children with testicular torsion (TT) treated in Beijing Children's Hospital from January 2006 to December 2018 were enrolled in this study. Patient data collected in this study included age, symptom duration, preoperative preparation time, cryptorchidism testicular torsion or not, spermatic cord torsion degree, orchiectomy/orchiopexy, testicular volume 3 months after operation by ultrasound in orchiopexy patients and haematologic parameters. The orchiopexy group comprised of 54 patients with a mean age of 135.6 +/- 43.73 months, and the orchiectomy group included 58 patients with a mean age of 119.36 +/- 60.82 months. The multivariate analysis showed that symptom duration (Odds Ratio = 1.11, $p < 0.001$), spermatic cord torsion degree (Odds Ratio = 1.006, $p = 0.002$) and mean platelet volume (MPV; Odds Ratio = 3.697, $p = 0.044$) were significant predictors of orchiectomy. The cut-off value for MPV during window time for orchiectomy was 10.55 fl (10⁻⁹ L) and provided a sensitivity of 47.8% and a specificity of 92.6%. This study found that symptom duration, spermatic cord torsion degree and MPV could be indicators of testicular viability in testicular torsion. MPV can provide valuable information before operation which can guide doctors and family members of the patients to select the appropriate treatment.

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Status

Embase

Author NameID

He, Meng; ORCID: <http://orcid.org/0000-0001-6925-2925>

Institution

(He, Zhang, Sun) Urology Department, Beijing Children's Hospital, Capital Medical University, Beijing, China

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118.

Laparoscopic Percutaneous Extraperitoneal Internal Ring Closure for Pediatric Inguinal Hernia: 1,142 Cases.

Wang Y.-J., Zhang Q.-L., Chen L., Lin Y., Zhang J.-Q., Wu D.-M., Huang W.-H., Zhou C.-M.

Embase

Journal of Laparoendoscopic and Advanced Surgical Techniques. 29 (6) (pp 845-851), 2019.

Date of Publication: 01 Jun 2019.

[Article]

AN: 628040482

Purpose: The purpose of this study was to summarize the clinical experience of the laparoscopic percutaneous extraperitoneal closure of the internal ring using an epidural needle for the treatment of inguinal hernias.

Method(s): There were 1,142 children with an isolated inguinal hernia who participated in this study from January 2013 to May 2018. An epidural needle was used to treat the indirect inguinal hernia with laparoscopic assistance. Symptoms and signs were followed up at 1 week, 3 months, and every 1-2 years after the operation.

Result(s): All 1,142 children underwent laparoscopic surgery successfully. All patients were discharged 1-2 days after the operation. During the hospitalization and follow-up, there were 21 patients with complications, including 6 cases of hernia recurrence, 7 cases of poor healing of the umbilical incision, 5 cases of suture granuloma and 3 cases of groin traction pain discomfort.

None of the following complications occurred: abdominal wall vascular injury, deferent duct injury, umbilical hernia, iatrogenic cryptorchidism, testicular atrophy, hydrocele, or scrotal oedema.

Conclusion(s): Laparoscopic percutaneous extraperitoneal closure of the internal ring using an epidural needle is a safe and feasible method for the treatment of inguinal hernias in children.

This method has the advantages of less trauma, no scarring and a good cosmetic effect.

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Embase

Institution

(Wang, Zhang, Chen, Lin, Zhang, Wu, Huang, Zhou) Department of Pediatric Surgery, Fujian Provincial Maternity and Children's Hospital, Fuzhou, China

Publisher

Mary Ann Liebert Inc. (E-mail: info@liebertpub.com)

Year of Publication

2019

119.

Experimental cryptorchidism enhances testicular susceptibility to dibutyl phthalate or acrylamide in Sprague-Dawley rats.

de Souza N.P., Cardoso A.P.F., Gomide L.M.M., Lima T.R.R., Miot H.A., Martino-Andrade A.J., Arnold L.L., Pennington K.L., Cohen S.M., de Camargo J.L.V., Nascimento e Pontes M.G.

Embase

Human and Experimental Toxicology. 38 (8) (pp 899-913), 2019. Date of Publication: 01 Aug 2019.

[Article]

AN: 627340738

Cryptorchidism (CPT), the most common male congenital abnormality, is variably associated with other male reproductive tract problems. We evaluated if cryptorchid rats develop enhanced testicular susceptibility to dibutyl phthalate (DBP) or acrylamide (AA) after extended exposure. Three studies with rats were performed: (1) in utero and postnatal exposure to DBP or AA; (2) establishment of CPT and orchiopexy; and (3) in utero and postnatal exposures to DBP or AA associated with CPT/orchiopexy. Seminiferous tubules were histologically scored according to the severity of lesions: (1) Rats exposed to DBP (score 1.5) or AA (score 1.1) presented mostly preserved spermatogenesis. Some seminiferous tubules showed vacuolated germinative epithelium, germ cell apoptosis, and a Sertoli cell-only (SCO) pattern. (2) CPT (score 3.3) resulted in decreased absolute testes weights, degenerated and SCO tubules, and spermatogenesis arrest that were reversed by orchiopexy (score 1.1). (3) Exposure to DBP or AA with CPT/orchiopexy led to atrophic testes, spermatogenesis arrest, germ cell exfoliation/multinucleation, and SCO tubules (both chemicals score 2.5). Exposure to chemicals

such as DBP or AA prevented the recovery of cryptorchid testes by orchiopexy. The possible role of environmental contaminants should be considered when looking for factors that modulate human testicular disorders associated with CPT.

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Author NameID

de Souza N.P.; ORCID: <http://orcid.org/0000-0001-6052-0356>

Institution

(de Souza, Cardoso, Gomide, Lima, Miot, de Camargo, Nascimento e Pontes) Department of Pathology, Center for Evaluation of the Impact of the Environmental on Human Health (TOXICAM), Botucatu Medical School, Botucatu Campus, Sao Paulo State University (UNESP), Sao Paulo, Brazil (Martino-Andrade) Department of Physiology, Polytechnic Centre, Federal University of Parana, Curitiba, Brazil

(Arnold, Pennington, Cohen) Department of Pathology and Microbiology and the Fred and Pamela Buffett Cancer Center, University of Nebraska Medical Center, Omaha, NE, United States

(Cohen) Havlik-Wall Professor of Oncology, University of Nebraska Medical Center, Omaha, NE, United States

Publisher

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Year of Publication

2019

120.

Phenotypic severity scoring system and categorisation for prune belly syndrome: application to a pilot cohort of 50 living patients.

Wong D.G., Arevalo M.K., Passoni N.M., Iqbal N.S., Jascur T., Kern A.J., Sanchez E.J., Satyanarayan A., Gattineni J., Baker L.A.

Embase

BJU International. 123 (1) (pp 130-139), 2019. Date of Publication: January 2019.

[Article]

AN: 623964249

Objective: To design a novel system of scoring prune belly syndrome (PBS) phenotypic severity at any presenting age and apply it to a large pilot cohort.

Patients and Methods: From 2000 to 2017, patients with PBS were recruited to our prospective PBS study and medical records were cross-sectionally analysed, generating individualised RUBACE scores. We designed the pragmatic RUBACE-scoring system based on six sub-scores (R: renal, U: ureter, B: bladder/outlet, A: abdominal wall, C: cryptorchidism, E: extra-genitourinary, generating the acronym RUBACE), yielding a potential summed score of 0-31. The 'E' score was used to segregate syndromic PBS and PBS-plus variants. The cohort was scored per classic Woodard criteria and RUBACE scores compared to Woodard category.

Result(s): In all, 48 males and two females had a mean (range) RUBACE score of 13.8 (8-25) at a mean age of 7.3 years. Segregated by phenotypic categories, there were 39 isolated PBS (76%), six syndromic PBS (12%) and five PBS-plus (10%) cases. The mean RUBACE scores for Woodard categories 1, 2, and 3 were 20.5 (eight patients), 13.8 (25), and 10.6 (17), respectively (P < 0.001).

Conclusion(s): RUBACE is a practical, organ/system level, phenotyping tool designed to grade PBS severity and categorise patients into isolated PBS, syndromic PBS, and PBS-plus groups. This standardised system will facilitate genotype-phenotype correlations and future prospective multicentre studies assessing medical and surgical treatment outcomes.

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Author NameID

Wong, Daniel G.; ORCID: <http://orcid.org/0000-0002-6627-9409> Arevalo, Michelle K.; ORCID: <http://orcid.org/0000-0001-6938-7352>

Baker, Linda A.; ORCID: <http://orcid.org/0000-0001-8272-4886>

Institution

(Wong, Arevalo, Passoni, Iqbal, Jascur, Kern, Sanchez, Satyanarayan, Baker) Department of Urology, University of Texas Southwestern Medical Center, Dallas, TX, United States (Passoni, Kern, Sanchez, Satyanarayan, Baker) Pediatric Urology, Children's Health Dallas, Dallas, TX, United States

(Gattineni) Pediatric Nephrology, Children's Health Dallas, Dallas, TX, United States

Publisher

Blackwell Publishing Ltd

Year of Publication

2019

121.

Novel combined insulin-like 3 variations of a single nucleotide in cryptorchidism.

Sinopidis X., Mourelatou R., Kostopoulou E., Karvela A., Rojas-Gil A.-P., Tsekoura E., Georgiou G., Spiliotis B.E.

Embase

Journal of Pediatric Endocrinology and Metabolism. 32 (9) (pp 987-994), 2019. Date of Publication: 01 Sep 2019.

[Article]

AN: 2002873111

Insulin-like 3 hormone (INSL3) is involved in the process of testicular descent, and has been thoroughly studied in cryptorchidism. However, INSL3 allelic variations found in the human genome were heterozygous and only a few of them were found exclusively in patients with cryptorchidism. Under this perspective, we aimed to study the presence of INSL3 allelic variations in a cohort of patients with cryptorchidism and to estimate their potential consequences. Blood samples were collected from 46 male patients with non-syndromic cryptorchidism and from 43 age-matched controls. DNA extraction and polymerase chain reaction (PCR) were performed for exons 1 and 2 of the INSL3 gene in all subjects. Sequencing analysis was carried out on the PCR products. All data were grouped according to testicular location. Seven variations of a single nucleotide (SNVs) were identified both in patients with cryptorchidism and in controls: Rs2286663 (c.27G > A), rs1047233 (c.126A > G) and rs6523 (c.178A > G) at exon 1, rs74531687 (c.191-30C > T) at the intron, rs121912556 (c.305G > A) at exon 2 and rs17750642 (c.*101C > A) and rs1003887 (c.*263G > A) at the untranslated region (UTR). The allelic variants rs74531687 and rs121912556 were found for the first time in the Greek population. The novel homozygotic combination of the three allelic variants rs1047233-rs6523-rs1003887 seemed to present a stronger correlation with more severe forms of cryptorchidism. The combination of specific INSL3 SNVs rather than the existence of each one of them alone may offer a new insight into the involvement of allelic variants in phenotypic variability and severity.

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Institution

(Sinopidis) Department of Pediatric Surgery, School of Medicine, University of Patras, Rion, Patras 26504, Greece (Mourelatou, Kostopoulou, Karvela, Rojas-Gil, Tsekoura, Spiliotis)

Department of Pediatrics, Research Laboratory, Division of Pediatric Endocrinology and Diabetes, School of Medicine, University of Patras, Patras, Greece

(Georgiou) Department of Pediatric Surgery, Children's Hospital, Patras, Greece

Publisher

De Gruyter (E-mail: peter.golla@degruyter.com)

Year of Publication

2019

122.

Pubertal growth spurt in patients with bilateral anorchia after testosterone replacement therapy.

Fouatih K., Belin F., Lambert A.S., Bouligand J., Bouvattier C.

Embase

Archives de Pediatrie. 26 (6) (pp 320-323), 2019. Date of Publication: September 2019.

[Article]

AN: 2002381713

Anorchia, the absence of testes in 46,XY boys, is a very rare condition. It has been suggested that the testicular tissue disappears during pregnancy, as a result of a vascular accident associated with torsion or a genetic cause. Because pubertal growth spurt is directly influenced by androgen exposure, we decided to evaluate the pubertal height gain in nine patients with anorchia who were followed up at the pediatric endocrinology unit of Bicetre University Hospital. We retrospectively included nine patients with bilateral anorchia whose puberty had been induced by androgen replacement therapy and for whom final height measurements were available. Data were obtained from medical records. Mean gain in pubertal height was 21.7 +/- 2.3 cm, lower than the expected gain during puberty (25 cm, P < 0.005). Despite limited experience in this rare

condition, androgen replacement therapy seems to allow for good pubertal growth spurt in adolescents with anorchia. However, formal protocols for androgen therapy during puberty may need to be optimized.

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Institution

(Fouatih, Belin) Service d'endocrinologie pediatrique, Paris Sud universite, hopital Bicetre, AP-HP, 78, avenue du General-Leclerc, Le Kremlin-Bicetre 94270, France (Lambert, Bouvattier)

Service d'endocrinologie pediatrique, faculte de medecine Paris Sud, hopital Bicetre, Le Kremlin-Bicetre, France

(Bouligand) Service d'hormonologie et de biologie moleculaire, faculte de medecine Paris sud, hopital Bicetre, Le Kremlin-Bicetre, France

Publisher

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Year of Publication

2019

123.

Definition of giant inguinoscrotal hernias in infants and evaluation of reliable surgical approach in a single-center study.

Anadolulu A.I., Kafadar M.T., Gercel G.

Embase

Annals of Medicine and Surgery. 45 (pp 127-129), 2019. Date of Publication: September 2019.

[Article]

AN: 2002339950

Objective: Inguinal hernia surgery is the most common surgery performed by pediatric surgeons. Giant inguinoscrotal hernia has not been clearly defined yet. The definition of giant inguinoscrotal hernia and the reliability of the surgical procedure were investigated in this study.

Material(s) and Method(s): Sixtyfour of totally 1548 male patients who have been operated with inguinal hernia from May 2015 to January 2018 were included in the study considering the diagnosis of giant inguinoscrotal hernia. The criteria for the diagnosis of giant inguinoscrotal hernia were determined as, observing that the hernia sac was filled with intestinal loops from the inguinal region to the scrotum during the physical examination, herniation of the intestines to the scrotum again as soon as the hernia was reduced and 2 cm and above inner ring diameter. High ligation and hernioplasty to 29 (45.3%) patients and hernioplasty using Zig maneuver to 35 (54.6%) patients were performed during the study.

Result(s): Postoperative wound infection was observed in 2 patients (6.8%) with high ligation and 1 (2.8%) patient with hernioplasty with Zig maneuver. Scrotal edema was detected in all the patients, which persisted until postoperative month 1. Recurrence was seen in 6 (20.6%) of 29 patients who operated using the high ligation method while it was seen in 2 (5.7%) of other 35 patients. None of the patients had testicular atrophy and/or iatrogenic undescended testis.

Conclusion(s): Giant inguinoscrotal hernias should be defined and evaluated as a group apart from classical inguinoscrotal hernias. Recurrence and morbidity rates were lower in patients who underwent hernioplasty using Zig maneuver.

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Institution

(Anadolulu) Health Sciences University Mehmet Akif Inan Training and Research Hospital Clinic of Pediatric Surgery, Sanliurfa, Turkey (Kafadar) Health Sciences University Mehmet Akif Inan Training and Research Hospital Clinic of General Surgery, Sanliurfa, Turkey

(Gercel) Sanliurfa Training and Research Hospital Clinic of Pediatric Surgery, Sanliurfa, Turkey

Publisher

Elsevier Ltd

Year of Publication

2019

124.

Single-Site laparoscopic percutaneous extraperitoneal closure of the internal ring using an epidural needle for children with Inguinal Hernia.

Zhang J.-Q., Zhang Q.-L., Chen L., Lin Y., Wang Y.-J., Wu D.-M., Zhou C.-M.

Embase

Medical Science Monitor. 25 (pp 4469-4473), 2019. Date of Publication: 2019.

[Article]

AN: 2002292974

Background: This study evaluated the safety and effectiveness of single-site laparoscopic percutaneous extraperitoneal closure of the internal ring using an epidural needle for children with inguinal hernia. Material/Methods: We retrospectively analyzed clinical data of 542 children with inguinal hernia who underwent single-site laparoscopic percutaneous extraperitoneal closure of the internal ring using an epidural needle at our hospital from June 2014 to June 2017.

Result(s): All patients successfully underwent surgery and none were converted to conventional surgery. Abdominal vascular injury, vasectomy injury, testicular vascular injury, umbilical hernia, iatrogenic cryptorchidism, testicular atrophy, hydrocele, hernia recurrence, and scrotal edema were not reported during the perioperative period. A follow-up of these patients was performed for 1224 months. During the follow-up period, umbilical hernia, iatrogenic cryptorchidism, testicular atrophy, and hydrocele were not noted, but 3 cases of hernia recurrence were found.

Conclusion(s): The single-site laparoscopic percutaneous extraperitoneal closure of the internal ring using an epidural needle for children with inguinal hernia is safe and effective, and this procedure has the advantages of minimal trauma, no scarring, and good cosmetic effect.

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Institution

(Zhang, Zhang, Chen, Lin, Wang, Wu, Zhou) Department of Pediatric Surgery, Fujian Provincial Maternity and Children's Hospital, Fuzhou, Fujian, China

Publisher

International Scientific Information, Inc. (E-mail: iza.pranga@isl-science.com)

Year of Publication

2019

Do pediatricians routinely perform genitourinary examinations during well-child visits? A review from a large tertiary pediatric hospital.

Gerber J.A., Balasubramanian A., Jorgez C.J., Shukla M.A., Jacob J.S., Zhu H., Sheth K.R., Mittal A., Tu D.D., Koh C.J., Janzen N., Wang M.-H., Austin P.F., Gonzales E.T., Roth D.R., Seth A.

Embase

Journal of Pediatric Urology. 15 (4) (pp 374.e1-374.e5), 2019. Date of Publication: August 2019. [Article]

AN: 2002134137

Background: The male genital examination is a common source of discomfort for the patient and medical provider. Performance of male genital examination is imperative; however, as many treatable diagnoses can be made. Undescended testicles (UDTs), hernias, testicular tumors, and urethral abnormalities are all potentially concerning findings which can be discovered on routine examination.

Objective(s): The objectives of this study are to determine the rate at which general pediatricians perform routine genitourinary (GU) examinations in the pediatric population and to determine the rate at which UDT are diagnosed or documented in the patient's history. The authors hypothesize the rate of pediatric GU examination during routine well-child visits to be in line with the previously reported rates in the adult literature. Study design: Nine hundred ninety-six consecutive male well-child visits conducted by general pediatricians at the study institution were reviewed. These visits were evaluated for documentation of a detailed GU examination as well as the presence of UDT from these examinations. In addition, past medical and surgical histories were reviewed to determine if a diagnosis of UDT was noted.

Result(s): Pediatricians at the study institution documented GU examinations 99.1% of the time during male well-child visits. Only 1.1% of the cohort had a documentation of UDT at any time point. Of the 11 patients with UDT, 6 boys (54.5%) had spontaneous descent with no referral to urology, whereas 5 (45.5%) required orchidopexy.

Discussion(s): Prior reports suggest 70-75% of routine office visits include a genital examination. None of these reports reviewed the pediatric population, thus making this review novel in this respect. In addition, the results are vastly different from these prior studies as the authors demonstrated over 99% of male well-child examinations included documentation of a thorough genital examination. A limitation of the study is its retrospective nature, which creates a lack of standardization across the data set. In addition, without being physically present in the examination room, one cannot discern whether an examination is simply being documented without actual performance because of the template format of the electronic medical record (EMR). Furthermore, the study was not designed to best evaluate the true rate of UDTs;

therefore, the reported rate of 1.1% cannot be accurately associated with a particular age at diagnosis.

Conclusion(s): Pediatricians do, in fact, document GU examinations on a routine basis. This finding cannot be taken with complete certainty as verification of actual examination performance is impractical. While the data demonstrated a lower than expected rate of UDT, depending upon age at diagnosis, this could indicate that although examinations are being documented, their accuracy may be diminished because of various factors at play in the healthcare system as a whole, including improper exam performance and EMR templates. Follow-up studies are required to verify these potentially changing rates of UDT and to determine if there is discordance between documentation and performance of GU examinations.

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Author NameID

Koh C.J.; ORCID: <http://orcid.org/0000-0001-8696-5193>

Institution

(Gerber, Balasubramanian, Zhu, Sheth, Mittal, Tu, Koh, Janzen, Wang, Austin, Gonzales, Roth, Seth) Division of Pediatric Urology, Department of Surgery, Texas Children's Hospital, Houston, TX, United States (Gerber, Jorgez, Shukla, Jacob, Sheth, Mittal, Tu, Koh, Janzen, Wang, Austin, Gonzales, Roth, Seth) Scott Department of Urology, Baylor College of Medicine, Houston, TX, United States

Publisher

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Year of Publication

2019

126.

Factors associated with delay in undescended testis referral.

Jiang D.D., Acevedo A.M., Bayne A., Austin J.C., Seideman C.A.

Embase

Journal of Pediatric Urology. 15 (4) (pp 380.e1-380.e6), 2019. Date of Publication: August 2019.
[Article]

AN: 2001921713

Introduction: Undescended testis (UDT) is one of the most common congenital disorders and is associated with infertility and testicular cancer. Multiple guidelines internationally have recommended orchiopexy by 18 months. Multiple large retrospective studies published in the last decade have found persistent delay in timing of orchiopexy.

Objective(s): The aim of the study was to determine timing at which UDTs are referred at the tertiary pediatric hospital and assess factors that are associated with delay in UDT referral. Study design: Based on clinical observations and previous data, a series of clinical and socio-economic variables were constructed to design a prospective database. All patients who underwent orchiopexy for UDT from March 1, 2017, to August 31, 2018, were reviewed for demographic and clinical data. Referral appointments after 18 months were considered delayed. Factors associated with delay in UDT referral were analyzed using univariate and multivariate analysis with logistic regression.

Result(s): One hundred seventy-eight patients underwent orchiopexy for UDT. The median age was 44 months, and 64% of them had delay in referral. On univariate analysis, normal birth testicular examination, diagnosis of 'retractile testicle,' long gap without seeing pediatrician, diagnosis by a new physician, and primary language non-English were associated with delayed UDT referral. On multivariate analysis, delayed referral was associated with normal testicular examination at birth, history of 'retractile testis,' diagnosis not by the regular primary care provider, and other health or social issues that may have led to delay.

Discussion(s): This is the first prospective study analyzing timing of referral for boys with cryptorchidism. It was found that timing of treatment of UDT with orchiopexy has not improved over the last decade. Major causes in delay in referral may be due to poor of education of families and lack of routine testicular examinations by referring providers. Secondary ascent may account a significant number of delayed orchiopexy cases.

Conclusion(s): Most patients at Doernbecher had delayed referral of cryptorchidism. Factors associated with delay were determined. To improve treatment of cryptorchidism, quality-based interventions and the importance of education and routine testicular examinations need to be focused on.[Figure presented]

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Institution

(Jiang, Acevedo) Department of Urology, Oregon Health & Sciences University, Portland, OR, United States (Bayne, Austin, Seideman) Department of Pediatric Urology, Doernbecher, Portland, OR, United States

Publisher

Elsevier Ltd

Year of Publication

2019

127.

Different clinical presentations and management in complete androgen insensitivity syndrome (CAIS).

Lanciotti L., Cofini M., Leonardi A., Bertozzi M., Penta L., Esposito S.

Embase

International Journal of Environmental Research and Public Health. 16 (7) (no pagination), 2019.

Article Number: 1268. Date of Publication: 01 Apr 2019.

[Review]

AN: 2001904248

Complete androgen insensitivity syndrome (CAIS) is an X-linked recessive genetic disorder resulting from maternally inherited or de novo mutations involving the androgen receptor gene, situated in the Xq11-q12 region. The diagnosis is based on the presence of female external genitalia in a 46, XY human individual, with normally developed but undescended testes and complete unresponsiveness of target tissues to androgens. Subsequently, pelvic ultrasound or magnetic resonance imaging (MRI) could be helpful in confirming the absence of Mullerian structures, revealing the presence of a blind-ending vagina and identifying testes. CAIS management still represents a unique challenge throughout childhood and adolescence, particularly regarding timing of gonadectomy, type of hormonal therapy, and psychological concerns. Indeed this condition is associated with an increased risk of testicular germ cell tumour (TGCT), although TGCT results less frequently than in other disorders of sex development (DSD). Furthermore, the majority of detected tumoral lesions are non-invasive and with a low probability of progression into aggressive forms. Therefore, histological, epidemiological, and prognostic features of testicular cancer in CAIS allow postponing of the gonadectomy until after pubertal age in order to guarantee the initial spontaneous pubertal development and avoid the

necessity of hormonal replacement therapy (HRT) induction. However, HRT is necessary after gonadectomy in order to prevent symptoms of hypoestrogenism and to maintain secondary sexual features. This article presents differential clinical presentations and management in patients with CAIS to emphasize the continued importance of standardizing the clinical and surgical approach to this disorder.

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Institution

(Lanciotti, Cofini, Leonardi, Penta, Esposito) Pediatric Clinic, Department of Surgical and Biomedical Sciences, Università degli Studi di Perugia, Perugia 06132, Italy (Bertozzi) Pediatric Surgery, Azienda Ospedaliera Santa Maria della Misericordia, Perugia 20122, Italy

Publisher

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Year of Publication

2019

128.

Surgery for no palpable testis before the age of one year: a risk for the testis?.

Marret J.B., Ravasse P., Boullier M., Blouet M., Dolet N., Petit T., Rod J.

Embase

Journal of Pediatric Urology. 15 (4) (pp 377.e1-377.e6), 2019. Date of Publication: August 2019.

[Article]

AN: 2001848749

Introduction: Surgery for undescended testis is now commonly recommended before the age of one year. However, the risk of testicular atrophy or miss location after surgery at a young age has not been clearly evaluated.

Objective(s): The objective of this study is to evaluate the rate of testicular atrophy after surgery for non-palpable testis before the age of one year. Materials: Fifty-five patients operated between 2005 and 2014 for non-palpable testes were reviewed for clinical and ultrasound (US) evaluation.

Median follow-up after surgery was of 68.5 months (range 26-130 months). The median age at surgery was of months (5-12 months). Eight patients (14.5%) had bilateral non-palpable testis; thus, 63 testes were evaluated. At surgery, 38 (60%) testes were located in the high inguinal canal; 25 (40%), in the abdominal cavity. Orchiopexy was performed with preservation of the testicular vessels for 58 testes. Fowler-Stephens (FS) procedure was performed for 5 testes. Testicular location was clinically evaluated, and testicular volume was measured using a standard sonogram technique in our pediatric radiology department. Ratio comparing the volume of the descended testis to the spontaneously scrotal located testis was calculated in unilateral forms. Result(s): After surgery, testes had scrotal location in 62 cases and inguinal location in one case. Seven cases of atrophy were confirmed after US control (11%), more frequently (odds ratio, OR 11.68 [1.9-72.5]) in abdominal testis (24%) than in inguinal testis (2.6%). Atrophy testicular was more frequent with FS technique (OR 7.1 [1.3-40.1]), but the population was weak (N = 5). Median volume ratio for unilateral form was 0.88 [0-1.8]; 14 patients presented a ratio greater than 1.

Discussion(s): The influence of the young age at surgery and the risk of post operative testicular atrophy had not been clearly evaluated. The term of 'no palpable testis' supports an heterogeneous group mixing abdominal and extra-abdominal testis sharing a uniform clinical presentation. Our rate of atrophy in the group of abdominal testes (24%) and inguinal testes (2.6%) is similar to the literature, which concerns older patients. The long-term sonogram assessment demonstrated a good development of the testis after surgery, especially in inguinal cases.

Conclusion(s): Surgery for no palpable testis before the age of one year does not lead to a superior risk of testicular atrophy compared with surgery at an older age and allows a good development of the testis. [Table presented]

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Institution

(Marret, Ravasse, Boullier, Dolet, Petit, Rod) Department of Pediatric Surgery, University Hospital of Caen, Avenue de la Cote de Nacre, Caen cedex 9 14 032, France (Marret, Ravasse, Rod) Caen University Hospital, Avenue de la Cote de Nacre, Caen 14 000, France

(Blouet) Department of Pediatric Radiology, University Hospital of Caen, Avenue de la Cote de Nacre, Caen cedex 9 14 032, France

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129.

Congenital hypogonadotrophic hypogonadism: Minipuberty and the case for neonatal diagnosis.

Swee D.S., Quinton R.

Embase

Frontiers in Endocrinology. 10 (FEB) (no pagination), 2019. Article Number: 97. Date of

Publication: 2019.

[Review]

AN: 628500953

Congenital hypogonadotrophic hypogonadism (CHH) is a rare but important etiology of pubertal failure and infertility, resulting from impaired gonadotrophin-releasing hormone secretion or action. Despite the availability of effective hormonal therapies, the majority of men with CHH experience unsatisfactory outcomes, including chronic psychosocial and reproductive sequelae. Early detection and timely interventions are crucial to address the gaps in medical care and improve the outlook for these patients. In this paper, we review the clinical implications of missing minipuberty in CHH and therapeutic strategies that can modify the course of disease, as well as explore a targeted approach to identifying affected male infants by integrating clinical and biochemical data in the early postnatal months.

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Status

Embase

Institution

(Swee, Quinton) Department of Endocrinology, Royal Victoria Infirmary, Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne, United Kingdom (Swee) Department of Endocrinology, Singapore General Hospital, Singapore, Singapore

(Quinton) Institute of Genetic Medicine, University of Newcastle upon Tyne, Newcastle upon Tyne, United Kingdom

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130.

Klinefelter Syndrome: Clinical Spectrum Based on 44 Consecutive Cases from a South Indian Tertiary Care Center.

Asirvatham A.R., Pavithran P.V., Pankaj A., Bhavani N., Menon U., Menon A., Abraham N., Nair V., Kumar H., Thampi M.V.

Embase

Indian Journal of Endocrinology and Metabolism. 23 (2) (pp 263-266), 2019. Date of Publication: 01 Mar 2019.

[Article]

AN: 627987202

Background: Klinefelter syndrome (KFS) is the commonest chromosomal abnormality, yet remains largely underdiagnosed due to its varied clinical presentation. This study was done to understand the clinical spectrum in our population.

Aim(s): We intended to study the clinical characteristics of children and adults with KFS in our population. We also desired to identify any special features of Klinefelter variants.

Method(s): Forty-four patients with karyotype diagnosis of KFS during the time period 2007-2015 were included in this retrospective study. Clinical details and hormonal profile were obtained from hospital information system.

Result(s): Our study population consisted of 17 (38.6%) participants in pediatric age group (age <18 years) and 27 (61.4%) adults. Clinical presentation prompting evaluation in the former group included cardiac anomalies (29.4%), dysmorphism (23.5%), hypogonadism (17.6%), developmental delay (11.8%), tall stature (11.8%), and cryptorchidism (5.9%). Among adults, 16 (59.2%) presented with hypogonadism and 9 (20.4%) had primary infertility. Six children (35.3%) had micropenis and four (three children, one adult) had unilateral undescended testis. Behavioral problems were detected in 19 (43.2%) subjects. Mean follicle stimulating hormone (FSH) and

luteinizing hormone (LH) values were 38 IU/mL and 18 IU/mL, respectively. The classical 47 XXY karyotype was detected in 38 (86.4%) subjects and 6 (13.6%) had karyotype consistent with Klinefelter variants.

Conclusion(s): KFS was diagnosed only after 18 years of age in two-thirds of patients.

Developmental delay, cardiac anomalies, behavioral abnormalities, and intellectual disabilities were the common presentations in pediatric subjects. Adults predominantly presented with hypogonadism. Individuals with Klinefelter variant karyotype sought medical attention predominantly for non-gonadal concerns.

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Institution

(Asirvatham) Department of Endocrinology, Diabetes and Metabolism, Sri Ramachandra Medical College and Research Institute, Chennai, Tamil Nadu, India (Pavithran, Pankaj, Bhavani, Menon, Menon, Abraham, Nair, Kumar) Department of Endocrinology and Podiatry, Amrita Institute of Medical Sciences, Kochi, Kerala 682 041, India

(Thampi) Department of Cytogenetics, Amrita Institute of Medical Sciences, Kochi, Kerala, India

Publisher

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131.

Frequency of inguinal herniotomy in Australia (1998-2017).

Vikraman J., Donath S., Hutson J.M.

Embase

Pediatric Surgery International. 35 (7) (pp 759-763), 2019. Date of Publication: 08 Jul 2019.

[Article]

AN: 627826295

Background/aim: Closure of the processus vaginalis (PV) is considered as the last step of testicular descent. Therefore, patent processus vaginalis (PV), and inguinal hernias are linked to cryptorchidism. As the National Australian incidence of orchidopexy has decreased over the previous 20 years, we aimed to explore the incidence of inguinal herniotomy (including hydrocele) over time in Australia.

Method(s): The National Department of Human Services (DHS) database, and Bureau of Statistics database were obtained for the years 1998-2017. The numbers of inguinal herniotomies in patients aged 0-4, 5-14 and 15-24 years were examined with ethical approval.

Result(s): Over the 20-year period, over 87,000 inguinal herniotomy procedures were performed in males. The incidence per year in males decreased across all ages over the 20-year period, but was most pronounced in infants and toddlers. Similar to males, the incidence in females decreased over time, with the ratio of procedures per head of population decreasing in children under 5 years of age. The ratio of male: females varied according to ages, and was between 2.8 and 6.2 males: 1 female.

Conclusion(s): This study suggests that fewer 0-4-year olds are undergoing inguinal herniotomy, compared with 20 years ago. This is likely due to a change in practice for the management of unilateral symptomatic hernias, from routine bilateral herniotomies, to unilateral surgery. As well as less aggressive surgical intervention for hydroceles in boys.

Level of Evidence: III.

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Status

Embase

Institution

(Vikraman, Hutson) Douglas Stephens Surgical Research Laboratory, Murdoch Childrens Research Institute, Royal Childrens Hospital, Flemington Road, Parkville, VIC 3052, Australia

(Vikraman, Hutson) Department of Urology, Royal Children's Hospital, Melbourne, Australia

(Vikraman, Hutson) Department of Paediatrics, University of Melbourne, Melbourne, Australia

(Donath) Clinical Epidemiology and Biostatistics, Murdoch Childrens Research Institute, Melbourne, Australia

Publisher

Springer Verlag (E-mail: service@springer.de)

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132.

Incidence of disorders of sexual development in neonates in Ghana: Prospective study.

Ameyaw E., Asafo-Agyei S.B., Hughes I.A., Zacharin M., Chanoine J.-P.

Embase

Archives of Disease in Childhood. 104 (7) (pp 636-638), 2019. Date of Publication: 01 Jul 2019.

[Article]

AN: 627780196

Objective The incidence of disorders of sexual development (DSD) is unknown in sub-Saharan Africa. We describe the characteristics and incidence of DSD in a cohort of infants born in Ghana. **Design** Trained research assistants performed systematic genital examination at birth. All infants with suspected abnormal genitalia were further examined by a paediatric endocrinologist. **Setting** Komfo Anokye Teaching Hospital, Kumasi, Ghana. **Patients** Consecutive infants born in a single centre over a 1-year period (May 2014 to April 2015). **Main outcome measures** Incidence of DSD. **Micropenis** was defined as a stretched length <2.1 cm and **clitoromegaly** as a clitoral length >8.6 mm. **Results** We examined 9255 infants (93% of all live births) within 72 hours of birth. Twenty-six neonates had a DSD. Nineteen infants had DSD without genital ambiguity: isolated micropenis (n=2), hypospadias (n=7), cryptorchidism (n=4) and clitoromegaly (n=6). Seven infants had DSD with ambiguity: clitoromegaly with a uterus on ultrasound and elevated 17-hydroxyprogesterone, suggesting XX DSD due to congenital adrenal hyperplasia (CAH)(n=4) and micropenis, hypospadias and gonads in a bifid scrotum or in the inguinal region, consistent with XY DSD (n=3). **Conclusion** The incidence of atypical genitalia was 28/10,000 (95% CI 17/10 000 to 39/10 000) live births. The incidence of CAH was 4.3/10 000 (95% CI 1.2/10 000 to 11.1/10 000) and was strongly associated with consanguinity.

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Status

Embase

Institution

(Ameyaw, Asafo-Agyei) Department of Child Health, Komfo Anokye Teaching Hospital, Kumasi, Ghana (Hughes) Department of Paediatrics, University of Cambridge, Cambridge, United Kingdom

(Zacharin) Pediatrics, Royal Children's Hospital Melbourne, Parkville, VIC, Australia

(Chanoine) Department of Pediatrics, University of British Columbia, Vancouver, BC, Canada

Publisher

BMJ Publishing Group (E-mail: subscriptions@bmjgroup.com)

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2019

133.

Review of the phenotypic spectrum associated with haploinsufficiency of MYRF.

Rossetti L.Z., Grinton K., Yuan B., Liu P., Pillai N., Mizerik E., Magoulas P., Rosenfeld J.A., Karaviti L., Sutton V.R., Lalani S.R., Scott D.A.

Embase

American Journal of Medical Genetics, Part A. 179 (7) (pp 1376-1382), 2019. Date of Publication: July 2019.

[Article]

AN: 627705707

The myelin regulatory factor gene (MYRF) encodes a transcription factor that is widely expressed. There is increasing evidence that heterozygous loss-of-function variants in MYRF can lead to abnormal development of the heart, genitourinary tract, diaphragm, and lungs. Here, we searched a clinical database containing the results of 12,000 exome sequencing studies. We identified three previously unreported males with putatively deleterious variants in MYRF: one with a point mutation predicted to affect splicing and two with frameshift variants. In all cases where parental DNA was available, these variants were found to have arisen de novo. The phenotypes identified in these subjects included a variety of congenital heart defects (CHD) (hypoplastic left heart syndrome, scimitar syndrome, septal defects, and valvular anomalies), genitourinary anomalies (ambiguous genitalia, hypospadias, and cryptorchidism), congenital diaphragmatic hernia, and pulmonary hypoplasia. The phenotypes seen in our subjects overlap those described in individuals diagnosed with PAGOD syndrome [MIM# 202660], a clinically defined syndrome characterized by pulmonary artery and lung hypoplasia, agonadism, omphalocele, and diaphragmatic defects that can also be associated with hypoplastic left heart and scimitar syndrome. These cases provide additional evidence that haploinsufficiency of MYRF causes a genetic syndrome whose cardinal features include CHD, urogenital anomalies, congenital diaphragmatic hernia, and pulmonary hypoplasia. We also conclude that consideration

should be given to screening individuals with PAGOD for pathogenic variants in MYRF, and that individuals with MYRF deficiency who survive the neonatal period should be monitored closely for developmental delay and intellectual disability.

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Status

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Author NameID

Rossetti, Linda Z.; ORCID: <http://orcid.org/0000-0002-8953-3246> Pillai, Nishitha; ORCID:

<http://orcid.org/0000-0002-2943-2136>

Rosenfeld, Jill A.; ORCID: <http://orcid.org/0000-0001-5664-7987>

Scott, Daryl A.; ORCID: <http://orcid.org/0000-0003-1460-5169>

Institution

(Rossetti, Ginton, Yuan, Liu, Pillai, Mizerik, Magoulas, Rosenfeld, Sutton, Lalani, Scott)

Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, United States (Yuan, Liu) Baylor Genetics, Houston, TX, United States

(Karaviti) Division of Pediatric Endocrinology, Baylor College of Medicine, Houston, TX, United States

(Scott) Department of Molecular Physiology and Biophysics, Baylor College of Medicine, Houston, TX, United States

Publisher

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Year of Publication

2019

134.

Testicular cancer in men with undescended testis: Insights from the Thames Valley Testicular Cancer database.

Bryant R.J., Hobbs C., Richardson C., Fox S., Joseph J., Verrill C., Woodcock V.K., Sullivan M.E., Protheroe A.S.

Embase

Journal of Clinical Urology. 12 (4) (pp 274-279), 2019. Date of Publication: 01 Jul 2019.

[Article]

AN: 626131621

Objective: Undescended testis (UDT) increases the risk of testicular cancer (TCa) development. Historical evidence suggests that malignant transformation of uncorrected UDT primarily results in seminomas, whereas mixed germ cell tumours predominate in corrected UDT; however, the risk of malignancy in the 'normal' contralateral testis is unclear. We investigated the contemporary Oxford TCa cohort to report the frequency of prior UDT and types of tumours developing in the prior UDT and normal contralateral testis.

Patients and Methods: A 607 patient contemporary TCa cohort within the Thames Valley Testicular Cancer database.

Result(s): Of men with new TCa, 8% had a history of UDT. Of men with TCa and prior UDT, 61% developed seminomas, whereas 56% of men with TCa without previous UDT developed this subtype. Among men with prior UDT, 77% developed tumours in the UDT, whilst 23% developed TCa in the contralateral normal testis.

Conclusion(s): Seminoma was the most frequent malignancy following UDT, with a greater frequency than without prior UDT. Around one in four TCa patients with UDT developed contralateral tumours, emphasising the need for self-examination of both testes. Advice should be given to any patient with a history of UDT stressing the importance of ongoing self-examination of both testes.

Level of Evidence: Level 4.

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Status

Embase

Author NameID

Bryant, Richard J; ORCID: <http://orcid.org/0000-0002-8330-9251> Protheroe, Andrew S; ORCID: <http://orcid.org/0000-0001-9413-0575>

Institution

(Bryant, Hobbs, Sullivan) Department of Urology, Churchill Hospital, United Kingdom (Bryant, Verrill) Nuffield Department of Surgical Sciences, University of Oxford, United Kingdom (Richardson, Fox, Joseph, Woodcock, Protheroe) Department of Oncology, University of Oxford, United Kingdom

(Verrill) Department of Cellular Pathology, John Radcliffe Hospital, United Kingdom

(Verrill) Oxford NIHR Biomedical Research Centre, University of Oxford, United Kingdom

Publisher

SAGE Publications Ltd (E-mail: info@sagepub.co.uk)

Year of Publication

135.

Orchidopexy for bilateral undescended testes: A multicentre study on its effects on fertility and comparison of two fixation techniques.

Uijldert M., Meissner A., Kuijper C.F., Repping S., de Jong T.P.V.M., Chrzan R.J.

Embase

Andrologia. 51 (3) (no pagination), 2019. Article Number: e13194. Date of Publication: April 2019.

[Article]

AN: 624810277

To evaluate fertility potential after orchidopexy for bilateral undescended testis and compare two surgical fixation techniques for effect on fertility. Men older than 22 years who had either tunica albuginea orchidopexy (TAO) or "no-touch" technique (NTO) in childhood for bilateral undescended testis (BUDT) were selected. Participants filled out a questionnaire followed by physical examination, had testicular ultrasound, blood sample and semen analysis. Statistical testing was performed using general linear modelling. Sixty-seven out of 166 individuals responded. Forty-nine completed the questionnaire, and nine (18.3%) reported having fathered children. Thirty-six showed up for further examination, 26 had TAO and 10 NTO. Impaired hormonal spermatogenesis regulation (34.6% vs. 20%), higher subfertility rate (46% vs. 20%) and lower means of motile spermatozoa (58.1×10^6 spz vs. 177.9×10^6 spz) were observed in the TAO versus the NTO group; none of these were statistically significant. Four (15.4%) of the TAO and two (20%) of the NTO group have azoospermia. Although the operation technique did not have a significant impact on fertility, unfavourable outcomes were more common after surgery involving the tunica albuginea of the testis. Larger sample sizes are needed to ascertain whether the trends favouring the NTO technique are of any significance.

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Author NameID

Uijldert, Mick; ORCID: <http://orcid.org/0000-0001-9067-4938> Chrzan, Rafal J.; ORCID: <http://orcid.org/0000-0001-8620-1898>

Institution

(Uijldert, Meisner) Department of Urology, Academic Medical Center, Amsterdam, Netherlands

(Uijldert, Meisner, Repping) Department of Obstetrics and Gynaecology, Center for Reproductive Medicine, Academic Medical Center, Amsterdam, Netherlands

(Kuijper, de Jong, Chrzan) Department of Paediatric Urology, Emma Children's Hospital, Academic Medical Center, Amsterdam, Netherlands

(Kuijper, de Jong) Department of Paediatric Urology, University Medical Center, Utrecht, Netherlands

(Chrzan) Department of Paediatric Urology, Jagiellonian University, Krakow, Poland

Publisher

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136.

A practical guide to cryptorchidism for the primary care physician.

Lovin J.M., Khater N., Mata J.A.

Embase

Family Medicine and Primary Care Review. 21 (1) (pp 78-82), 2019. Date of Publication: 2019.

[Review]

AN: 2001956948

Cryptorchidism, also known as undescended testis, affects 3-5% of full term male infants at birth and 23% of preterm or low birth weight infants. Current guidelines recommend that all boys with cryptorchidism without testicular descent by 6 months of age be referred to an appropriate specialist for evaluation, with surgery preferably performed by 18 months. We aim to examine areas of controversy still being debated by reviewing published articles, in order to provide primary providers with a practical guide to diagnosis and management of the undescended testis. Our review examined published articles from 2000 to 2018, related to undescended testes, and their management. 32 articles were reviewed from 2000 to 2018 and showed with a high level of evidence that failure of testicular descent by 6 months of age (gestational) should prompt referral

to an appropriate specialist. Physical exam is crucial. The main concerns for patients with cryptorchidism are the increased risk of testicular cancer and sub fertility. Routine use of scrotal ultrasound is not recommended in the evaluation of cryptorchidism. Diagnostic laparoscopy serves to confirm the presence of an intra-abdominal testicle. Surgical intervention within 18 months of age is imperative. Cryptorchidism should be corrected surgically between 6 months and 18 months of age. Early detection with diagnostic laparoscopy is the standard of care for treatment of palpable, undescended testis.

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Status

Embase

Author NameID

Khater, Nazih; ORCID: <http://orcid.org/0000-0002-6846-0009> Mata, John A.; ORCID: <http://orcid.org/0000-0003-0511-0965>

Institution

(Lovin, Khater, Mata) Department of Urology, Louisiana State University Health - Shreveport, Shreveport, LA, United States

Publisher

Polish Society of Family Medicine

Year of Publication

2019

137.

Spinal anesthesia in children: most pediatric urologists are not on board.

Rehfuss A., Bogaert G., Kogan B.A.

Embase

Journal of Pediatric Urology. 15 (3) (pp 263.e1-263.e5), 2019. Date of Publication: May 2019.

[Article]

AN: 2001741500

Objective: In 2016, the Food and Drug Administration issued a warning on general anesthetic medications used for lengthy procedures (>3 h) in children younger than 3 years. Spinal anesthesia can be a safe alternative to general anesthesia for many pediatric urology procedures. It can shorten total operating room (OR) time, provide excellent pain control, and allow parents to

reunite with their child immediately after surgery. However, use of spinal anesthesia can also directly affect the operating surgeon (awake patient, time constraints of spinal, and prolonged preoperative time). Members of the Societies for Pediatric Urology (SPU) and European Society of Pediatric Urology (ESPU) were surveyed to get their opinions on the use of spinal anesthesia for routine pediatric urology procedures. It was hypothesized that half of pediatric urologists would favor spinal anesthesia and that SPU members would be more likely to favor spinal anesthesia than their European colleagues.

Material(s) and Method(s): A short survey with five clinical scenarios was created. Scenarios assessed physicians' recommendations regarding timing and the type of anesthesia (general or spinal) for common pediatric urology procedures: undescended testicle, inguinal hernia, hypospadias, phimosis, and phimosis with penoscrotal webbing. Surveys were emailed to members of the SPU and ESPU. Responses and demographic information were collected and analyzed.

Result(s): The survey was completed by 113 SPU members (46% response rate for members who opened the invitation) and 109 ESPU members. For all clinical scenarios, < 20% of pediatric urologists from the SPU and <25% from the ESPU favor doing any procedure with spinal anesthesia. The majority of respondents practice in children's hospitals with pediatric anesthesiologists, but roughly half of the responders (54% SPU and 43% ESPU) do not think their anesthesia colleagues would be comfortable performing spinal anesthesia. Furthermore, only 51% of SPU and 36% of ESPU members discuss the possible neurodevelopmental side-effects of anesthesia with parents; similarly, less than half of all respondents think their anesthesia colleagues address these potential side-effects when obtaining consent. The only significant difference between SPU and ESPU responses was that ESPU members tended to delay penile surgery more than SPU respondents.

Conclusion(s): Whether general anesthesia has any effect on the developing brain of children undergoing routine pediatric urology procedures is unclear. Yet, few pediatric urologists, independent of their region of practice, prefer spinal to general anesthesia. Collaboration in the OR is the key to success, and it is important that pediatric urologists and pediatric anesthesiologists work together to balance the benefits and risks of general and spinal anesthesia. [Table presented]

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Institution

(Reh fuss, Kogan) Albany Medical College, United States (Bogaert) University Hospitals Leuven, United States

Publisher

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138.

Endocrine disrupting chemicals interfere with Leydig cell hormone pathways during testicular descent in idiopathic cryptorchidism.

Fenichel P., Chevalier N., Lahlou N., Coquillard P., Wagner-Mahler K., Pugeat M., Panaia-Ferrari P., Brucker-Davis F.

Embase

Frontiers in Endocrinology. 10 (JAN) (no pagination), 2019. Date of Publication: 2019.

[Short Survey]

AN: 627170392

Cryptorchidism, a frequent genital malformation in male newborn, remains in most cases idiopathic. On the basis of experimental, epidemiological, and clinical data, it has been included in the testicular dysgenesis syndrome and believed to be influenced, together with genetic and anatomic factors, by maternal exposure to endocrine disrupting chemicals (EDCs). Here, we analyze how EDCs may interfere with the control of testicular descent, which is regulated by two Leydig cell hormones, testosterone, and insulin like peptide 3 (INSL3).

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Embase

Institution

(Fenichel, Chevalier, Brucker-Davis) Department of Reproductive Endocrinology, University Hospital of Nice, Nice, France (Fenichel, Chevalier, Brucker-Davis) Institut National de la Recherche Medicale, UMR U1065, Universite Nice-Sophia Antipolis, Nice, France

(Lahlou) Department of Hormonology and Metabolic Disorders, Hopital Cochin, APHP, Paris-Descartes University, Paris, France

(Coquillard) Institut Sophia-Agrobiotech (INRA-CNRS, Nice University), Nice, France

(Wagner-Mahler) Department of Pediatrics, CHU Lenval, Nice, France
(Pugeat) Institut National de la Recherche Medicale, U1060 CaRMen, Federation
d'Endocrinologie, Hospices Civils de Lyon-1, Bron, France
(Panaia-Ferrari) Department of Biochemistry, University Hospital of Nice, Nice, France
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Year of Publication
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139.

Klinefelter syndrome: From pediatrics to geriatrics.

Shiraishi K., Matsuyama H.

Embase

Reproductive Medicine and Biology. 18 (2) (pp 140-150), 2019. Date of Publication: April 2019.

[Review]

AN: 627063421

Background: Klinefelter syndrome (KS) is one of the major causes of nonobstructive azoospermia (NOA). Microdissection testicular sperm extraction (micro-TESE) is often performed to retrieve sperm. Infertility specialists have to care for KS patients on a lifelong basis.

Method(s): Based on a literature review and our own experience, male infertility treatment and KS pathophysiology were considered on a lifelong basis. Main findings: Patients diagnosed early often have an increased number of aberrant X chromosomes. Cryptorchidism and hypospadias are often found, and surgical correction is required. Cryopreservation of testicular sperm during adolescence is an issue of debate because the sperm retrieval rate (SRR) in KS patients decreases with age. The SRR in adult KS patients is higher than that in other patients with NOA; however, low testosterone levels after micro-TESE will lower the general health and quality of life. KS men face a number of comorbidities, such as malignancies, metabolic syndrome, diabetes, cardiovascular disease, bone disease, and immune diseases, which ultimately results in increased mortality rates.

Conclusion(s): A deeper understanding of the pathophysiology of KS and the histories of KS patients before they seek infertility treatment, during which discussions with multidisciplinary teams are sometimes needed, will help to properly treat these patients.

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Author NameID

Shiraishi, Koji; ORCID: <http://orcid.org/0000-0002-3278-7747>

Institution

(Shiraishi, Matsuyama) Department of Urology, Yamaguchi University School of Medicine, Ube, Japan

Publisher

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E-mail: vgorayska@wiley.com)

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140.

Features of the metabolic syndrome in late adolescence are associated with impaired testicular function at 20 years of age.

Hart R.J., Doherty D.A., Mori T.A., Adams L.A., Huang R.-C., Minaee N., Handelsman D.J., McLachlan R., Norman R.J., Dickinson J.E., Olynyk J.K., Beilin L.J.

Embase

Human Reproduction. 34 (3) (pp 389-402), 2019. Date of Publication: 01 Mar 2019.

[Article]

AN: 626797029

STUDY QUESTION Are early signs of metabolic disorder in late adolescence associated with features of impaired testicular function many years before the majority seek parenthood?

SUMMARY ANSWER Adolescents with features of metabolic disorder at 17 years, or insulin resistance (IR) at 20 years of age, show impaired testicular function and altered hormone levels compared to those without metabolic disorder. WHAT IS KNOWN ALREADY Controversial evidence suggests a recent decline in sperm production potentially linked to environmental influences, but its cause remains unclear. Concomitant increases in obesity and diabetes suggest that lifestyle factors may contribute to this decline in testicular function. Although obesity has

been associated with adverse testicular function in some studies, it remains unclear whether poor testicular function merely reflects, or causes, poor metabolic health. If metabolic disorder were present in adolescence, prior to the onset of obesity, this may suggest that metabolic disorder maybe a precursor of impaired testicular function.

STUDY DESIGN, SIZE, DURATION The Western Australian Pregnancy Cohort (Raine) Study is a longitudinal study of children born in 1989-1991 who have undergone detailed physical assessments since birth (1454 male infants born). At 17 years of age, 490 boys underwent a hepatic ultrasound examination, serum cytokine assessment (n = 520) and a metabolic assessment (n = 544). A further metabolic assessment was performed at 20 years (n = 608). Testicular assessment was performed at 20 years; 609 had reproductive hormones measured, 404 underwent a testicular ultrasound and 365 produced a semen sample.

PARTICIPANTS/MATERIALS, SETTING, METHODS Testicular volume was estimated by ultrasonography, and semen analysis was performed according to World Health Organization guidelines. Concentrations of LH, FSH and inhibin B (inhB) in serum were measured by immunoassay and total testosterone by liquid chromatography-mass spectrometry. At 17 years of age, a liver ultrasound examination was performed to determine the presence of non-alcoholic fatty liver disease (NAFLD), and serum analysed for the cytokines interleukin-18 and soluble tumour necrosis factor receptor 1 and 2 (sTNFR1, sTNFR2). At 17 and 20 years of age, fasting blood samples were analysed for serum liver enzymes, insulin, glucose, triglycerides (TG), total cholesterol, high density lipoprotein and low density lipoprotein cholesterol, high sensitivity C-reactive protein and uric acid. The homeostatic model assessment (HOMA) was calculated and approximated IR was defined by a HOMA >4. Anthropometric data was collected and dual energy X-ray absorptiometry measurement performed for lean and total fat mass. As at this young age the prevalence of metabolic syndrome was expected to be low, a two-step cluster analysis was used using waist circumference, TGs, insulin, and systolic blood pressure to derive a distinct high-risk group with features consistent with the metabolic syndrome and increased cardiometabolic risk.

MAIN RESULTS AND THE ROLE OF CHANCE Men at age 17 years with increased cardiometabolic risk had lower concentrations of serum testosterone (medians: 4.0 versus 4.9 ng/mL) and inhB (193.2 versus 221.9 pg/mL) ($P < 0.001$ for both) compared to those within the low risk metabolic cluster. Men with ultrasound evidence of NAFLD (n = 45, 9.8%) had reduced total sperm output (medians: 68.0 versus 126.00 million, $P = 0.044$), testosterone (4.0 versus 4.7 ng/mL, $P = 0.005$) and inhB (209.1 versus 218.4 pg/mL, $P = 0.032$) compared to men without NAFLD. Men with higher concentrations of sTNFR1 at 17 years of age had a lower sperm output and serum concentration of inhB, with an increase in LH and FSH (all $P < 0.05$ after adjustment for age, BMI, abstinence and a history of cryptorchidism, varicocele, cigarette smoking, alcohol and drug use), compared to those without an elevated sTNFR1. Multivariable regression analysis, adjusting for confounders, demonstrated that men in the high-risk metabolic cluster at 20 years had a lower serum testosterone and inhB ($P = 0.003$ and $P = 0.001$,

respectively). A HOMA-IR > 4 was associated with a lower serum testosterone (P = <0.001) and inhB (P = 0.010) and an increase in serum FSH (P = 0.015). LIMITATIONS, REASONS FOR CAUTION This study is limited by the sample size and multiple comparisons, and causality cannot be proven from an observational study. Due to a 3-year interval between some metabolic assessments and assessment of testicular function, we cannot exclude the introduction of a bias into the study, as some of the participants and their testicular function will not have been fully mature at the 17-year assessment. WIDER IMPLICATIONS OF THE FINDINGS Irrespective of a proven causation, our study findings are important in that a significant minority of the men, prior to seeking parenthood, presented co-existent features of metabolic disorder and signs of testicular impairment. Of particular note is that the presence of NAFLD at 17 years of age, although only present in a minority of men, was associated with an almost 50% reduction in sperm output at 20 years of age, and that the presence of IR at 20 years was associated with a 20% reduction in testicular volume.

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Status

Embase

Institution

(Hart, Doherty, Dickinson) Division of Obstetrics and Gynaecology, University of Western Australia (UWA), Perth, Western Australia, Australia (Hart) Claremont Western Australia Australia

(Doherty, Minaee) Women and Infants Research Foundation, Perth, Western Australia, Australia

(Mori, Beilin) Medical School, UWA, Royal Perth Hospital, Western Australia, Australia

(Adams) Medical School, UWA, Sir Charles Gairdner Hospital, Nedlands, Western Australia, Australia

(Huang) Telethon Kids Institute, UWA, Perth, Western Australia, Australia

(Handelsman) ANZAC Research Institute, University of Sydney, Sydney, New South Wales, Australia

(McLachlan) Hudson Institute, Monash University, Melbourne, Australia

(Norman) Robinson Institute, Research Institute, School of Medicine, University of Adelaide and Fertility SA, Adelaide, Australia

(Olynyk) Department of Gastroenterology, Fiona Stanley and Fremantle Hospital and School of Medical and Health Sciences, Edith Cowan University, Joondalup, Western Australia, Australia

Publisher

Oxford University Press

Year of Publication

2019

141.

EED and EZH2 constitutive variants: A study to expand the Cohen-Gibson syndrome phenotype and contrast it with Weaver syndrome.

Griffiths S., Loveday C., Zachariou A., Behan L.-A., Chandler K., Cole T., D'Arrigo S., Dieckmann A., Foster A., Gibney J., Hunter M., Milani D., Pantaleoni C., Roche E., Sherlock M., Springer A., White S.M., Tatton-Brown K.

Embase

American Journal of Medical Genetics, Part A. 179 (4) (pp 588-594), 2019. Date of Publication: April 2019.

[Article]

AN: 626751218

Overgrowth-intellectual disability (OGID) syndromes are characterized by increased growth (height and/or head circumference $\geq +2$ SD) in association with an intellectual disability. Constitutive EED variants have previously been reported in five individuals with an OGID syndrome, eponymously designated Cohen-Gibson syndrome and resembling Weaver syndrome. Here, we report three additional individuals with constitutive EED variants, identified through exome sequencing of an OGID patient series. We compare the EED phenotype with that of Weaver syndrome (56 individuals), caused by constitutive EZH2 variants. We conclude that while there is considerable overlap between the EED and EZH2 phenotypes with both characteristically associated with increased growth and an intellectual disability, individuals with EED variants more frequently have cardiac problems and cervical spine abnormalities, boys have cryptorchidism and the facial gestalts can usually be distinguished.

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Embase

Author NameID

Griffiths, Sara; ORCID: <http://orcid.org/0000-0003-1736-2430>

Institution

(Griffiths, Tatton-Brown) Department of Clinical Genetics, St George's University of London, London, United Kingdom (Loveday, Tatton-Brown) Division of Genetics and Epidemiology, Institute of Cancer Research, Sutton, United Kingdom

(Zachariou) Division of Clinical Studies, Institute of Cancer Research, Sutton, United Kingdom

(Behan, Sherlock) Department of Endocrinology, Beaumont Hospital, Dublin, Ireland

(Chandler) Manchester Centre for Genomic Medicine, Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Science Centre, Manchester, United Kingdom

(Cole, Foster) Department of Clinical Genetics, Birmingham Women's and Children's NHS Foundation Trust, Birmingham, United Kingdom

(D'Arrigo, Pantaleoni) Developmental Neurology Unit, Fondazione IRCCS Istituto Neurologico C. Besta, Milan, Italy

(Dieckmann) Department of Neuropediatrics, Jena University Hospital, Jena, Germany

(Behan, Gibney, Roche) Department of Paediatric Growth, Diabetes and Endocrinology, Tallaght University Hospital, Dublin, Ireland

(Hunter, Springer) Department of Genetics, Monash Health, Melbourne, Australia

(Hunter, Springer) Department of Paediatrics, Monash University, Melbourne, Australia

(Milani) Pediatric Highly Intensive Care Unit, Fondazione IRCCS Ca' Granda, Ospedale Maggiore Policlinico, Milan, Italy

(Roche) Department of Endocrinology and Diabetes Mellitus, Tallaght University Hospital, Dublin, Ireland

(Roche) Trinity College Dublin, The University of Dublin, Dublin, Ireland

(Sherlock) Royal College of Surgeons in Ireland, Dublin, Ireland

(White) Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Melbourne, Australia

(White) Department of Paediatrics, University of Melbourne, Melbourne, Australia

(Tatton-Brown) South West Thames Regional Genetics Service, London, United Kingdom

Publisher
Wiley-Liss Inc. (E-mail: info@wiley.com)
Year of Publication
2019

Correction to: Maternal smoking during pregnancy and risk of cryptorchidism: a systematic review and meta-analysis (European Journal of Pediatrics, (2019), 178, 3, (287-297), 10.1007/s00431-018-3293-9).

Yu C., Wei Y., Tang X., Liu B., Shen L., Long C., Lin T., He D., Wu S., Wei G.

Embase

European Journal of Pediatrics. 178 (3) (pp 299-300), 2019. Date of Publication: 12 Mar 2019.

[Erratum]

AN: 625851604

The publisher regrets that the original version of this article contained an error. Table 1 was shown in the wrong version, thus corrected table is shown in this article. The original article has been corrected.

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Status

Embase

Author NameID

Wu, Shengde; ORCID: <http://orcid.org/0000-0002-0083-561X>

Institution

(Yu, Wei, Tang, Liu, Shen, Long, Lin, He, Wu, Wei) Department of Urology, Children's Hospital of Chongqing Medical University, Room 806, Kejiao Building (No. 6 Building), No. 136, 2nd Zhongshan Road, Yuzhong District, Chongqing City 400014, China (Yu, Wei, Tang, Wu, Wei) Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, Chongqing, China

(Liu, Lin, He, Wu, Wei) Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing, China

(Shen) China International Science and Technology Cooperation Base of Child Development and Critical Disorders, Chongqing, China

(Long) Chongqing Key Laboratory of Pediatrics Chongqing, Chongqing, China

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Year of Publication

2019

143.

Congenital inguinal hernia, hydrocoele and undescended testis.

Khoo A.K., Cleeve S.J.

Embase

Surgery (United Kingdom). 37 (4) (pp 225-230), 2019. Date of Publication: April 2019.

[Review]

AN: 2001661368

Congenital inguinal hernias (CIH), hydrocoeles and undescended testes (UDT) are common groin conditions in neonates, infants and children that are encountered by general practitioners, paediatricians, general surgeons and paediatric surgeons. CIH, hydrocoeles and UDT share a common embryological origin. Clinical differentiation between the three conditions can be challenging, particularly as they may exist in isolation or combination in the same patient. Accurate clinical distinction is imperative as the management and outcome is different for each condition. Surgery and outcomes for these conditions is discussed.

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Institution

(Khoo, Cleeve) Royal London Hospital, London, UK; Royal London Hospital, London, UK

Publisher

Elsevier Ltd

Year of Publication

2019

144.

Bilateral orchidopexies: synchronous or metachronous? Survey of BAPS and BAPU members and single-centre comparison.

Mostafa I.A., Shalaby M.S., Woodward M.N.

Embase

Journal of Pediatric Surgery. 54 (2) (pp 310-312), 2019. Date of Publication: February 2019.

[Article]

AN: 2001350415

Background/Aim: Approximately 20% of undescended testes (UDT) are bilateral. It is unclear whether bilateral orchidopexy (BO) should be undertaken synchronously (SBO) or metachronously (MBO). Our aim was to investigate current UK practice and the complications of SBO vs MBO.

Material(s) and Method(s): Following approval of BAPS and BAPU ethics committee, a survey was circulated to UK consultant pediatric surgeons and urologists regarding practice. A departmental retrospective review was additionally carried out for patients undergoing BO between 2005 and 2017.

Result(s): Forty-three consultant surgeons from 20 centres completed the survey. Overall, SBO was preferred by 70% for bilateral palpable UDT versus 30% for bilateral impalpable UDT. When one side was palpable and the other impalpable, 70% preferred SBO. Pediatric urologists were significantly more likely to undertake SBO than pediatric general surgeons. One hundred eighty-eight patients (376 testicular units) were identified who had undergone BO with a median follow up of 9 months. 144/188 (76.6%) underwent SBO, while 44 had MBO. There was no statistical difference in the complication rate between the two groups (7.6% in SBO vs 9.1% in MBO; $p = 0.66$).

Conclusion(s): The majority of the responding UK consultants, in particular pediatric urologists, favor SBO. This potentially offers a reduction in cost, more rapid completion of treatment, and is not associated with additional complications by comparison to MBO. We recommend SBO to be standard practice for bilateral UDT whenever possible.

Level of Evidence: Level III, Retrospective Comparative Study.

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Status

Embase

Institution

(Mostafa, Shalaby, Woodward) Department of Paediatric Surgery, Bristol Royal Hospital for Children, Bristol, United Kingdom (Shalaby) Department of Paediatric Surgery, Ain Shams University, Cairo, Egypt

Publisher

W.B. Saunders

Year of Publication

2019

145.

Congenital diaphragmatic hernia is associated with nonscrotal testes.

Janssen S., Heiwegen K., van Rooij I.A., van Alfen-van der Velden J., de Blaauw I., Botden S.M.

Embase

Journal of Pediatric Surgery. 54 (3) (pp 445-448), 2019. Date of Publication: March 2019.

[Article]

AN: 2000609614

Background: Congenital diaphragmatic hernia (CDH) is a rare birth defect with a high mortality and morbidity. Nonscrotal testes (NST) are the most reported genital anomaly in boys. Both defects have known associated anomalies, but little is known about the association between CDH and NST. This study evaluates this association and the location of the NST in a large cohort of male CDH survivors. Moreover, we analyzed possible associative factors for NST in CDH patients.

Method(s): A cohort of CDH patients, born between January 2000 and March 2014 and treated in a high volume expertise center, was evaluated retrospectively. Boys with a minimum follow-up of 18 months were included. The patients were evaluated for testes location, performed orchidopexy, and possible associative factors such as birth weight, gestational age, other congenital anomalies and CDH characteristics (surgical treatment, approach and ECMO).

Result(s): Seventy-five CDH patients were included. Twenty-seven (36%) were diagnosed with NST, of which 22 (29%) received orchidopexy. In 54 patients (72%) there were reports on testes location at birth and location was known for all patients at the age of 18 months, although side of NST was unknown in four. The location of the NST was mostly ipsilateral to the CDH (n = 20, 87%), of which eight (35%) had a bilateral NST with a unilateral CDH. There were no significant differences in birth weight, gestational age, and CDH specific characteristics in patients with or without NST.

Conclusion(s): This study shows a strong association between CDH and NST, with a prevalence of 36%. However, no specific characteristics of the CDH were related to the NST. The testes of all male CDH patients should be thoroughly evaluated in the first year of their life, to ensure a proper and timely treatment.

Level of Evidence: Level IV; case series.

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Status

Embase

Institution

(Janssen, Heiwegen, de Blaauw, Botden) Department of Surgery-Division of Pediatric Surgery, Radboudumc-Amalia Children's Hospital, Nijmegen, Netherlands (van Rooij) Radboud Institute for Health Sciences, Department for Health Evidence, Radboudumc, Nijmegen, Netherlands (van Alfen-van der Velden) Department of Pediatrics, Radboudumc-Amalia Children's Hospital, Nijmegen, Netherlands

Publisher

W.B. Saunders

Year of Publication

2019

146.

Diagnostic Accuracy of Virtual Touch Quantification and Virtual Touch Imaging Quantification Sonoelastography Techniques in Pediatric Undescended Testes Patients.

Ozkan M.B., Bilgic M., Bicakci U., Germiyanoglu C.

Embase

Journal of Diagnostic Medical Sonography. 35 (2) (pp 96-103), 2019. Date of Publication: 01 Mar 2019.

[Article]

AN: 625851253

The purpose of this study was to determine the usefulness of virtual touch quantification (VTQ) and virtual touch imaging quantification (VTIQ) techniques for assessing undescended testes stiffness, by age and location. One testicle from each of 84 participants (31 of whom were healthy volunteers) was assessed by the point shear-wave elastography (p-SWE) method, using both VTQ and VTIQ techniques. The patients were grouped by location of assessment (intra-abdominal, inguinal, and scrotal) and age (group 1, healthy patients; group 2, patients age one and younger; and group 3, patients older than age one). Although the VTQ and VTIQ methods for group 2 showed similar results in the Bland-Altman plot, for group 3, the VTIQ method was more effective than the VTQ method. The use of p-SWE with VTIQ may be an appropriate technique

for determining the tissue stiffness of testes located in the inguinal canal and for patients over one year of age.

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Status

Embase

Author NameID

Ozkan, Mehmet Burak; ORCID: <http://orcid.org/0000-0003-4672-0671>

Institution

(Ozkan) Department of Pediatric Radiology Department, Dr Sami Ulus Research and Training Hospital, Altindag, Ankara, Turkey (Bilgic) Department of Pediatric Radiology, Omu, Samsun, Turkey

(Bicakci) Department of Pediatric Surgery, Omu, Samsun, Turkey

(Germiyanoglu) Department of Urology, Omu, Turkey

Publisher

SAGE Publications Inc. (E-mail: claims@sagepub.com)

Year of Publication

2019

147.

An updated mortality risk analysis of the post-pubertal undescended testis.

Shah S., Feustel P.J., Knuth J., Welliver C.

Embase

Canadian Urological Association Journal. 13 (1) (pp E1-E6), 2019. Date of Publication: January 2019.

[Article]

AN: 625684914

Introduction: The undescended testicle (UDT) presents a problem in post-pubertal (PP) men, as it carries an increased risk of developing a germ cell tumour (GCT). Management of the PP patient with an UDT must weigh the relative risk (RR) of perioperative mortality (POM) from orchiectomy against the lifetime risk of death from a GCT.

Method(s): The most recent data on GCT mortality were obtained from the National Centre for Health Statistics. Standard life tables were used to calculate the cumulative risk over a man's

lifetime based on age. The increased RR of GCT in men with UDT was determined by weighing the observed and expected rates from literature review. Life table data was then multiplied by the RR to define the risk of GCT in men with UDT. Data from patients undergoing similar risk surgical procedures, stratified by American Society of Anesthesiologists (ASA) class, was used to determine POM.

Result(s): Lifetime risk of dying from GCT decreases with increasing age. POM exceeded risks of death from GCT for men after age 50.2 for ASA class 1 and age 35.4 for ASA class 2. Men with an ASA class higher than 2 have a higher risk of POM compared to GCT for all ages.

Conclusion(s): We found different ages from previous reports at which observation is advised. We consider prophylactic orchiectomy only in men who are under 50.2 years if ASA class 1 and under 35.4 years if ASA class 2. Men with an ASA class 3 or higher should always undergo observation.

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Embase

Institution

(Shah, Welliver) Department of Surgery, Division of Urology, Albany Medical College, Albany, NY, United States (Feustel) Center for Neuropharmacology and Neuroscience, Albany Medical College, Albany, NY, United States

(Knuth) Department of Anesthesiology, Albany Medical College, Albany, NY, United States

(Welliver) Albany Stratton VA Medical Center, Albany, NY, United States

Publisher

Canadian Urological Association (E-mail: josephine.sciortino@cua.org)

Year of Publication

2019

148.

Anomalies of the genitourinary tract in children with 22q11.2 deletion syndrome.

Van Batavia J.P., Crowley T.B., Burrows E., Zackai E.H., Sanna-Cherchi S., McDonald-McGinn D.M., Kolon T.F.

Embase

American Journal of Medical Genetics, Part A. 179 (3) (pp 381-385), 2019. Date of Publication: March 2019.

[Article]

AN: 625654466

The 22q11.2 deletion syndrome (22q11.2DS) involves multiple organ systems with variable phenotypic expression. Genitourinary tract abnormalities have been noted to be present in up to 30-40% of patients. At our institution, an internationally recognized, comprehensive, and multidisciplinary 22q11.2DS care center has been providing care to these children. We sought to report on the incidence of genitourinary tract anomalies in this large cohort and, therefore, retrospectively reviewed all patients who underwent a complete evaluation from 1992 to March 2017. We identified all children with any genital or urinary tract anomaly. For all children with a diagnosis of hydronephrosis, the underlying etiology was determined, when possible. Overall, 1,073 of 1,267 children with 22q11.2DS underwent renal evaluations at our institution. Hundered Sixty-Two (15.1%) children had structural abnormalities of their kidneys/urinary tracts. The majority of children with hydronephrosis (63%) had isolated upper tract dilation without any additional diagnoses. Boys were significantly more likely to be diagnosed with a genital abnormality than girls (7.7 vs. 0.5%, $p < 0.001$). Of the 649 boys in the entire cohort, 24 (3.7%) had cryptorchidism and 24 (3.7%) had hypospadias, which was noted to be mild in all except one boy. Overall, findings of hydronephrosis, unilateral renal agenesis, and multicystic dysplastic kidney occur at higher rates than expected in the general population. Given these findings, in addition to routine physical examination, we believe that all patients with 22q11.2DS warrant screening RBUS at time of diagnosis.

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Embase

Author NameID

Van Batavia, Jason P.; ORCID: <http://orcid.org/0000-0002-1592-7824> Crowley, Terrence B.;

ORCID: <http://orcid.org/0000-0002-3272-5458>

Institution

(Van Batavia, Kolon) Division of Urology, Children's Hospital of Philadelphia, Philadelphia, PA, United States (Crowley, Burrows, Zackai, McDonald-McGinn) Division of Human Genetics, Children's Hospital of Philadelphia, Philadelphia, PA, United States

(Sanna-Cherchi) Division of Nephrology, New York-Presbyterian Hospital Columbia University Medical Center, New York, NY, United States

Publisher

Wiley-Liss Inc. (E-mail: info@wiley.com)

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2019

149.

Family and neighborhood socioeconomic inequality in cryptorchidism and hypospadias: A nationwide study from Sweden.

Li X., Sundquist J., Hamano T., Sundquist K.

Embase

Birth Defects Research. 111 (2) (pp 78-87), 2019. Date of Publication: 15 Jan 2019.

[Article]

AN: 625545678

Objectives: To examine whether there is an association between neighborhood deprivation and incidence of cryptorchidism and hypospadias, after accounting for family-level and individual-level sociodemographic characteristics.

Method(s): All boys born in Sweden between January 1, 2001 and December 31, 2010 were followed. Data were analyzed by multilevel logistic regression, with family-level and individual-level characteristics at the first level and level of neighborhood deprivation at the second level.

Result(s): During the study period, among a total of 497,584 boys, 8,584 (1.7%) and 3,704 (0.7%) were diagnosed with cryptorchidism and hypospadias, respectively. Cumulative rates for cryptorchidism and hypospadias increased with increasing levels of neighborhood deprivation. In the study population, 1.5 per 100 and 2.0 per 100 boys, in the least and most deprived neighborhoods were diagnosed with cryptorchidism and 0.7 per 100 and 0.9 per 100 boys were diagnosed with hypospadias. Incidence of hospitalization for cryptorchidism and hypospadias increased with increasing neighborhood-level deprivation across all family-level and individual-level sociodemographic categories. The odds ratio (OR) for cryptorchidism and hypospadias for those living in high-deprivation neighborhoods versus those living in low-deprivation neighborhoods was 1.13 (95% confidence interval [CI] = 1.05-1.21) and 1.24 (95% CI = 1.12-1.37). High neighborhood deprivation remained significantly associated with higher odds of hypospadias after adjustment for family-level and individual-level sociodemographic characteristics (OR = 1.20, 95% CI = 1.08-1.35).

Conclusion(s): This study is the largest so far on neighborhood influences on cryptorchidism and hypospadias. Our results suggest that neighborhood deprivation is associated with a moderate incidence of hypospadias independent of family-level and individual-level sociodemographic characteristics.

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Embase

Author NameID

Li, Xinjun; ORCID: <http://orcid.org/0000-0002-5559-4657>

Institution

(Li, Sundquist, Sundquist) Center for Primary Health Care Research, Lund University, Malmo, Sweden (Sundquist, Sundquist) Department of Family Medicine and Community Health, Icahn School of Medicine at Mount Sinai, New York, NY, United States

(Sundquist, Hamano, Sundquist) Department of Functional Pathology, Center for Community-Based Healthcare Research and Education (CoHRE), School of Medicine, Shimane University, Izumo, Japan

(Hamano) Department of Sports Sociology and Health Sciences, Kyoto Sangyo University, Kyoto, Japan

Publisher

John Wiley and Sons Inc. (P.O.Box 18667, Newark NJ 07191-8667, United States)

Year of Publication

2019

150.

Molecular genetics and phenotype/genotype correlation of 5-alpha reductase deficiency in a highly consanguineous population.

Alswailem M.M., Alzahrani O.S., Alghofaili L., Qasem E., Almohanaa M., Alsagheir A., Bin Abbas B., Attia N.A., Al Shaikh A., Alzahrani A.S.

Embase

Endocrine. 63 (2) (pp 361-368), 2019. Date of Publication: 15 Feb 2019.

[Article]

AN: 624185140

Context and objectives: 5-alpha reductase deficiency is a rare 46,XY disorder of sex development. We present detailed phenotypic and genotypic features of a cohort of 24 subjects from a highly consanguineous population of Saudi Arabia Subjects and Methods: We studied the clinical presentation and hormonal profiles of 24 subjects diagnosed with 5-alpha reductase deficiency and performed genetic testing on DNA isolated from their peripheral blood using polymerase chain reaction and direct sequencing of the SRD5A2.

Result(s): All subjects had 46,XY karyotype and presented with atypical appearance of external genitalia ranging from clitoromegaly, micophallus with hypospadias, undescended testes to completely normally looking female genitalia. Thirteen (54%) of them had severe under virilization and were assigned female sex at birth. The other 11 subjects were raised as males. Stimulated Testosterone:Dihydrotestosterone ratio was high in all 16 subjects in whom it was measured. The genetic testing revealed 2 nonsense mutations (p.R103X and p.R227X) in 2 unrelated subjects, 3 missense mutations (p.P181L, p.A228T, p.R246Q) in 11 subjects and a splice site mutation (IVS1-2A > G) in 11 other subjects. There was significant phenotypic variability even in subjects with the same mutation and also within the same family.

Conclusion(s): This is the first and largest report of the clinical and molecular genetics of 5-alpha reductase deficiency from the Middle East. It shows weak genotype/phenotype correlation and significant phenotypic heterogeneity. IVS1-2A > G mutation is the most common mutation and is likely to be a founder mutation in this part of the world.

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Embase

Institution

(Alswailem, Alghofaili, Qasem, Almohanaa, Alzahrani) Department of Molecular Oncology, King Faisal Specialist Hospital & Research Centre, Riyadh, Saudi Arabia (Alzahrani, Alsagheir, Bin Abbas) Department of Pediatrics, King Faisal Specialist Hospital & Research Centre, Riyadh, Saudi Arabia

(Attia, Al Shaikh) Department of Pediatrics, King Abdulaziz Medical City, Jeddah, Saudi Arabia (Alzahrani) Department of Medicine, King Faisal Specialist Hospital & Research Centre, Riyadh, Saudi Arabia

Publisher

Humana Press Inc. (E-mail: humana@humanapr.com)

Year of Publication

2019

151.

Surgical Management of Undescended Testis - Timetable and Outcome: A Debate.

Thorup J., Cortes D.

Embase

Sexual development : genetics, molecular biology, evolution, endocrinology, embryology, and pathology of sex determination and differentiation. 13 (1) (pp 11-19), 2019. Date of Publication: 2019.

[Review]

AN: 627156722

Around 1.4-3.8% of boys in the Western world are operated because of cryptorchidism. This means that orchidopexy remains one of the most common surgical procedures performed in boys. As a consequence, several consensus reports, guidelines, and reviews dealing with the management of cryptorchidism have been published recently. Based on our research and 30 years' experience with the management of cryptorchidism, the intention of the present publication is to advise on the surgical management and comment on the expected outcome, especially with focus on the controversies related to guidelines and reviews. Except for late referral and waiting lists, which may be practical problems, there is no reason to postpone orchidopexy for nonsyndromic congenital cryptorchid testes beyond 6 months. There is good evidence that such strategy improves the fertility potential and decreases the risk of testicular cancer. In cases with genuine gonadotropin insufficiency, early surgical correction is not enough and adjuvant LH-RH treatment should be implemented to improve the fertility potential. Cryopreservation may be an option in case of treatment failure of adjuvant LH-RH. A prerequisite for such management includes serum hormone assessment and evaluation of testicular biopsies at orchidopexy.

Ascended testes contribute to 20-60% of operative cases and should be treated when diagnosed.

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PMC Identifier

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Publisher

NLM (Medline)

Year of Publication

2019

152.

Optimal Management of Undescended Testis in Boys with Cerebral Palsy. A Debate.

Springer A.

Embase

Sexual development : genetics, molecular biology, evolution, endocrinology, embryology, and pathology of sex determination and differentiation. 13 (1) (pp 20-25), 2019. Date of Publication: 2019.

[Review]

AN: 627156327

Cerebral palsy is a rare condition following injury of the developing brain and including nonprogressive neurological disorders, spasticity, intellectual impairment and others. Boys with cerebral palsy have a high incidence of undescended testis. Although the motives for treatment (infertility, cancer prevention, psychological aspects, testicular torsion) are not different in boys without neurological impairment, the decision-making process in boys with cerebral palsy is very difficult. Besides medical and surgical arguments the discussion involves challenging ethical issues.

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PMC Identifier

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153.

Is Hormonal Treatment of Congenital Undescended Testes Justified? A Debate.

Hadziselimovic F.

Embase

Sexual development : genetics, molecular biology, evolution, endocrinology, embryology, and pathology of sex determination and differentiation. 13 (1) (pp 3-10), 2019. Date of Publication: 2019.

[Review]

AN: 627155363

Abnormal germ cell development in cryptorchidism is not a result of a congenital dysgenesis but is preceded by a hormone imbalance and perturbation in germ cell-specific gene expression during abrogated mini-puberty. Adequate treatment with low doses of GnRHa enables 86% of men to achieve a normal sperm count and, most importantly, prevent development of azoospermia. GnRHa treatment induces a significant transcriptional response, including protein coding genes involved in pituitary development, the hypothalamic-pituitary-gonadal axis, and testosterone synthesis. Furthermore, hormonal treatment to achieve epididymo-testicular descent as a first choice of treatment of cryptorchidism has a long tradition in Europe. It eliminates the necessity of subsequent surgery. Moreover, in the cases of non-responders it facilitates orchidopexy and contributes considerably to a reduced incidence of unilateral and the more serious bilateral complete post-surgical testicular atrophy.

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154.

Changing Trends in the Referral and Timing of Treatment for Congenital Cryptorchidism: A Single-Center Experience from Bosnia and Herzegovina.

Zvizdic Z., Islamovic B., Milisic E., Jonuzi A., Vranic S.

Embase

Journal of Pediatric Surgery. (no pagination), 2019. Date of Publication: 2019.

[Article]

AN: 2002808318

Background: Cryptorchidism is the most common male urogenital tract disorder identified at birth. Treatment delays of cryptorchidism may be associated with significant complications such as subfertility and testicular cancer. The currently recommended age for performing orchidopexy is between 6 and 12 months of age and no later than 18 months. The aim of this study was to investigate the trends in the pattern of referral and age of boys at the time of operative treatment of congenital cryptorchidism at the largest tertiary care center in Bosnia and Herzegovina.

Method(s): The study included all boys who underwent orchidopexy for congenital cryptorchidism during two equivalent periods: 2008-2010 and 2015-2017. We assessed the referral age of patients, the age of patients at the time of orchidopexy, laterality of cryptorchidism, position of cryptorchidic testes palpated before surgery, the intraoperative position of cryptorchidic testis, a clinical position of the testis at follow up, and risk factors for late orchidopexy.

Result(s): In total, 324 patients with 386 testes underwent orchidopexy for congenital cryptorchidism during the study periods. Of these patients, 62 received a bilateral orchidopexy (19.1%). Total referral age of patients with congenital cryptorchidism was 23 months (range, 4-74.5 months). Total median age at surgery was 24 months (range, 6-74 months). One hundred and eleven patients (28.8%) underwent surgery at less than the age of 12 months, 136 (35.2%) at less than the age of 18 months, and 250 (64.8%) patients underwent surgery after the age of 18 months. The analysis of the observed two periods (2008-2010 and 2015-2017) showed a statistically significant decrease in the mean referral age and the mean age at surgery over the last 5 years (2015-2017) ($p = 0.007$ and $p = 0.003$, respectively).

Conclusion(s): Current guidelines for timely operative treatment for congenital cryptorchidism have not been fully implemented in Bosnia and Herzegovina but a gradual improvement is evident. The main factor contributing to delays in orchidopexy was delayed or neglected referral by referring physicians. Optimizing the time of orchidopexy will require an improved coordination at all levels of pediatric health care to diminish the long-term consequences of cryptorchidism.

Type of Study: Retrospective.

Level of Evidence: III.

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PMC Identifier

31500876 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31500876>]

Status

Article-in-Press

Institution

(Zvizdic, Milisic, Jonuzi) Clinic of Pediatric Surgery, University Clinical Center Sarajevo, Sarajevo, Bosnia and Herzegovina (Islamovic) Public Medical Center Visoko, Visoko, Bosnia and Herzegovina

(Vranic) College of Medicine, QU Health, Qatar University, Doha, Qatar

Publisher

W.B. Saunders

Year of Publication

2019

155.

Cryptorchidism, gonocyte development, and the risks of germ cell malignancy and infertility: A systematic review.

Loebenstein M., Thorup J., Cortes D., Clasen-Linde E., Hutson J.M., Li R.

Embase

Journal of Pediatric Surgery. (no pagination), 2019. Date of Publication: 2019.

[Review]

AN: 2002324074

Background/Aim: Cryptorchidism, or undescended testis (UDT) occurs in 1%-4% of newborn males and leads to a risk of infertility and testicular malignancy. Recent research suggests that infertility and malignancy in UDT may be caused by abnormal development of the neonatal germ cells, or gonocytes, which normally transform into spermatogonial stem cells (SSC) or undergo apoptosis during minipuberty at 2-6 months in humans (2-6 days in mice). We aimed to identify the current knowledge on how UDT is linked to infertility and malignancy.

Method(s): Here we review the literature from 1995 to the present to assess the possible causes of infertility and malignancy in UDT, from both human studies and animal models.

Result(s): Both the morphological steps and many of the genes involved in germ cell development are now characterized, but the factors involved in gonocyte transformation and apoptosis in both normal and cryptorchid testes are not fully identified. During minipuberty there is evidence for the hypothalamic-pituitary axis stimulating gonocyte transformation, but without known direct control by LH and androgen, although FSH may have a role. An arrested gonocyte maybe the origin of later malignancy at least in syndromic cryptorchid testes in humans, which is

consistent with the recent finding that gonocytes are normally absent in a rodent model of congenital cryptorchidism, where malignancy has not been reported.

Conclusion(s): The results of this review strengthen the view that malignancy and infertility in men with previous UDT may be caused by abnormalities in germ cell development during minipuberty.

Type of Study: Systematic review (secondary, filtered) Level of evidence: Level I.

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PMC Identifier

31327540 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31327540>]

Status

Article-in-Press

Institution

(Loebenstein, Hutson, Li) Douglas Stephens Surgical Research Group, Murdoch Children's Research Institute, Melbourne, Australia (Loebenstein, Hutson, Li) Department of Paediatrics, University of Melbourne, Australia

(Hutson) Department of Urology, The Royal Children's Hospital, Melbourne, Australia

(Thorup) Department of Paediatric Surgery, Rigshospitalet, Copenhagen, Denmark

(Thorup, Cortes) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Cortes) Section of Endocrinology, Department of Pediatrics, Copenhagen University Hospital Hvidovre, Denmark

(Clasen-Linde) Department of Pathology, Copenhagen University Hospital Rigshospitalet, Denmark

Publisher

W.B. Saunders

Year of Publication

2019

156.

Sertoli Cell Number Correlates with Serum Inhibin B in Infant Cryptorchid Boys.

Hildorf S., Dong L., Thorup J., Clasen-Linde E., Yding Andersen C., Cortes D.

Embase

Sexual development : genetics, molecular biology, evolution, endocrinology, embryology, and pathology of sex determination and differentiation. 13 (2) (pp 74-82), 2019. Date of Publication: 2019.

[Article]

AN: 626907071

Postnatal maturation of Sertoli cells is crucial for male fertility. The aim of this study was to assess the association between the Sertoli cell number per tubule cross-section (SC/T), the serum level of the Sertoli cell-produced inhibin B, and the A-dark spermatogonia number per tubule (Ad/T) in cryptorchid boys. Forty infant cryptorchid boys aged 4-35 months (median: 13 months) were included in the study. During orchiopexy, blood samples for serum inhibin B, luteinizing hormone (LH), and follicle stimulating hormone (FSH) and testicular biopsies were obtained. Histological sections were evaluated by quantitative immunohistochemical and immunofluorescence analysis including VASA and SOX9 (Sertoli cell marker) in order to measure the tubular germ cell number (G/T), Ad/T, and SC/T. The SC/T correlated negatively with age ($p < 0.0002$) and positively with G/T, Ad/T, inhibin B, FSH, and LH (all $p < 0.01$). Inhibin B correlated with LH ($p < 0.0001$), but not with FSH ($p = 0.2077$). The SC/T:G/T ratio positively correlated with age ($p < 0.0001$). Boys with Ad spermatogonia at surgery had a higher number of Sertoli cells compared to boys without Ad spermatogonia. In conclusion, a correlation between Sertoli cell number and inhibin B was proven, indicating that inhibin B possibly reflects the function of Sertoli cells in infant cryptorchid boys.

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PMC Identifier

30889614 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=30889614>]

Publisher

NLM (Medline)

Year of Publication

2019

157.

Association of Interleukin-31 gene polymorphisms with risk of cryptorchidism in a Chinese population.

Zou B., Yu Z., Huang J., Tan C., Wang H., Fu J., Li X., Wang X., Cui S., Tang T.

Embase

Medicine. 98 (22) (pp e15861), 2019. Date of Publication: 01 May 2019.

[Article]

AN: 628141033

This study aims to investigate the possible association between Interleukin-31 (IL-31) gene polymorphisms and cryptorchidism risk. Two single nucleotide polymorphisms of IL-31, rs7977932 (C/G) and rs4758680 (C/A), were selected to be investigated in this study. Polymerase chain reaction-restriction fragment length polymorphism methods were used to discriminate the selected single nucleotide polymorphisms of IL-31 gene. A hospital-based case-control study of 112 cryptorchidism patients and 425 healthy controls was conducted. The frequencies of the C allele of rs4758680 in the patients with cryptorchidism were significantly higher compared with those in controls (89% vs 83%, $P = .02$, OR = 0.58, 95% CI = 0.37-0.92). Compared with CC genotype in dominant model, notable decreased frequencies of A carriers (CA/AA genotypes) were observed in cryptorchidism patients ($P = .03$, OR = 0.58, 95% CI = 0.35-0.96). Results demonstrated that IL-31 gene polymorphisms were associated with the genetic susceptibility to cryptorchidism in a Chinese population. Compared with CC genotype, the A carriers (CA/AA genotypes) of rs4758680 were protect factors in cryptorchidism susceptibility.

PMC Identifier

31145337 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31145337>]

Institution

(Zou, Huang, Tan, Wang, Fu, Li, Wang, Cui, Tang) Department of Urology, Armenia (Zou, Huang, Tan, Wang, Fu, Li, Wang, Cui, Tang) Urogenital Diseases Lab, Affiliated Hospital of North Sichuan Medical College, Nanchong, China

(Yu) Department of Urology, Chongqing Three Gorges Central Hospital, Wanzhou, China

Publisher

NLM (Medline)

Year of Publication

2019

158.

Endocrine Complications of Noonan Syndrome beyond Short Stature.

Venugopal V., Romero C.J.

Embase

Pediatric endocrinology reviews : PER. 16 (Supplement 2) (pp 465-470), 2019. Date of Publication: 01 May 2019.

[Article]

AN: 627933680

Noonan syndrome (NS) is a diagnosis that is made clinically based on features including typical facies, congenital heart defects, short stature and developmental delay. Approximately 50% of the patients have identified mutations in the PTPN11 gene, and a smaller percentage of mutations have been reported in other genes such as SOS1, RAF1 and RIT1. Despite normal birth length, patients typically reach adult height below normal. Other than growth, endocrine complications of NS are not as commonly reported. These include possible pathology in thyroid function, pubertal development and bone metabolism. Some investigators have looked to see if genetic mutations in these patients could pose a risk for future endocrinopathies. This chapter reviews reports on endocrine dysfunction other than growth in patients with NS. The information is meant to enhance awareness in those providers who care for these patients to the possibility of other existing endocrinopathies. Most importantly, it supports and highlights the endocrinologist's role in the care of patients with NS.

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PMC Identifier

31115198 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31115198>]

Institution

(Venugopal) Senior Associate Consultant, Mayo Clinic Health System, United States (Romero)
Division of Pediatric Endocrinology and Metabolism, Icahn School of Medicine at Mount Sinai,
United States

Publisher

NLM (Medline)

Year of Publication

2019

159.

The History of Noonan Syndrome.

Miller B.S.

Embase

Pediatric endocrinology reviews : PER. 16 (Supplement 2) (pp 424-427), 2019. Date of Publication: 01 May 2019.

[Article]

AN: 627933009

Early in her career, Jacqueline Noonan, a pediatric cardiologist, recognized that a number of children with valvular pulmonary stenosis had similar facial features. Dr. Noonan reported the clinical characteristics of this condition including short stature, hypertelorism, ptosis, mild mental retardation, undescended testes, and skeletal malformations. Further characterization of Noonan Syndrome led to the development of clinical criteria for the diagnosis of the condition.

Identification of the first genetic cause of Noonan Syndrome, mutation of ptpn11 was reported in 2001. Multiple subsequent genes have been identified as causes of Noonan Syndrome and the related Rasopathies.

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PMC Identifier

31115193 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31115193>]

Institution

(Miller) Division of Endocrinology, University of Minnesota Masonic Children's Hospital, 2450 Riverside Avenue, Minneapolis, MN 55454, United States

Publisher

NLM (Medline)

Year of Publication

2019

160.

Prevalence and surgical management of pubic hypertrophy in hypospadias patients: results from a high-volume surgeon.

Bandini M., Sekulovic S., Stanojevic N., Spiridonescu B., Pesic V., Sansalone S., Slavkovic M., Briganti A., Salonia A., Montorsi F., Djinoic R.

Embase

International braz j urol : official journal of the Brazilian Society of Urology. 45 (6) (pp 1238-1248), 2019. Date of Publication: 01 Nov 2019.

[Article]

AN: 630094041

INTRODUCTION: Pubic hypertrophy, defined as an abnormal and abundant round mass of fatty tissue located over the pubic symphysis, is frequently underestimated in patients with hypospadias. We examined the prevalence of this condition, as well as the outcomes associated with its surgical treatment. **MATERIAL AND METHODS:** Within 266 hypospadias patients treated at our clinic, we assessed the prevalence of pubic hypertrophy, and we schematically described the surgical steps of pubic lipectomy. Multivariable logistic regression (MLR) tested for predictors of pubic hypertrophy. Finally, separate MLRs tested for predictors of fistula and any complications after pubic lipectomy.

RESULT(S): Of 266 hypospadias patients, 100 (37.6%) presented pubic hypertrophy and underwent pubic lipectomy. Patients with pubic hypertrophy more frequently had proximal hypospadias (44 vs. 7.8%), disorders of sex development (DSD) (10 vs. 0.6%), cryptorchidism (12 vs. 2.4%), and moderate (30degree-60degree) or severe (>60degree) penile curvature (33 vs. 4.2%). In MLR, the location of urethral meatus (proximal, Odds ratio [OR]: 10.1, $p < 0.001$) was the only significant predictor of pubic hypertrophy. Finally, pubic lipectomy was not associated with increased risk of fistula (OR: 1.12, $p = 0.7$) or any complications (OR: 1.37, 95% CI: 0.64-2.88, $p = 0.4$) after multi-variable adjustment.

CONCLUSION(S): One out of three hypospadias patients, referred to our center, presented pubic hypertrophy and received pubic lipectomy. This rate was higher in patients with proximal hypospadias suggesting a correlation between pubic hypertrophy and severity of hypospadias. Noteworthy, pubic lipectomy was not associated with increased risk of fistula or any complications.

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PMC Identifier

31808413 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31808413>]

Institution

(Bandini, Sekulovic, Stanojevic, Spiridonescu, Pesic, Slavkovic, Djinovic) Sava Perovic Foundation, Center for Genito-Urinary Reconstructive Surgery, BelMedic General Hospital, Belgrade, Serbia (Bandini, Briganti, Salonia, Montorsi) Division of Oncology and Unit of Urology, IRCCS Ospedale San Raffaele, Vita-Salute San Raffaele University, Milan, URI, Italy (Spiridonescu) Clinical Institute Fundeni, Center for Urology and Renal Transplantation, Bucharest, Romania (Sansalone) Department of Experimental Medicine and Surgery, University of Tor Vergata, Rome, Italy

Publisher

NLM (Medline)

Year of Publication

2019

161.

Ectopic Posterior Pituitary, Polydactyly, Midfacial Hypoplasia and Multiple Pituitary Hormone Deficiency due to a Novel Heterozygous IVS11-2A>C(C.1957-2A>C) Mutation in GLI2 Gene. Demiral M., Demirbilek H., Unal E., Durmaz C.D., Ceylaner S., Ozbek M.N.

Embase

Journal of clinical research in pediatric endocrinology. (no pagination), 2019. Date of Publication: 29 Nov 2019.

[Article]

AN: 630012098

We, report a novel heterozygous IVS11-2A>C(c.1957-2A>C) mutation in GLI2 gene with an extremely distinct phenotypical expression in two siblings and their father from an unrelated family. The index case was a boy who developed cholestasis and hypoglycaemia at the neonatal period. He had postaxial polydactyly, mid-facial hypoplasia, high palatal arch, micropenis, and bilateral cryptorchidism. Laboratory examination revealed a diagnosis of multiple pituitary hormone deficiency. There were severe anterior pituitary hypoplasia, absent pituitary stalk and ectopic posterior pituitary on pituitary MR imaging which suggested pituitary stalk interruption syndrome (PSIS) with no other midline structural abnormality. In molecular genetic analysis, a novel heterozygous splicing IVS11-2A>C (c.1957-2A>C) mutation detected in GLI2 gene. His father and a 6-year-old brother with the identical mutation also had unilateral postaxial polydactyly and mid-facial hypoplasia whilst no pituitary hormone deficiency. Present novel heterozygous mutation detected in the GLI2 gene suggested an extremely variable clinical phenotype in individuals with identical mutation, even in those within the same family and incomplete penetrance of GLI2 mutations.

PMC Identifier

31782289 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31782289>]

Institution

(Demiral, Unal, Ozbek) Gazi Yasargil Training and Research Hospital Clinics of Paediatric Endocrinology, Turkey (Demirbilek) Hacettepe University, Faculty of Medicine, Department of Paediatric Endocrinology, Ankara, Turkey

(Durmaz) Gazi Yasargil Training and Research Hospital, Medical Genetics, Turkey
(Ceylaner) InterGen Genetic Diagnosis Center, Medical Genetics, Ankara, Turkey

Publisher

NLM (Medline)

Year of Publication

2019

162.

The impact of age at orchiopexy on testicular cancer outcomes.

Higgins M., Smith D.E., Gao D., Wilcox D., Cost N.G., Saltzman A.F.

Embase

World journal of urology. (no pagination), 2019. Date of Publication: 28 Nov 2019.

[Article]

AN: 630011051

PURPOSE: To estimate how many boys with UDT must undergo orchiopexy to prevent one case of TC, one death from TC and one exposure to TC treatment beyond radical orchiectomy as compared to being treated at an older age.

METHOD(S): This retrospective study utilized data from a 2007 Swedish study of males who underwent orchiopexy for UDT (Pettersson et al.). TC incidence for boys undergoing orchiopexy for UDT was assessed based on the age at orchiopexy (0-6 years, 7-9 years, 10-12 years, 13-15 years). The incidence of TC in each age cohort was calculated and used to determine the number needed to treat (NNT) for each age group using assumptions based on published TC outcomes.

RESULT(S): For an index patient ≤ 6 years, 372 boys need to undergo orchiopexy to prevent a single case of TC, 1488 boys to prevent exposure to TC therapy beyond radical orchiectomy, and 5315 boys to prevent a single TC-related death compared to treatment at an older age.

CONCLUSION(S): While there is evidence supporting benefits of early orchiopexy, the NNT to affect TC outcomes is very high. Even those with delayed orchiopexies have low risk for TC poor outcomes. This information can be used when counseling patients and families faced with UDT about the risks related to TC, especially with comorbidities.

PMC Identifier

31781896 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31781896>]

Author NameID

Saltzman, Amanda F.; ORCID: <http://orcid.org/0000-0002-3338-1116>

Institution

(Higgins, Saltzman) Department of Urology, University of Kentucky, KY, 800 Rose Street, Lexington 40536, United States (Smith, Gao) Department of Pediatrics, University of Colorado School of Medicine and University of Colorado Cancer Center, CO, Aurora, United States (Wilcox, Cost, Saltzman) Division of Urology, Department of Surgery, University of Colorado School of Medicine and Children's Hospital Colorado, CO, Aurora, United States

Publisher

NLM (Medline)

Year of Publication

2019

163.

Maternal rheumatoid arthritis and systemic lupus erythematosus and risk of cryptorchidism and hypospadias in boys: a Danish nationwide study.

Knudsen S.S., Bech B.H., Deleuran B.W., Ramlau-Hansen C.H., Arendt L.H.

Embase

Rheumatology (Oxford, England). (no pagination), 2019. Date of Publication: 18 Nov 2019.

[Article]

AN: 629888578

OBJECTIVES: RA and SLE are the most prevalent autoimmune rheumatic diseases affecting young women. Both diseases are characterized by systemic inflammation that may affect placental function and fetal development during pregnancy, and both diseases are associated with adverse pregnancy and child outcomes. We investigated the associations between maternal RA or SLE and the two genital malformations, cryptorchidism and hypospadias.

METHOD(S): In this nationwide register-based study including all male singleton live births in Denmark from 1995 to 2016, we assessed the occurrence of cryptorchidism and hypospadias according to the prenatal disease-state of the mothers. Using Cox proportional hazards models we calculated adjusted hazard ratios, accounting for varying age at diagnosis.

RESULT(S): Among 690 240 boys, 1026 had a mother with RA and 352 had a mother with SLE. We found adjusted hazard ratios of 1.72 (95% CI: 1.15; 2.57) for cryptorchidism among boys born to mothers with RA and 1.46 (95% CI: 0.69; 3.06) for boys born to mothers with SLE, compared

with the general population. As the number of hypospadias cases was low, multivariate analysis was not feasible. The crude hazard ratios were 0.51 (95% CI: 0.16; 1.58) and 1.00 (95% CI: 0.25; 4.03) for RA and SLE, respectively.

CONCLUSION(S): Boys born to mothers with RA had higher risk of cryptorchidism, compared with unexposed boys. Boys born to mothers with SLE showed a similar tendency, however with less precision of the estimate. No conclusion could be reached on the risk of hypospadias, due to the low number of events.

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PMC Identifier

31740964 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31740964>]

Institution

(Knudsen, Bech, Ramlau-Hansen) Department of Public Health, Aarhus University, Aarhus

(Knudsen, Deleuran) Department of Rheumatology, Aarhus University Hospital, Aarhus

(Deleuran) Department of Biomedicine, Aarhus University, Aarhus

(Arendt) Department of Gynaecology and Obstetrics, Horsens Regional Hospital, Horsens, Denmark

Publisher

NLM (Medline)

Year of Publication

2019

164.

The Fertility Potential Is Compromised in About 20-25% of Non-Syndromic Cryptorchid Boys Despite Orchidopexy Within the First Year of Life.

Hildorf S., Clasen-Linde E., Cortes D., Fossum M., Thorup J.

Embase

The Journal of urology. (pp 101097JU0000000000000615), 2019. Date of Publication: 23 Oct 2019.

[Article]

AN: 629661135

PURPOSE: One of the concerns regarding cryptorchidism is the risk of impaired fertility. Current guidelines recommend orchidopexy for cryptorchidism between 6 to 12 months to optimize the fertility outcome. The aim of this study was to evaluate the fertility potential of boys with non-syndromic cryptorchidism who underwent orchidopexy within the recommended age in order to clarify the need for eventual supplemental treatment modalities. **MATERIALS AND METHODS:** A retrospective evaluation of mini-puberty hormones (follicle stimulating hormone, luteinizing hormone, and inhibin B) and testicular biopsies from boys with cryptorchidism who underwent early orchidopexy within the first year, between 2010 and 2019, was performed. Based on histological examination, the germ cell number and the type A dark (Ad) spermatogonia number per seminiferous tubule cross-section (G/T and AdS/T, respectively) were analyzed in relation to normal values.

RESULT(S): 25% (83/333) of the boys with non-syndromic cryptorchidism (21% bilateral cryptorchidism) had reduced G/T. 21% of the boys (70/333) had low serum inhibin B. Of these boys having low serum inhibin B, 46% (32/70) boys had decreased G/T and 33% (23/70) boys decreased AdS/T (<0.01). Totally, 23% (75/333) of the boys had no Ad spermatogonia present. **CONCLUSION(S):** Despite early and successful orchidopexy 20-25% of boys with cryptorchidism may risk later infertility based on hormonal and histological data. Blood test and testicular biopsies are mandatory to identify boys with a high risk of infertility where additional treatment modalities and follow-up may be needed.

PMC Identifier

31642739 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31642739>]

Institution

(Hildorf, Fossum, Thorup) Department of Pediatric Surgery, Copenhagen University Hospital Rigshospitalet, Surgical Clinic C, Denmark (Clasen-Linde) Department of Pathology, Copenhagen University Hospital Rigshospitalet, Denmark

(Cortes, Fossum, Thorup) Faculty of Health and Medical Sciences, University of Copenhagen, Denmark

(Cortes) Section of Endocrinology, Department of Pediatrics, Copenhagen University Hospital Hvidovre, Denmark

Publisher

NLM (Medline)

Year of Publication

2019

165.

Gonadotropin treatment for male partial congenital hypogonadotropic hypogonadism in Chinese patients.

Hao M., Nie M., Yu B.-Q., Gao Y.-J., Wang X., Ma W.-L., Huang Q.-B., Zhang R., Mao J.-F., Wu X.-Y.

Embase

Asian journal of andrology. (no pagination), 2019. Date of Publication: 23 Aug 2019.

[Article]

AN: 629180295

Partial congenital hypogonadotropic hypogonadism (PCHH) is caused by an insufficiency in, but not a complete lack of, gonadotropin secretion. This leads to reduced testosterone production, mild testicular enlargement, and partial pubertal development. No studies have shown the productivity of spermatogenesis in patients with PCHH. We compared the outcomes of gonadotropin-induced spermatogenesis between patients with PCHH and those with complete congenital hypogonadotropic hypogonadism (CCHH). This retrospective study included 587 patients with CHH who were treated in Peking Union Medical College Hospital (Beijing, China) from January 2008 to September 2016. A total of 465 cases were excluded from data analysis for testosterone or gonadotropin-releasing hormone treatment, cryptorchidism, poor compliance, or incomplete medical data. We defined male patients with PCHH as those with a testicular volume of ≥ 4 ml and patients with a testicular volume of < 4 ml as CCHH. A total of 122 compliant, noncryptorchid patients with PCHH or CCHH received combined human chorionic gonadotropin and human menopausal gonadotropin and were monitored for 24 months. Testicular size, serum luteinizing hormone levels, follicle-stimulating hormone levels, serum total testosterone levels, and sperm count were recorded at each visit. After gonadotropin therapy, patients with PCHH had a higher spermatogenesis rate (92.3%) than did patients with CCHH (74.7%). During 24-month combined gonadotropin treatment, the PCHH group took significantly less time to begin producing sperm compared with the CCHH group (median time: 11.7 vs 17.8 months, $P < 0.05$). In conclusion, after combined gonadotropin treatment, patients with PCHH have a higher spermatogenesis success rate and sperm concentrations and require shorter treatment periods for sperm production.

PMC Identifier

31464203 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=31464203>]

Institution

(Hao, Nie, Yu, Gao, Wang, Ma, Huang, Zhang, Mao, Wu) NHC Key laboratory of Endocrinolog, Peking Union Medical College Hospital; Department of Endocrinology, Peking Union Medical

College Hospital, Peking Union Medical College, Chinese Academy of Medical Sciences, Beijing 100730, China (Hao) Department of Endocrinology, First Affiliated Hospital of Harbin Medical University, Harbin 150001, China

Publisher

NLM (Medline)

Year of Publication

2019

166.

Prune Belly Syndrome. [Review]

Pomajzl AJ; Sankararaman S.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

StatPearls Publishing. 2019 01.

[Review]

UI: 31334968

Prune belly syndrome, also referred to as Eagle-Barrett syndrome or the triad syndrome, is a rare congenital disorder characterized by the triad of deficient abdominal musculature, cryptorchidism, and urinary tract abnormalities.[1] Children born with this condition present on a broad spectrum ranging from incompatibility with life to aging normally and having children of their own. The severity of renal dysplasia mostly determines the survival and prognosis among the survivors.[2] Perinatal mortality ranges between 10 to 25% in contemporary studies and directly correlates to the severity of pulmonary hypoplasia as a result of oligohydramnios from reduced fetal urine production from renal dysplasia and urinary tract abnormalities leading to Potter sequence.[3]

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Book Title

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1

Authors Full Name

Pomajzl, AJ; Sankararaman, Senthilkumar.

Institution

Pomajzl, AJ. Creighton University School of Medicine Sankararaman, Senthilkumar. University Hospitals Rainbow Babies & Children's Hospital, Cleveland

Publisher

StatPearls Publishing

Year of Publication

2019

167.

Kallmann Syndrome. [Review]

Sonne J; Lopez-Ojeda W.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

StatPearls Publishing. 2019 01.

[Review]

UI: 30855798

Kallmann Syndrome (KS) is a congenital form of hypogonadotropic hypogonadism (HH) that manifests with hypo- or anosmia. This decrease in gonadal function is due to a failure in the differentiation or migration of neurons that arise embryologically in the olfactory mucosa to take up residence in the hypothalamus serving as gonadotropin-releasing hormone (GnRH) neurons. A deficit in the GnRH hormone results in decreased levels of sex steroids leading to a lack of sexual maturity and the absence of secondary sexual characteristics. Typical diagnosis occurs when a child fails to begin puberty. The condition, first described in 1944, is a rare pediatric genetic disease that is estimated to affect 1 in 48,000 individuals [1]. Treatment involves life-long hormone replacement therapy. However, treatment for male infants may include early hormone treatment or surgery to correct undescended testicles [2]. Unfortunately, later in life, these patients have an increased risk for developing osteoporosis due to their decreased sex hormones production and are often prescribed Vitamin D supplementation and bisphosphonates [3]. Kallmann syndrome, like other HH conditions, is characterized by reproductive features centered around a lack of sexual maturation during the years of puberty. These signs can include a lack of testicular development as determined by testicular volume in men, and a failure to start menstruation (amenorrhoea) in women. Poorly defined secondary sexual characteristics can include a lack of pubic hair and underdeveloped mammary glands. Micropenis may also be

present in a small portion of male cases, while cryptorchidism or undescended testicles may have been present at birth. All of these traits are related to low levels of luteinizing hormone (LH) and follicle stimulating hormone (FSH), which consequentially results in low testosterone in males and estrogen and progesterone in women [2]. In addition to the reproductive deficits of HH conditions, there will also be the presence of other non-reproductive characteristics which are often defects of embryological origin. KS is defined by its additional presentation of anosmia or hyposmia. Approximately 60% of patients with GnRH deficiency present with an impaired sense of smell and could be identified as having KS, cleft palate and lip, hypodontia and cleft hand or foot are also frequently present along with unilateral renal agenesis [4]. Cerebral impairments may also be present, including central hearing impairment, mirror movements of the hands (synkinesis) and ataxia. Color-blindness and ocular window defects have also been observed [5][6].

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Book Title

StatPearls

Version ID

1

Authors Full Name

Sonne, James; Lopez-Ojeda, Wilfredo.

Institution

Sonne, James. University of Central Florida Lopez-Ojeda, Wilfredo. Kaiser Permanente School of Medicine

Publisher

StatPearls Publishing

Year of Publication

2019

168.

Cryptorchidism. [Review]

Leslie SW; Sajjad H; Villanueva CA.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

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[Review]

UI: 29261861

Cryptorchidism is the absence of at least one testicle from the scrotum. It is the most common birth defect involving the male genitalia. About 3% of full-term and 30% of premature male infants are born with one or both testicles undescended. Approximately 80% of cryptorchid testes descend by the third month of life. This makes the true incidence around 1%. [1] Cryptorchidism may occur on one or both sides, but more commonly affects the right testicle. The testicle may be anywhere along the "path of descent," such as: [2]: Located high in the retroperitoneal abdomen to the inguinal ring. In the inguinal canal. Ectopic from the path of descent. Hypoplastic. Dysgenetic. Missing or Absent. Unilateral (two-thirds). The undescended testicle can usually be palpated in the inguinal canal. In a minority of patients, the missing testicle may be located in the abdomen or be nonexistent. Undescended testicles are associated with decreased fertility (bilateral cases), increased testicular germ cell tumors (overall risk under 1%), testicular torsion, inguinal hernias, and psychological problems. Without surgical correction, an undescended testicle may descend during the first three months of life. To reduce risks, undescended testes may be brought into the scrotum with an orchiopexy. Cryptorchidism, hypospadias, testicular cancer, and poor semen quality make up testicular dysgenesis syndrome (TDS). This syndrome is thought to be due to harmful environmental factors that disrupt embryonal programming and gonadal development during fetal life.

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Book Title

StatPearls

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1

Authors Full Name

Leslie, Stephen W.; Sajjad, Hussain; Villanueva, Carlos A..

Institution

Leslie, Stephen W.. Creighton University Medical Center Sajjad, Hussain. RMU and Allied Hospitals

Villanueva, Carlos A.. Children's Hospital and Medical Center

Publisher

StatPearls Publishing

Year of Publication

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169.

Chlordecone exposure and risk of congenital anomalies: the Timoun Mother-Child Cohort Study in Guadeloupe (French West Indies).

Rouget F; Kadhel P; Monfort C; Viel JF; Thome JP; Cordier S; Multigner L.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Environmental Science & Pollution Research. 2019 Aug 03.

[Journal Article]

UI: 31376129

Chlordecone is an organochlorine pesticide that was extensively used to control the banana root borer population in the French West Indies until 1993. Its persistence in soil has led to widespread pollution of the environment, and human beings, including pregnant women, are still exposed to this chemical. High levels of exposure to chlordecone during gestation have been shown to cause congenital anomalies, including undescended testes in rodents. We assessed the associations between chlordecone concentrations in maternal and cord plasma and the risk of congenital anomalies in the Timoun Mother-Child Cohort Study (2004-2007) that included 1068 pregnant women in Guadeloupe. Odds ratios were estimated using unconditional logistic regression analysis, controlling for confounding factors. The median plasma concentrations in maternal and cord plasma were 0.39 µg/L and 0.20 µg/L, respectively. Thirty-six children were diagnosed with malformations according to the European Registration of Congenital Anomalies guidelines and 25 with undescended testes. There was no association between maternal or cord plasma concentration of chlordecone and the risk of overall malformations nor undescended testes. These results suggest that prenatal exposure to the currently observed environmental levels of chlordecone in French West Indies does not increase the risk of birth defects.

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1

Status

Publisher

Author NameID

Rouget, Florence; ORCID: <http://orcid.org/0000-0001-5514-0893>

Authors Full Name

Rouget, Florence; Kadhel, Philippe; Monfort, Christine; Viel, Jean Francois; Thome, Jean Pierre; Cordier, Sylvaine; Multigner, Luc.

Institution

Rouget, Florence. CHU de Rennes, Univ Rennes, Inserm, EHESP, Irset (Institut de recherche en sante, environnement et travail) - UMR_S 1085, F-35000, Rennes, France.

florence.rouget@inserm.fr. Kadhel, Philippe. CHU de Pointe-a-Pitre, Univ Antilles, Univ Rennes, Inserm, EHESP, Irset (Institut de recherche en sante, environnement et travail) - UMR_S 1085, F-97110, Pointe-a-Pitre, France.

Monfort, Christine. Univ Rennes, Inserm, EHESP, Irset (Institut de recherche en sante, environnement et travail) - UMR_S 1085, F-35000, Rennes, France.

Viel, Jean Francois. CHU de Rennes, Univ Rennes, Inserm, EHESP, Irset (Institut de recherche en sante, environnement et travail) - UMR_S 1085, F-35000, Rennes, France.

Thome, Jean Pierre. LEAE-CART (Laboratoire d'Ecologie Animale et d'Ecotoxicologie-Centre de Recherche Analytique et Technologique), Universite de Liege, B-4000, Liege, Belgium.

Cordier, Sylvaine. Univ Rennes, Inserm, EHESP, Irset (Institut de recherche en sante, environnement et travail) - UMR_S 1085, F-35000, Rennes, France.

Multigner, Luc. Univ Rennes, Inserm, EHESP, Irset (Institut de recherche en sante, environnement et travail) - UMR_S 1085, F-35000, Rennes, France.

Year of Publication

2019

170.

The impact of primary location and age at orchiopexy on testicular atrophy for congenital undescended testis.

Tseng CS; Huang KH; Kuo MC; Hong CH; Chen CH; Lu YC; Huang CY; Pu YS; Chang HC; Chiang IN.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Scientific Reports. 9(1):9489, 2019 07 01.

[Journal Article]

UI: 31263127

In this study, we investigated post-orchiopexy testicular growth of undescended testes (UDTs) at different primary locations and determined the risk factors for testicular atrophy (TA). We conducted a retrospective chart review of boys who had undergone orchiopexy for UDTs during January 2001-December 2013. Patient profile, age at operation, primary UDT location, and

testicular volume were noted. TA was defined as $\geq 50\%$ loss of volume after orchiopexy. The primary endpoints were testicular growth and TA after orchiopexy. The secondary endpoint was risk factors for TA. In total, 182 boys had undergone regular ultrasonography; the median follow-up period was 34 months. Among 230 UDTs, 18 (7.8%) atrophic testicles were identified within a median interval of 13 months after orchiopexy. TA rates were 3.3% (1/30), 6.9% (12/173), and 18.5% (5/27) in primary suprascrotal, canalicular, and above-inguinal UDTs, respectively. The survival probability of UDT was 91%, 92% and 100% when orchiopexy was performed in age ≤ 1 year, $1 < \text{age} \leq 2$ years, and 100% in age > 2 years, respectively. Multivariate analysis revealed that inguinal and above-inguinal UDTs (hazard ratio [HR] 11.76, 95% confidence interval [CI] 1.55-89.33, $p = 0.017$) and genetic or endocrine disorders (HR 3.19, 95% CI 1.19-8.56, $p = 0.021$) were the risk factors for TA, but not age at operation, premature birth, and laterality. Thus, TA incidence was higher when patients had high primary testicular locations. Early orchiopexy before two years of age may be associated with higher TA risk, while most testicles have promising growth after orchiopexy.

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1

Status

In-Process

Author NameID

Tseng, Chi-Shin; ORCID: <https://orcid.org/0000-0002-1971-8552>

Authors Full Name

Tseng, Chi-Shin; Huang, Kuo-How; Kuo, Ming-Chieh; Hong, Chung-Hung; Chen, Chung-Hsin; Lu, Yu-Chuan; Huang, Chao-Yuan; Pu, Yeong-Shiau; Chang, Hong-Chiang; Chiang, I-Ni.

Institution

Tseng, Chi-Shin. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan. Huang, Kuo-How. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Kuo, Ming-Chieh. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Hong, Chung-Hung. Graduate Institute of Electronics Engineering, National Taiwan University, Taipei, Taiwan.

Chen, Chung-Hsin. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Lu, Yu-Chuan. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Huang, Chao-Yuan. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Pu, Yeong-Shiau. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Chang, Hong-Chiang. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan.

Chiang, I-Ni. Department of Urology, National Taiwan University Hospital, National Taiwan University, Taipei, Taiwan. inichiang@gmail.com.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6602959>

Year of Publication

2019

171.

Identification of three novel SRD5A2 mutations in Chinese patients with 5alpha-reductase 2 deficiency.

Cheng T; Wang H; Han B; Zhu H; Yao HJ; Zhao SX; Zhu WJ; Zhai HL; Chen FG; Song HD; Cheng KX; Liu Y; Qiao J.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Asian Journal of Andrology. 21(6):577-581, 2019 Nov-Dec.

[Journal Article. Research Support, Non-U.S. Gov't]

UI: 31031332

In this study, we investigated the genetics, clinical features, and therapeutic approach of 14 patients with 5alpha-reductase deficiency in China. Genotyping analysis was performed by direct sequencing of PCR products of the steroid 5alpha-reductase type 2 gene (SRD5A2). The 5alpha-reductase activities of three novel mutations were investigated by mutagenesis and an in vitro transfection assay. Most patients presented with a microphallus, variable degrees of hypospadias, and cryptorchidism. Eight of 14 patients (57.1%) were initially reared as females and changed their social gender from female to male after puberty. Nine mutations were identified in the 14 patients. p.G203S, p.Q6X, and p.R227Q were the most prevalent mutations. Three mutations (p.K35N, p.H162P, and p.Y136X) have not been reported previously. The nonsense mutation p.Y136X abolished enzymatic activity, whereas p.K35N and p.H162P retained partial enzymatic activity. Topical administration of dihydrotestosterone during infancy or early childhood

combined with hypospadias repair surgery had good therapeutic results. In conclusion, we expand the mutation profile of SRD5A2 in the Chinese population. A rational clinical approach to this disorder requires early and accurate diagnosis, especially genetic diagnosis.

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1

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Authors Full Name

Cheng, Tong; Wang, Hao; Han, Bing; Zhu, Hui; Yao, Hai-Jun; Zhao, Shuang-Xia; Zhu, Wen-Jiao; Zhai, Hua-Ling; Chen, Fu-Guo; Song, Huai-Dong; Cheng, Kai-Xiang; Liu, Yang; Qiao, Jie.

Institution

Cheng, Tong. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China. Wang, Hao. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Han, Bing. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Zhu, Hui. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Yao, Hai-Jun. Department of Urology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Zhao, Shuang-Xia. Central Laboratory, Clinical Research Center, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Zhu, Wen-Jiao. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Zhai, Hua-Ling. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Chen, Fu-Guo. Department of Plastic Surgery, Research Center of Tissue Engineering, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Song, Huai-Dong. Central Laboratory, Clinical Research Center, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Cheng, Kai-Xiang. Department of Plastic Surgery, Research Center of Tissue Engineering, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Liu, Yang. Department of Plastic Surgery, Research Center of Tissue Engineering, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

Qiao, Jie. Department of Endocrinology, Shanghai Ninth People's Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai 200011, China.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6859664>

Year of Publication

2019

172.

Evaluation and Management of the Undescended Testis in Puerto Rico: A Single Surgeon's 10 Years of Experience.

Soto-Palou FG; Escudero-Chu K; Pineyro-Ruiz C; Jorge JC; Perez-Brayfield M.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Puerto Rico Health Sciences Journal. 38(4):269-271, 2019 12.

[Journal Article. Research Support, N.I.H., Extramural]

UI: 31935315

OBJECTIVE: Cryptorchidism is an abnormality of the male genitourinary tract in which one or both testes fail to descend into the scrotum. The American Urological Association (AUA) clinical guidelines for the evaluation and treatment of cryptorchidism were recently published. We reviewed our experience with the evaluation and management of our patients and examined our findings with respect to the AUA and European Association of Urology (EAU) guidelines.

METHODS: Data were obtained from pediatric patients who underwent a surgical intervention for an undescended testis from 2007 through 2017 at HIMA Hospital and the University Pediatric Hospital (both in Puerto Rico); all the surgeries were performed by the same surgeon. A total of 754 patients were identified; 142 patients were excluded due to lack of follow-up data (N = 612). The data obtained included age, testes locations, radiologic and surgical findings, and postoperative results.

RESULTS: At their initial evaluations, a large proportion of the patients (46.4%) came accompanied with radiographic imaging. These findings were consistent with those of the

physical examination in 58.5% of the patients and with the surgical findings in 63.1% (sensitivity 77.9%, specificity 45.8%). Our data showed that the median referral age was 24 months, which suggests that there was a significant delay in diagnosis. At the time of surgery, the average age of the patients who required an orchiectomy was 3.93 years, while those who underwent an orchiopexy had an average age of 3.28 years.

CONCLUSION: Our data reveal that, despite its lack of usefulness, radiologic imaging continues to be included in the diagnostic workups of children newly identified with cryptorchidism in Puerto Rico. In addition, and contrary to the guidelines, there tends to be a significant delay in treatment with surgical intervention. It is important to continue to educate our referring physicians on the AUA and EUA guidelines in order to create awareness and encourage the proper diagnostic and treatment approach for cryptorchidism.

Version ID

1

Status

In-Process

Authors Full Name

Soto-Palou, Francois G; Escudero-Chu, Karina; Pineyro-Ruiz, Coriness; Jorge, Juan Carlos; Perez-Brayfield, Marcos.

Institution

Soto-Palou, Francois G. Urology Section, Department of Surgery, School of Medicine, University of Puerto Rico Medical Sciences Campus, San Juan, PR. Escudero-Chu, Karina. Urology Section, Department of Surgery, School of Medicine, University of Puerto Rico Medical Sciences Campus, San Juan, PR.

Pineyro-Ruiz, Coriness. Department of Anatomy and Neurobiology, School of Medicine, University of Puerto Rico Medical Sciences Campus, San Juan, PR.

Jorge, Juan Carlos. Department of Anatomy and Neurobiology, School of Medicine, University of Puerto Rico Medical Sciences Campus, San Juan, PR.

Perez-Brayfield, Marcos. Urology Section, Department of Surgery, School of Medicine, University of Puerto Rico Medical Sciences Campus, San Juan, PR.

Year of Publication

2019

Testicular expression of long non-coding RNAs is affected by curative GnRHa treatment of cryptorchidism.

Hadziselimovic F; Verkauskas G; Vincel B; Stadler MB.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
Basic & Clinical Andrology. 29:18, 2019.

[Journal Article]

UI: 31890219

Background: Cryptorchidism is a frequent endocrinopathy in boys that has been associated with an increased risk of developing testicular cancer and infertility. The condition is curable by combined surgery and hormonal treatment during early pre-pubertal stages using gonadotropin releasing hormone agonist (GnRHa). However, whether the treatment also alters the expression of testicular long non-coding RNAs (lncRNAs) is unknown. To gain insight into the effect of GnRHa on testicular lncRNA levels, we re-analyzed an expression dataset generated from testicular biopsies obtained during orchidopexy for bilateral cryptorchidism.

Results: We identified EGFR-AS1, linc-ROR, LINC00221, LINC00261, LINC00282, LINC00293, LINC00303, LINC00898, LINC00994, LINC01121, LINC01553, and MTOR-AS1 as potentially relevant for the stimulation of cell proliferation mediated by GnRHa based on their direct or indirect association with rapidly dividing cells in normal and pathological tissues. Surgery alone failed to alter the expression of these transcripts.

Conclusion: Given that lncRNAs can cooperate with chromatin-modifying enzymes to promote epigenetic regulation of genes, GnRHa treatment may act as a surrogate for mini-puberty by triggering the differentiation of Ad spermatogonia via lncRNA-mediated epigenetic effects. Our work provides additional molecular evidence that infertility and azoospermia in cryptorchidism, resulting from defective mini-puberty cannot be cured with successful orchidopexy alone.

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1

Status

PubMed-not-MEDLINE

Author NameID

Verkauskas, Gilvydas; ISNI: 0000 0001 2243 2806 Verkauskas, Gilvydas; GRID: 0000 0001 2243 2806

Vincel, Beata; ISNI: 0000 0001 2243 2806

Vincel, Beata; GRID: 0000 0001 2243 2806

Stadler, Michael B; ISNI: 0000 0001 2110 3787

Stadler, Michael B; GRID: 0000 0001 2110 3787

Stadler, Michael B; ISNI: 0000 0001 2223 3006

Stadler, Michael B; GRID: 0000 0001 2223 3006

Authors Full Name

Hadziselimovic, Faruk; Verkauskas, Gilvydas; Vincel, Beata; Stadler, Michael B.

Institution

Hadziselimovic, Faruk. Cryptorchidism Research Institute, Children's Day Care Center, 4410 Liestal, Switzerland. Verkauskas, Gilvydas. 2Children's Surgery Centre, Faculty of Medicine, Vilnius University, 01513 Vilnius, Lithuania.

Vincel, Beata. 3Children's Surgery Centre, Clinic of Gastroenterology, Nephrourology and Surgery, Institute of Clinical Medicine, Faculty of Medicine, Vilnius University, Vilnius, Lithuania.

Stadler, Michael B. 4Friedrich Miescher Institute for Biomedical Research, Basel, Switzerland.

Stadler, Michael B. 5Swiss Institute of Bioinformatics, Basel, Switzerland.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6933710>

Other Abstract

Publisher: La cryptorchidie est. une endocrinopathie frequente chez les garcons. Elle est. associee a un risque eleve de cancer des testicules et d'infertilite. La cryptorchidie peut etre soignee par une therapie incluant une intervention chirurgicale et un traitement hormonal par l'agoniste de l'hormone GnRH. Alors que l'effet de la therapie sur l'expression des ARNm a ete analyse, ses consequences pour la transcription des longs ARNs non codants (ARNInc) testiculaires restent inconnues. Afin de mieux comprendre les effets du GnRHa sur les concentrations cellulaires des ARNInc dans le testicule, nous avons analyse des donnees d'expression d'ARN par sequencage (ARN-Seq) generees en utilisant des biopsies testiculaires obtenues dans le cadre d'une orchidopexie pour cryptorchidie bilaterale.; Language: French

Year of Publication

2019

174.

Age at Surgery and Outcomes of Undescended Testes at King Salman Armed Forces Hospital, Tabuk, Saudi Arabia.

Mohammad Alnoaiji MS; Alrashidi TN; Ghmaird AS; Alsalem SS; Alanazi MS; Albazei AI; Alenzi MO; Aljuhani MA; Alotaibi RS; Alanazi SA; Althomali AM; Almohammadi AM; Alshahrani EH. OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present Cureus. 11(12):e6413, 2019 Dec 18.

[Journal Article]

UI: 31886102

Objective The aim of this study was to investigate the age at diagnosis and surgery of undescended testes and patients' outcomes. **Methods** This is a retrospective study that reviewed the files of patients who underwent orchidopexy at the King Salman Armed Forces Hospital (KSAFH), Tabuk, Saudi Arabia (SA), between January 1, 2015, and March 30, 2019. All children from birth until 13 years old who were admitted within the specified time frame and underwent orchidopexy were included in this study. The gathered data were analyzed through the Statistical Package for Social Sciences software (SPSS, version 23; SPSS Inc., Chicago, IL, USA). **Results** A total of 175 patients were included in this study. The rate of orchidopexy at our institution was 12.2%. The median ages at diagnosis and surgery were 12 and 24 months, respectively. The median duration between diagnosis and surgery was eight months. The most common site of undescended testis was inguinal (80.6%). Bilateral undescended testes were recorded in 24.6% of cases, and 25.7% of cases were impalpable. The size of the undescended testis was average in half the cases, small in 44.6% and atrophic in 6.4% of cases. Postoperative complications were reported in 4.0% of cases. Cox regression analysis revealed that the age at diagnosis was a significant risk factor affecting the time of surgery. **Conclusion** The findings of this study revealed that most cases of undescended testes in Tabuk were operated beyond the age recommended by international guidelines. The age at diagnosis seems to significantly affect the time of surgery. Copyright © 2019, Mohammad Alnoaiji et al.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Mohammad Alnoaiji, Mohammad S; Alrashidi, Tahani N; Ghmaird, Asmaa S; Alsalem, Sarah S; Alanazi, Malak S; Albazei, Alanuad I; Alenzi, Maryam O; Aljuhani, Mastorah A; Alotaibi, Rana S; Alanazi, Sara A; Althomali, Aeshah M; Almohammadi, Ahad M; Alshahrani, Eid H.

Institution

Mohammad Alnoaiji, Mohammad S. Paediatric Surgery, King Salman Armed Forces Hospitals, Tabouk, SAU. Alrashidi, Tahani N. Pediatric Surgery, University of Tabuk, Tabuk, SAU.

Ghmaird, Asmaa S. Pediatrics, University of Tabuk, Tabuk, SAU.

Alsalem, Sarah S. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Alanazi, Malak S. Otolaryngology Head and Neck Surgery, University of Tabuk, Tabuk, SAU.
Albazei, Alanuad I. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Alenzi, Maryam O. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Aljuhani, Mastorah A. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Alotaibi, Rana S. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Alanazi, Sara A. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Althomali, Aeshah M. Pediatric Surgery, University of Tabuk, Tabuk, SAU.
Almohammadi, Ahad M. Otolaryngology Head and Neck Surgery, University of Tabuk, Tabuk, SAU.
Alshahrani, Eid H. Pediatric Surgery, University of Bisha, Bisha, SAU.

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<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6921998>

Year of Publication

2019

175.

The role of tumor size, ultrasonographic findings, and serum tumor markers in predicting the likelihood of malignant testicular histology.

Song G; Xiong GY; Fan Y; Huang C; Kang YM; Ji GJ; Chen JC; Xin ZC; Zhou LQ.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Asian Journal of Andrology. 21(2):196-200, 2019 Mar-Apr.

[Journal Article]

UI: 30648671

The clinical predictive factors for malignant testicular histology remain unclear because of the low prevalence. Therefore, the aim of this study was to investigate predictors of malignant histology for testicular masses and decide more testis-sparing surgeries before surgery. This retrospective study enrolled 325 consecutive testicular mass patients who underwent radical orchiectomy (310/325) or testicular preserving surgery (15/325) from January 2001 to June 2016. The clinicopathological factors, including tumor diameter, cryptorchidism history, ultrasound findings, serum alpha-fetoprotein, and human chorionic gonadotropin (HCG) levels, were collected

retrospectively for statistical analysis. A predictive nomogram was also generated to evaluate the quantitative probability. Among all patients, 247 (76.0%) were diagnosed with a malignant testicular tumor and 78 (24.0%) with benign histology. Larger tumor diameter (per cm increased, hazard ratio [HR] = 1.284, P = 0.036), lower ultrasound echo (HR = 3.191, P = 0.001), higher ultrasound blood flow (HR = 3.320, P < 0.001), and abnormal blood HCG (HR = 10.550, P < 0.001) were significant predictive factors for malignant disease in all testicular mass patients. The nomogram generated was well calibrated for all predictions of malignant probability, and the accuracy of the model nomogram measured by Harrell's C statistic (C-index) was 0.92. According to our data, the proportion of patients who underwent radical orchiectomy for benign tumors (24.0%) was much larger than generally believed (10.0%). Our results indicated that the diameter, ultrasonic echo, ultrasonic blood flow, and serum HCG levels could predict the malignancy in testicular mass patients.

Version ID

1

Status

MEDLINE

Authors Full Name

Song, Gang; Xiong, Geng-Yan; Fan, Yu; Huang, Cong; Kang, Yong-Ming; Ji, Guang-Jie; Chen, Jin-Chao; Xin, Zhong-Cheng; Zhou, Li-Qun.

Institution

Song, Gang. Department of Urology, Peking University First Hospital, Beijing 100034, China.

Song, Gang. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.

Xiong, Geng-Yan. Department of Urology, Peking University First Hospital, Beijing 100034, China.

Xiong, Geng-Yan. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.

Fan, Yu. Department of Urology, Peking University First Hospital, Beijing 100034, China.

Fan, Yu. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.

Huang, Cong. Department of Urology, Peking University First Hospital, Beijing 100034, China.

Huang, Cong. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.

Kang, Yong-Ming. Department of Urology, Peking University First Hospital, Beijing 100034, China.

Kang, Yong-Ming. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.

Kang, Yong-Ming. Department of Urology, Suining Central Hospital, Suining 629000, China.
Ji, Guang-Jie. Department of Urology, Peking University First Hospital, Beijing 100034, China.
Ji, Guang-Jie. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.
Chen, Jin-Chao. Department of Urology, Peking University First Hospital, Beijing 100034, China.
Chen, Jin-Chao. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.
Xin, Zhong-Cheng. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.
Xin, Zhong-Cheng. Department of Andrology, Peking University First Hospital, Beijing 100034, China.
Zhou, Li-Qun. Department of Urology, Peking University First Hospital, Beijing 100034, China.
Zhou, Li-Qun. Institute of Urology, Peking University, National Urological Cancer Center, Beijing 100034, China.
PMC Identifier
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6413548>
Year of Publication
2019

176.

Orchiopexy.
Anonymous.
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AORN Journal. 109(1):P10-P12, 2019 01.
[Journal Article]
UI: 30592503
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1
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MEDLINE
Year of Publication

177.

Aging and Senescence in Canine Testes.

Merz SE; Klopffleisch R; Breithaupt A; Gruber AD.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Veterinary Pathology. 56(5):715-724, 2019 09.

[Journal Article]

UI: 31060479

Senescent cells accumulate with age but tissue-based studies of senescent cells are limited to selected organs from humans, mice, and primates. Cell culture and xenograft studies have indicated that senescent cells in the microenvironment may play a role in tumor proliferation via paracrine activities. Dogs develop age-related conditions, including in the testis, but cellular senescence has not been confirmed. We hypothesized that senescent cells accumulate with age in canine testes and in the microenvironment of testicular tumors. We tested the expression of the established senescence markers gammaH2AX and p21 on normal formalin-fixed, paraffin-embedded testes from 15 young dogs (<18 months of age) and 15 old dogs (7-15 years of age) and correlated the findings with age-dependent morphological changes. A statistically significant age-dependent increase in the percentage of p21-expressing cells was observed for testicular fibroblasts (4-fold) and Leydig cells (8-fold). However, p21-expressing cells were still a rare event. In contrast, the percentage of gammaH2AX-positive cells did not increase with age. P21- and gammaH2AX-expressing cells were rare in the microenvironments of tumors. Age-dependent morphological changes included an increased mean number of Leydig cells per intertubular triangle (2.95-fold) and a decreased spermatogenesis score. To our surprise, no age-related changes were recorded for interstitial collagen content, mean tubular diameter, and epithelial area. Opposed to our expectations based on previous in vitro data, we did not identify evidence of a correlation between age-associated accumulation of senescent cells and testicular tumor development. Understanding the role of the microenvironment in senescence obviously remains a challenging task.

Version ID

1

Status

MEDLINE

Author NameID

Klopfleisch, Robert; ORCID: <https://orcid.org/0000-0002-6308-0568> Breithaupt, Angele; ORCID: <https://orcid.org/0000-0002-6373-5923>

Authors Full Name

Merz, Sophie E; Klopfleisch, Robert; Breithaupt, Angele; Gruber, Achim D.

Institution

Merz, Sophie E. 1 Institute of Veterinary Pathology, Freie Universitat, Berlin, Germany.

Klopfleisch, Robert. 1 Institute of Veterinary Pathology, Freie Universitat, Berlin, Germany.

Breithaupt, Angele. 1 Institute of Veterinary Pathology, Freie Universitat, Berlin, Germany.

Gruber, Achim D. 1 Institute of Veterinary Pathology, Freie Universitat, Berlin, Germany.

Year of Publication

2019

178.

High Transverse Scrotal Incision Orchiopexy for Undescended Testes.

Ali MS; Khan N; Uddin MB; Hossain MS; Mushtabshirah L.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Mymensingh Medical Journal: MMJ. 28(3):542-546, 2019 Jul.

[Journal Article]

UI: 31391424

Bianchi squire first described scrotal incision orchiopexy as an alternative to the traditional approach in the 1980s. In maximum premature and some mature infants, palpable testis is a common surgical problem. There are several surgical techniques applied to overcome this surgical problems like combined inguinal and scrotal incision or single high transverse scrotal incision. The goal of this study assessed single high transverse scrotal incision for the management of PUT as regards to evaluate operative time, postoperative success and final cosmetic results. One hundred twenty patients were managed at the Paediatric Surgery Department of Mymensingh Medical College Hospital, Mymensingh, Bangladesh with PUT during the period from 2015 to March 2018. We operated all cases between 6 to 12 months and excluded more than 12 months

of age and recurrent cases. This technique involves manipulation of the testis down to the scrotum so that it is secured between the thumb and index finger as a fixation is performed. After fixation of testis high transverse scrotal incision was given, all layers were separated. Then enter into inguinal canal by cutting of external ring. After dissecting the testis come down through the incision. After making dartos pouch through the same incision and orchidopexy done. All infants were followed-up at 1 month, 2 months and 6 months to detect operative times as well as position, testicular atrophy and the final cosmetic appearance. Patient age ranged from 6 months to 12 months. A total of 120 PUTs were operated upon in 100 patients. PUT was bilateral in 15 patients, right-sided in 55 cases and left-sided in 30 cases. Among 120 testes 40 testes were located distal to external inguinal ring (EIR), 70 testes were at internal inguinal ring (IIR) and 10 testes were in between EIR and IIR. A total of 100 patients were successfully placed within scrotum using a single incision. Operative time ranged from 20-36 minutes. There were no cases of testicular atrophy or ascent. The only complications were 3 wound infections (3%), which were successfully treated with antibiotics. Single high transverse scrotal incision was sufficient to deal with PUT especially, in young infants (age 6 months). The procedure results in shorter operative times, similar success and complication rates, and a more cosmetically appealing outcome compared to inguinal orchiopexy.

Version ID

1

Status

MEDLINE

Authors Full Name

Ali, M S; Khan, N; Uddin, M B; Hossain, M S; Mushtabshirah, L.

Institution

Ali, M S. Dr Mohammad Showkot Ali, Assistant Professor, Department of Paediatric Surgery, Mymensingh Medical College (MMC), Mymensingh, Bangladesh.

Year of Publication

2019

179.

Testicular Volume Changes in Laparoscopic Staged Fowler-Stephens Orchiopexy: Studying the Impact of Testicular Vessel Division.

Abdelhalim A; Chamberlin JD; Young I; Fahim M; Chuang KW; McAleer IM; Wehbi E; Stephany HA; Khoury AE.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Urology. 127:113-118, 2019 05.

[Journal Article]

UI: 30779890

OBJECTIVE: To study the effect of testicular vessel division on testicular volume during laparoscopic staged Fowler Stephens orchiopexy (LSFSO).

METHODS: Testicular dimensions were prospectively measured intraoperatively at both first (S1) and second stages (S2) of LSFSO, and with scrotal ultrasound 3-12 months postoperatively. Testicular volumes were compared to reference ranges. Volume changes were tracked with a change of >20% considered clinically significant.

RESULTS: A total of 52 nonpalpable testes treated with LSFSO between 2008 and 2018 were included in the study. After a median follow-up of 6.8 (3-91.3) months, 46 (88.5%) testes were palpable in a scrotal location without adjunctive procedures and 39 (75%) maintained vascular flow on duplex ultrasound. One testis retracted to an inguinal position and was successfully treated with inguinal orchiopexy for an overall success of 90.4% (47/52). Of 36 testes with intra- and postoperative testicular volume documentation, only 2 (5.6%) had significant volume loss after S1. Both testes had catch-up growth after S2. Eight (22.2%) testes had significant volume loss after S2. At follow-up, 24 (66.7%) testes were smaller than the mean for age, of which 20 (83.3%) were small at baseline. Only 41.7% of testes larger than mean for age at follow-up, were small at baseline (P=.02).

CONCLUSION: Significant testicular volume loss does not occur after testicular vessel division at S1, but expected in approximately 1 quarter of testes after S2. We propose that testicular atrophy after LSFSO is primarily due to defective testicular development and rarely due to vascular compromise during S2.

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Version ID

1

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Authors Full Name

Abdelhalim, Ahmed; Chamberlin, Joshua D; Young, Irene; Fahim, Miriam; Chuang, Kai-Wen; McAleer, Irene M; Wehbi, Elias; Stephany, Heidi A; Khoury, Antoine E.

Institution

Abdelhalim, Ahmed. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA; Department of Urology, Mansoura Urology and Nephrology Center, Mansoura University, Mansoura, Egypt. Electronic address: a_halim_2010@yahoo.com. Chamberlin, Joshua D. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. Young, Irene. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. Fahim, Miriam. Eastern Virginia Medical School, VA. Chuang, Kai-Wen. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. McAleer, Irene M. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. Wehbi, Elias. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. Stephany, Heidi A. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. Khoury, Antoine E. Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, CA. Year of Publication 2019

180.

Adrenal hypoplasia congenita in identical twins.

Al Amer AM; Al Rubaya KM; Alzahrani AS.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Saudi Medical Journal. 40(1):87-92, 2019 Jan.

[Journal Article. Twin Study]

UI: 30617386

We are presenting a monozygotic twin brothers presented at different ages with different presentations. Twin-A presented at age of 18 days with salt losing crisis. Investigations revealed high plasma renin with low-normal aldosterone. Adrenocorticotrophic hormone, stimulation test

revealed low 17-OH progesterone at 0 and 60 minutes. Adrenocorticotrophic hormone level and serum cortisol were normal, which excluded initial impression of congenital adrenal hyperplasia. He was diagnosed to have isolated primary hypoaldosteronism. At age of 18 months, he was noticed to have hyperpigmentation of lips and gum. Adrenal failure was suspected, and hydrocortisone was added. Twin-B presented at 9 years and 6 months of age with adrenal crisis. Both were having unilateral undescended testes. Adrenal hypoplasia congenita (AHC) was suspected after his twin's presentation. Molecular analysis for gene study for both of them revealed adrenal insufficiency, NR0B1 (DAX1) gene mutation. In conclusion, gene analysis is important for the diagnosis of AHC and for genetic counseling.

Version ID

1

Status

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Authors Full Name

Al Amer, Alia M; Al Rubaya, Khloud M; Alzahrani, Ali S.

Institution

Al Amer, Alia M. Unit of Pediatric Endocrinology and Diabetes, Maternity and Children Hospital, Dammam, Kingdom of Saudi Arabia. E-mail. Aliamalamer@gmail.com.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6452601>

Year of Publication

2019

181.

Scrotal Ultrasound Is Not Routinely Indicated in the Management of Cryptorchidism, Retractable Testes, and Hydrocele in Children.

Shields LBE; White JT; Peppas DS; Rosenberg E.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Lobal Pediatric Health. 6:2333794X19890772, 2019.

[Journal Article]

UI: 31803796

Cryptorchidism, or undescended testes, is the most common congenital genitourinary anomaly. A failure or delay of treatment may result in reduced fertility or an increased risk of testicular cancer. The American Urological Association (AUA) recommends that a scrotal ultrasound (SUS) not be performed in the preoperative management of cryptorchidism. This study investigated how likely pediatricians were to perform SUS despite the AUA guidelines. We retrospectively studied 243 patients referred to a single pediatric urology practice for clinically diagnosed testis pathology including undescended testis, hydrocele, and retractile testis over a 4-year period (January 1, 2015, to December 30, 2018). A total of 72 patients (29.6%) underwent a SUS ordered by their pediatrician prior to the pediatric urology visit. Pediatricians should be aware that SUS performed prior to pediatric urological evaluation does not alter management and is associated with a significant financial cost in patients with cryptorchidism or hydrocele.

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Version ID

1

Status

PubMed-not-MEDLINE

Author NameID

Shields, Lisa B E; ORCID: <https://orcid.org/0000-0002-1526-4063>

Authors Full Name

Shields, Lisa B E; White, Jeffrey T; Peppas, Dennis S; Rosenberg, Eran.

Institution

Shields, Lisa B E. Norton Healthcare, Louisville, KY, USA. White, Jeffrey T. Norton Healthcare, Louisville, KY, USA.

Peppas, Dennis S. Norton Healthcare, Louisville, KY, USA.

Rosenberg, Eran. Norton Healthcare, Louisville, KY, USA.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6876163>

Year of Publication

2019

182.

In-utero Exposure to Maternal Stressful Life Events and Risk of Cryptorchidism: The Raine Study.

Brauner EV; Hickey M; Hansen AM; Doherty DA; Handelsman DJ; Juul A; Hart R.
OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
Frontiers in Endocrinology. 10:530, 2019.

[Journal Article]

UI: 31428056

Cryptorchidism, registered at birth or later, is the most common birth defect in males in western countries, estimated to affect around 2-3% of newborn boys, declining to around 2% at 3 months. We have previously described a potential association between stressful life events (SLEs) in pregnancy and reduced semen quality and testosterone levels in adult offspring. Both outcomes are believed to share a common etiology with cryptorchidism thus increased risk of cryptorchidism in boys exposed to prenatal SLEs may be plausible. The risk of cryptorchidism associated with prenatal SLE amongst 1,273 male Generation 2 offspring was estimated using the Western Australian Pregnancy (Raine) Study. SLEs are discrete experiences that disrupt an individual's usual activities causing a life change and readjustment, such as death of a relative or friend, divorce, illness or job loss. Mothers prospectively reported SLEs, during pregnancy at gestational weeks (GW) 18 and 34 using a standardized 10-point questionnaire. A boy was diagnosed as cryptorchid if one or both testes was non-palpable in the scrotum and not able to be manipulated into the scrotum. Twenty-four (2%) cryptorchid boys were identified. Mean (standard deviation) of SLE exposures in GW34 was 1.1 (1.2) for non-cryptorchid boys and slightly higher 1.5 (1.8) for cryptorchid boys, similar differences were observed in GW18. Adjusted odds ratio [OR] and 95% confidence intervals (CI) for risk of cryptorchidism in early (18-weeks) and late gestation (34-weeks) according to prenatal SLE exposures were: 1.06 (95% CI: 0.77-1.45) and 1.18 (95% CI: 0.84-1.67), respectively. This is the first-time report on the possible relationships between exposure to early and late pregnancy SLEs and risk of cryptorchidism in a birth cohort. Prenatal SLE exposure was not associated with a statistically significant increase in the risk of cryptorchidism in male offspring. A small case population limits the statistical power of the study and future larger studies are required to evaluate this potential association.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Brauner, Elvira V; Hickey, Martha; Hansen, Ase Marie; Doherty, Dorota A; Handelsman, David J;
Juul, Anders; Hart, Roger.

Institution

Brauner, Elvira V. Department of Growth and Reproduction, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark. Brauner, Elvira V. The International Research and Research Training Centre in Endocrine Disruption of Male Reproduction and Child Health, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark.

Hickey, Martha. Department of Obstetrics and Gynaecology, University of Melbourne, Melbourne, VIC, Australia.

Hansen, Ase Marie. Department of Public Health, University of Copenhagen, Copenhagen, Denmark.

Hansen, Ase Marie. The National Research Centre for the Working Environment, Copenhagen, Denmark.

Doherty, Dorota A. Division of Obstetrics and Gynaecology, University of Western Australia, Perth, WA, Australia.

Handelsman, David J. ANZAC Research Institute, University of Sydney, Sydney, NSW, Australia.

Juul, Anders. Department of Growth and Reproduction, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark.

Juul, Anders. The International Research and Research Training Centre in Endocrine Disruption of Male Reproduction and Child Health, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark.

Hart, Roger. Division of Obstetrics and Gynaecology, University of Western Australia, Perth, WA, Australia.

Hart, Roger. Fertility Specialists of Western Australia, Bethesda Hospital, Claremont, WA, Australia.

PMC Identifier
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6688069>

Year of Publication
2019

183.

Postnatal Testicular Activity in Healthy Boys and Boys With Cryptorchidism. [Review]
Kuri-Hanninen T; Koskeniemi J; Dunkel L; Toppari J; Sankilampi U.
OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Frontiers in Endocrinology. 10:489, 2019.

[Journal Article. Review]

UI: 31396156

Cryptorchidism, or undescended testis, is a well-known risk factor for testicular cancer and impaired semen quality in adulthood, conditions which have their origins in early fetal and postnatal life. In human pregnancy, the interplay of testicular and placental hormones as well as local regulatory factors and control by the hypothalamic-pituitary (HP) axis, lead to testicular descent by term. The normal masculine development may be disrupted by environmental factors or genetic defects and result in undescended testes. Minipuberty refers to the postnatal re-activation of the HP-testicular (T) axis after birth. During the first weeks of life, gonadotropin levels increase, followed by activation and proliferation of testicular Leydig, Sertoli and germ cells. Consequent rise in testosterone levels results in penile growth during the first months of life. Testicular size increases and testicular descent continues until three to five months of age. Insufficient HPT axis activation (e.g., hypogonadotropic hypogonadism) is often associated with undescended testis and therefore minipuberty is considered an important phase in the normal male reproductive development. Minipuberty provides a unique window of opportunity for the early evaluation of HPT axis function during early infancy. For cryptorchid boys, hormonal evaluation during minipuberty may give a hint of the underlying etiology and aid in the evaluation of the later risk of HPT axis dysfunction and impaired fertility. The aim of this review is to summarize the current knowledge of the role of minipuberty in testicular development and descent.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Kuiri-Hanninen, Tanja; Koskeniemi, Jaakko; Dunkel, Leo; Toppari, Jorma; Sankilampi, Ulla.

Institution

Kuiri-Hanninen, Tanja. Department of Pediatrics, Kuopio University Hospital, Kuopio, Finland.

Koskeniemi, Jaakko. Research Centre for Integrative Physiology and Pharmacology, Institute of Biomedicine, University of Turku, Turku, Finland.

Koskeniemi, Jaakko. Department of Pediatrics, Turku University Hospital, Turku, Finland.

Dunkel, Leo. Barts and the London, William Harvey Research Institute, Queen Mary University of London, London, United Kingdom.

Toppari, Jorma. Research Centre for Integrative Physiology and Pharmacology, Institute of Biomedicine, University of Turku, Turku, Finland.

Toppari, Jorma. Department of Pediatrics, Turku University Hospital, Turku, Finland.

Sankilampi, Ulla. Department of Pediatrics, Kuopio University Hospital, Kuopio, Finland.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6663997>

Year of Publication

2019

184.

Trans-scrotal Incision Approach versus Traditional Trans-scrotal Incision Orchiopexy in Children with Cryptorchidism: A Randomized Trial Study.

Nazem M; Hosseinpour M; Alghazali A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Advanced Biomedical Research. 8:34, 2019.

[Journal Article]

UI: 31259163

Background: Although undescended testis (UDT) is the most prevalent malformation in male neonates, the best mode of UDT treatment remains controversial. This study aimed to compare trans-scrotal incision approach with traditional trans-scrotal incision orchiopexy in children suffering from cryptorchidism.

Materials and Methods: This single-blind randomized clinical trial was done on 100 children with UDT who needed surgery. The participants were alternately undergoing trans-scrotal incision orchiopexy (Group I) and traditional inguinal incision orchiopexy (Group II). The success rate and incidence of postoperative complications were evaluated 1 week and 1 month and 6 months after the operation in the two groups.

Results: Both the groups were similar in baseline characteristics including age and laterality ($P > 0.05$). There was no significant difference between the two groups in terms of the incidence of wound infection, testicular atrophy, testicular hypotrophy, and relapse ($P > 0.05$). In addition, the success rates were 98% in Group I and 94% in Group II ($P > 0.05$).

Conclusion: Both surgical methods have a high success rate, and there is no significant difference in the incidence of complications; however, in terms of beauty, satisfaction, and shortening the duration of surgery and the duration of hospitalization, trans-scrotal approach was more successful than the traditional method.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Nazem, Masoud; Hosseinpour, Mehrdad; Alghazali, Abdulhakim.

Institution

Nazem, Masoud. Department of Pediatrics Surgery, Isfahan University of Medical Sciences, Isfahan, Iran. Hosseinpour, Mehrdad. Department of Pediatrics Surgery, Isfahan University of Medical Sciences, Isfahan, Iran.

Alghazali, Abdulhakim. Department of General Surgery, Isfahan University of Medical Sciences, Isfahan, Iran.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6543866>

Year of Publication

2019

185.

Genes located in Y-chromosomal regions important for male fertility show altered transcript levels in cryptorchidism and respond to curative hormone treatment.

Gegenschatz-Schmid K; Verkauskas G; Stadler MB; Hadziselimovic F.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Basic & Clinical Andrology. 29:8, 2019.

[Journal Article]

UI: 31171972

Background: Undescended (cryptorchid) testes in patients with defective mini-puberty and low testosterone levels contain gonocytes that fail to differentiate normally, which impairs the development of Ad spermatogonia and ultimately leads to adult infertility. Treatment with the gonadotropin-releasing hormone agonist GnRHa increases luteinizing hormone and testosterone and rescues fertility in the majority of pathological cryptorchid testes. Several Y-chromosomal genes in the male-specific Y region (MSY) are essential for spermatogenesis, testis development

and function, and are associated with azoospermia, infertility and cryptorchidism. In this study, we analyzed the expression of MSY genes in testes with Ad spermatogonia (low infertility risk patients) as compared to testes lacking Ad spermatogonia (high infertility risk) before and after curative GnRHa treatment, and in correlation to their location on the Y-chromosome.

Results: Twenty genes that are up- or down-regulated in the Ad- group are in the X-degenerate or the ampliconic region, respectively. GnRHa treatment increases mRNA levels of 14 genes in the ampliconic region and decreases mRNA levels of 10 genes in the X-degenerate region.

Conclusion: Our findings implicate Y-chromosomal genes, including USP9Y, UTY, TXLNGY, RBMY1B, RBMY1E, RBMY1J and TSPY4, some of which are known to be important for spermatogenesis, in the curative hormonal treatment of cryptorchidism-induced infertility.

Version ID

1

Status

PubMed-not-MEDLINE

Author NameID

Verkauskas, Gilvydas; ISNI: 0000 0001 2243 2806 Verkauskas, Gilvydas; GRID: 0000 0001 2243 2806

Stadler, Michael B; ISNI: 0000 0001 2110 3787

Stadler, Michael B; GRID: 0000 0001 2110 3787

Stadler, Michael B; ISNI: 0000 0001 2223 3006

Stadler, Michael B; GRID: 0000 0001 2223 3006

Authors Full Name

Gegenschatz-Schmid, Katharina; Verkauskas, Gilvydas; Stadler, Michael B; Hadziselimovic, Faruk.

Institution

Gegenschatz-Schmid, Katharina. Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, 4410 Liestal, Switzerland. Verkauskas, Gilvydas. 2Children's Surgery Centre, Faculty of Medicine, Vilnius of University, 01513 Vilnius, Lithuania.

Stadler, Michael B. 3Friedrich Miescher Institute for Biomedical Research, Basel, Switzerland.

Stadler, Michael B. 4Swiss Institute of Bioinformatics, Basel, Switzerland.

Hadziselimovic, Faruk. Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, 4410 Liestal, Switzerland.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6545630>

Other Abstract

Publisher: La non descente des testicules chez les garçons cryptorchides qui présentent une mini-puberté déficiente et un taux réduit de testostérone (T) ont des gonocytes incapables de

se différencier normalement en spermatogonie Ad. Cette dernière entraîne une infertilité. Le traitement avec l'agoniste du GnRH (GnRHa) augmente les taux de LH et T et permet de sauvegarder la fertilité chez la majorité des testicules cryptorchides pathologiques. Plusieurs gènes du chromosome Y localisés dans la région spécifique du mâle (MSY) sont essentiels pour la spermatogenèse, ainsi que pour le développement et la fonction testiculaires, et sont associés à l'azoospermie, l'infertilité et la cryptorchidie. Dans cette étude, nous avons analysé l'expression des gènes dans la région MSY des testicules avec et sans spermatogonies Ad, avant et après traitement par GnRHa. Les résultats sont corrélés avec la localisation des gènes dans le chromosome Y.; Language: French

Year of Publication

2019

186.

Familial bilateral cryptorchidism is caused by recessive variants in RXFP2.

Ayers K; Kumar R; Robevska G; Bruell S; Bell K; Malik MA; Bathgate RA; Sinclair A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Medical Genetics. 56(11):727-733, 2019 Nov.

[Journal Article]

UI: 31167797

BACKGROUND: Cryptorchidism or failure of testicular descent is the most common genitourinary birth defect in males. While both the insulin-like peptide 3 (INSL3) and its receptor, relaxin family peptide receptor 2 (RXFP2), have been demonstrated to control testicular descent in mice, their link to human cryptorchidism is weak, with no clear cause-effect demonstrated.

OBJECTIVE: To identify the genetic cause of a case of familial cryptorchidism.

METHODS: We recruited a family in which four boys had isolated bilateral cryptorchidism. A fourth-degree consanguineous union in the family was reported. Whole exome sequencing was carried out for the four affected boys and their parents, and variants that segregated with the disorder and had a link to testis development/descent were analysed. Functional analysis of a RXFP2 variant in cell culture included receptor localisation, ligand binding and cyclic AMP (cAMP) pathway activation.

RESULTS: Genomic analysis revealed a homozygous missense variant in the RXFP2 gene (c.1496G>A .p.Gly499Glu) in all four affected boys and heterozygous in both parents. No other variant with a link to testis biology was found. The RXFP2 variant is rare in genomic databases and predicted to be damaging. It has not been previously reported. Functional analysis demonstrated that the variant protein had poor cell surface expression and failed to bind INSL3 or respond to the ligand with cAMP signalling.

CONCLUSION: This is the first reported genomic analysis of a family with multiple individuals affected with cryptorchidism. It demonstrates that recessive variants in the RXFP2 gene underlie familial cryptorchidism and solidifies the link between this gene and testicular descent in humans.

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1

Status

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Author NameID

Ayers, Katie; ORCID: <http://orcid.org/0000-0002-6840-3186>

Authors Full Name

Ayers, Katie; Kumar, Rakesh; Robevska, Gorjana; Bruell, Shoni; Bell, Katrina; Malik, Muneer A; Bathgate, Ross A; Sinclair, Andrew.

Institution

Ayers, Katie. Department of Paediatrics, University of Melbourne, Melbourne, Victoria, Australia
katie.ayers@mcri.edu.au drrakesh.pgi@gmail.com. Ayers, Katie. Murdoch Children's Research Institute, Parkville, Melbourne, Victoria, Australia.

Kumar, Rakesh. Paediatric Endocrinology and Diabetes Unit, Department of Paediatric Medicine, Post Graduate Institute of Medical Education and Research, Chandigarh, India
katie.ayers@mcri.edu.au drrakesh.pgi@gmail.com.

Robevska, Gorjana. Murdoch Children's Research Institute, Parkville, Melbourne, Victoria, Australia.

Bruell, Shoni. Florey Institute of Neuroscience and Mental Health, Parkville, Melbourne, Victoria, Australia.

Bruell, Shoni. Department of Biochemistry and Molecular Biology, University of Melbourne, Melbourne, Victoria, Australia.

Bell, Katrina. Murdoch Children's Research Institute, Parkville, Melbourne, Victoria, Australia.

Malik, Muneer A. Department of Paediatric Surgery, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India.

Bathgate, Ross A. Florey Institute of Neuroscience and Mental Health, Parkville, Melbourne, Victoria, Australia.

Bathgate, Ross A. Department of Biochemistry and Molecular Biology, University of Melbourne, Melbourne, Victoria, Australia.

Sinclair, Andrew. Department of Paediatrics, University of Melbourne, Melbourne, Victoria, Australia.

Sinclair, Andrew. Murdoch Children's Research Institute, Parkville, Melbourne, Victoria, Australia.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6860408>

Year of Publication

2019

187.

Total laparoscopic repair of Spigelian hernia with undescended testis.

Deshmukh SS; Kothari PR; Gupta AR; Dikshit VB; Patil P; Kekre GA; Deshpande A; Kulkarni AA; Hukeri A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Minimal Access Surgery. 15(3):265-267, 2019 Jul-Sep.

[Journal Article]

UI: 30618422

Spigelian hernia is very rare in the paediatric age group. We present the case of an 11-month-old male child who presented with left Spigelian hernia with the left undescended testis in its sac.

Hernia repair with orchidopexy was done using total laparoscopic approach. It is the first reported case of total laparoscopic repair of Spigelian hernia with undescended testis in the paediatric age group.

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Authors Full Name

Deshmukh, Shahaji Satish; Kothari, Paras R; Gupta, Abhaya R; Dikshit, Vishesh Balbhadra; Patil, Prashant; Kekre, Geeta Anil; Deshpande, Aditi; Kulkarni, Apoorva Achyut; Hukeri, Aboli.

Institution

Deshmukh, Shahaji Satish. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India. Kothari, Paras R. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Gupta, Abhaya R. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Dikshit, Vishesh Balbhadra. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Patil, Prashant. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Kekre, Geeta Anil. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Deshpande, Aditi. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Kulkarni, Apoorva Achyut. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

Hukeri, Aboli. Department of Paediatric Surgery, Lokmanya Tilak Municipal Medical College and General Hospital, Mumbai, Maharashtra, India.

PMC Identifier

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2019

188.

The relation between isolated micropenis in childhood with CAG and GGN repeat polymorphisms in the androgen receptor gene.

Tug E., Guntekin Ergun S., Ergun M.A., Dilek F.N., Percin E.F.

Embase

Turkish Journal of Medical Sciences. 48 (2) (pp 430-434), 2018. Date of Publication: 2018.

[Article]

AN: 621966988

Background/aim: In micropenis cases accompanied by external genital abnormalities such as hypospadias and cryptorchidism, infertility and spermatogenic failures have been reported to correlate with androgen receptor (AR) gene CAG and GGN repeat polymorphisms. While there is one study on isolated micropenis and CAG repeats, no study related to GGN repeats has been reported. We investigated the relation between CAG and GGN repeats in the AR gene with development of penis length in boys with isolated micropenis.

Material(s) and Method(s): A total of 24 Turkish boys with isolated micropenis (<-2.5 SD) and 64 healthy controls who had normal basal serum gonadotropin levels were examined. Genotyping was performed by DNA sequencing of the patients and controls.

Result(s): The distribution of CAG and GGN repeat lengths in our patients and controls was within the normal range and did not significantly differ between the patients and the controls.

Conclusion(s): CAG repeat length in the AR constitutes one of multiple genetic factors relevant to the development of isolated micropenis, and the expansion of this repeat can be detected as a likely modifying factor. Moreover, the interactions of other genes that may be involved in the etiology of isolated micropenis with CAG and GGN repeats have to be taken into consideration.

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Institution

(Tug, Guntekin Ergun, Ergun, Percin) Department of Medical Genetics, School of Medicine, Gazi University, Ankara, Turkey (Dilek) Dr. Sami Ulus Obstetrics Child Health and Illness Training and Research Hospital, Ankara, Turkey

Publisher

Turkiye Klinikleri (Talapapa Bulvary no. 102, Hamammonu 1 06230, Turkey)

Year of Publication

2018

Refining the phenotype associated with biallelic DNAJC21 mutations.

D'Amours G., Lopes F., Gauthier J., Saillour V., Nassif C., Wynn R., Alos N., Leblanc T., Capri Y., Nizard S., Lemyre E., Michaud J.L., Pelletier V.-A., Pastore Y.D., Soucy J.-F.

Embase

Clinical Genetics. 94 (2) (pp 252-258), 2018. Date of Publication: August 2018.

[Article]

AN: 623019542

Inherited bone marrow failure syndromes (IBMFS) are caused by mutations in genes involved in genomic stability. Although they may be recognized by the association of typical clinical features, variable penetrance and expressivity are common, and clinical diagnosis is often challenging.

DNAJC21, which is involved in ribosome biogenesis, was recently linked to bone marrow failure.

However, the specific phenotype and natural history remain to be defined. We correlate molecular data, phenotype, and clinical history of 5 unreported affected children and all individuals reported in the literature. All patients present features consistent with IBMFS: bone marrow failure, growth retardation, failure to thrive, developmental delay, recurrent infections, and skin, teeth or hair abnormalities. Additional features present in some individuals include retinal abnormalities, pancreatic insufficiency, liver cirrhosis, skeletal abnormalities, congenital hip dysplasia, joint hypermobility, and cryptorchidism. We suggest that DNAJC21-related diseases constitute a distinct IBMFS, with features overlapping Shwachman-Diamond syndrome and Dyskeratosis congenita, and additional characteristics that are specific to DNAJC21 mutations. The full phenotypic spectrum, natural history, and optimal management will require more reports.

Considering the aplastic anemia, the possible increased risk for leukemia, and the multisystemic features, we provide a checklist for clinical evaluation at diagnosis and regular follow-up.

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Author NameID

D'Amours G.; ORCID: <http://orcid.org/0000-0002-1068-3154>

Institution

(D'Amours, Nizard, Lemyre, Michaud, Soucy) Service de Genetique Medicale, CHU Sainte-Justine, Montreal, Canada (D'Amours) Faculte de Medecine, Universite de Montreal, Montreal, Canada

(Lopes, Saillour, Nassif, Michaud) Centre de Recherche, CHU Sainte-Justine, Montreal, Canada

(Lopes) Life and Health Sciences Research Institute (ICVS), School of Medicine, University of Minho, Braga, Portugal

(Lopes) ICVS/3B's - PT Government Associate Laboratory, Braga/Guimaraes, Portugal
(Gauthier, Soucy) Laboratoire de Diagnostic Moleculaire, CHU Sainte-Justine, Montreal, Canada
(Saillour, Michaud) Centre Integre de Genomique Clinique Pediatrique, Montreal, Canada
(Wynn) Blood and Marrow Transplant Unit, Royal Manchester Children's Hospital, Manchester, United Kingdom
(Alos) Service d'Endocrinologie, CHU Sainte-Justine, Montreal, Canada
(Alos, Nizard, Lemyre, Michaud, Pelletier, Pastore, Soucy) Departement de Pediatrie, Universite de Montreal, Montreal, Canada
(Leblanc) Departement d'Hematologie Pediatrique, CHU Robert-Debre, Paris, France
(Capri) Service de Genetique Clinique, CHU Robert-Debre, Paris, France
(Pelletier) Departement de Pediatrie, CHU Sainte-Justine, Montreal, Canada
(Pastore) Service d'Hematologie-Oncologie, CHU Sainte-Justine, Montreal, Canada
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190.

Efficacy and safety of human chorionic gonadotropin for treatment of cryptorchidism: A meta-analysis of randomised controlled trials.

Wei Y., Wang Y., Tang X., Liu B., Shen L., Long C., Lin T., He D., Wu S., Wei G.

Embase

Journal of Paediatrics and Child Health. 54 (8) (pp 900-906), 2018. Date of Publication: August 2018.

[Article]

AN: 621702995

Aim: Although human chorionic gonadotropin (hCG) has long been employed in the management of cryptorchidism, its safety and efficacy is still controversial. Hence, in the present study, we conducted a meta-analysis of the treatment of cryptorchidism using hCG.

Method(s): We searched the Medline, Embase, CINAHL, EBSCO, The Cochrane Library, China National Knowledge Infrastructure and WanFang databases. Data were extracted by two reviewers using the designed extraction form. Data up to July 2015 were obtained using the

terms 'cryptorchidism', 'chorionic gonadotropin' and 'randomised controlled trials'. All the publications were downloaded, and the respective authors were contacted for any further details and clarifications, if deemed necessary. The data analysis included randomised controlled trials that compared hCG with other hormone treatments offered to prepubescent males presenting with cryptorchidism. Testicular descent rate was used as the final positive outcome of the treatments offered. The software Review Manager (RevMan 5.3, The Cochrane Collaboration, London, UK) was used to review the management and data analysis. Risk ratios (RRs) with 95% confidence intervals (CIs) were pooled with a fixed effect model if no heterogeneity was present. Result(s): A total of seven trials satisfied the selection criteria. The overall quality of the studies downloaded from various databases was low. Data from these seven studies were divided into three subgroups depending on the design of the trials: Two studies compared hCG with a placebo, and three studies compared hCG with gonadotropin-releasing hormone (GnRH) in unilateral cryptorchidism, whereas two other studies compared hCG with GnRH in bilateral cryptorchidism. Analysis of these trials revealed no significant differences between the effectiveness of hCG treatment and GnRH treatment in bilateral (RR 0.05, 95% CI (-0.29-0.40), two trials, n = 104, P = 0.76) as well as unilateral cryptorchidism (RR 0.04, 95% CI (-0.12, 0.21), three trials, n = 81, P = 0.61). A meta-analysis of these studies showed that hCG treatment is not superior to placebo (RR 7.74, 95% CI (0.14-425.72), two trials, n = 31, P = 0.32). Conclusion(s): A meta-analysis of the seven studies led us to conclude that hCG treatment is no more effective than placebo, and there were no significant differences in the effectiveness of hCG versus GnRH treatment.

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Author NameID

Wu, Shengde; ORCID: <http://orcid.org/0000-0002-0083-561X>

Institution

(Wei, Wang, Tang, Liu, Shen, Long, Lin, He, Wu, Wei) Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China (Wei, Wang, Tang, Wu, Wei) Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, Chongqing, China

(Liu, Lin, He, Wu, Wei) Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing, China

(Shen) China International Science and Technology Cooperation Base of Child Development and Critical Disorders, Chongqing, China

(Long) Chongqing Key Laboratory of Pediatrics, Chongqing, China

Publisher

Blackwell Publishing

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2018

191.

Testicular Cancer in New Zealand (TCNZ) study: Protocol for a national case-control study.

Gurney J.K., Stanley J., McGlynn K., Richiardi L., Shaw C., Edwards R., Merriman T.R., Robson B., Koea J., McLeod M., Kennedy M.A., Sarfati D.

Embase

BMJ Open. 8 (8) (no pagination), 2018. Article Number: e025212. Date of Publication: 01 Aug 2018.

[Article]

AN: 623793948

Testicular cancer (TC) is by far the most common cancer to affect young men; however, the exposures that cause this disease are still poorly understood. Our own research has shown that Morimori have the highest rates of this disease in New Zealand - a puzzling observation, since internationally TC is most commonly a disease of men of European ancestry. These trends provide us with a unique opportunity: to learn more about the currently unknown exposures that cause TC, and to explain why Morimori have the highest rates of this disease in New Zealand. Using epidemiology and genetics, our experienced research team will conduct a nationwide study which aims to answer these internationally important questions. Aim of study The overall aim of the current national case-control study is to identify the key exposures in the development of TC in New Zealand, and explore which factors might explain the difference in the incidence of TC between Mori and non-Mori. Methods and analysis Outside of our own investigations into cryptorchidism, we still do not know which exposures are driving the significant incidence disparity between ethnic groups in NZ. The aim of the proposed research is to use a population-based case-control study to identify the key exposures in the development of TC in New Zealand. We will recruit 410 TC cases and 410 controls, and collect (1) environmental exposure data, via

interview and (2) genetic information, via genome-wide genotyping. Ethics and dissemination Ethical approval for this study was sought and received from the New Zealand Ministry of Health's Health and Disability Ethics Committee (reference # 17/NTA/248). Following a careful data interpretation process, we will disseminate the findings of this study to a wide and varied audience ranging from general academia, community groups and clinical settings, as well as to the participants themselves.

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Institution

(Gurney, Stanley, Sarfati) Cancer and Chronic Conditions (C3) Research Group, Department of Public Health, University of Otago, Wellington, New Zealand (McGlynn) Division of Cancer Epidemiology and Genetics, National Cancer Institute, Maryland, United States

(Richiardi) Cancer Epidemiology Unit, University of Turin, Turin, Italy

(Shaw, Edwards, Robson, McLeod) Department of Public Health, University of Otago, Wellington, New Zealand

(Merriman) Department of Biochemistry, University of Otago, Dunedin, New Zealand

(Koea) Department of Surgery, Waitemata District Health Board, Auckland, New Zealand

(Kennedy) Department of Pathology, University of Otago, Christchurch, New Zealand

Publisher

BMJ Publishing Group (E-mail: subscriptions@bmjgroup.com)

Year of Publication

2018

192.

Placental Weight and Risk of Cryptorchidism and Hypospadias in the Collaborative Perinatal Project.

Ghazarian A.A., Trabert B., Graubard B.I., Longnecker M.P., Klebanoff M.A., McGlynn K.A.

Embase

American Journal of Epidemiology. 187 (7) (pp 1354-1361), 2018. Date of Publication: 01 Jul 2018.

[Article]

AN: 623483853

Cryptorchidism and hypospadias are the most common congenital anomalies of the genitourinary tract in males, but their etiology remains unclear. Placental insufficiency has been suggested to be linked to both conditions. Placental weight is a commonly used proxy measure for placental insufficiency; thus, we examined placental weight and other placental characteristics in relation to cryptorchidism and hypospadias in the Collaborative Perinatal Project, a US mother-child cohort study. Pregnant women were recruited between 1959 and 1965. The analysis contrasted boys with cryptorchidism (n = 413) and boys with hypospadias (n = 145) with boys without cryptorchidism (n = 23,799) and boys without hypospadias (n = 22,326). Odds ratios and 95% confidence intervals were calculated using unconditional logistic regression. In categorical analyses in which the middle tertile was the referent, cryptorchidism was inversely associated with placental weight (odds ratio = 0.66, 95% confidence interval: 0.46, 0.95) among white boys and positively associated with the lowest tertile of placental weight among black boys (odds ratio = 1.70, 95% confidence interval: 1.11, 2.59). We conclude that lower placental weight may be related to risk of cryptorchidism. Further investigation of placental functioning may offer insights into the etiology of cryptorchidism.

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Institution

(McGlynn) National Cancer Institute, 9609 Medical Center Drive, Room 6-E446, Bethesda, MD 20892, United States (Ghazarian, Trabert, Graubard, McGlynn) Division of Cancer Epidemiology and Genetics, National Cancer Institute, Bethesda, MD, United States

(Ghazarian) Division of Cancer Control and Population Sciences, National Cancer Institute, Bethesda, MD, United States

(Longnecker) Epidemiology Branch, National Institute of Environmental Health Sciences, Research Triangle Park, NC, United States

(Klebanoff) Ohio Perinatal Research Network, Department of Pediatrics, College of Medicine, Ohio State University, Nationwide Children's Hospital, Columbus, OH, United States

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193.

Association between male genital anomalies and adult male reproductive disorders: a population-based data linkage study spanning more than 40 years.

Schneuer F.J., Milne E., Jamieson S.E., Pereira G., Hansen M., Barker A., Holland A.J.A., Bower C., Nassar N.

Embase

The Lancet Child and Adolescent Health. 2 (10) (pp 736-743), 2018. Date of Publication: October 2018.

[Article]

AN: 2001193103

Background: The male genital anomalies hypospadias and undescended testes have been linked to adult male reproductive disorders, testicular cancer, and decreased fertility. Few population-based studies have evaluated their effects on adult fertility outcomes and, in the case of undescended testes, the importance of early corrective surgery (orchidopexy).

Method(s): We did a population-based cohort study of all liveborn boys in Western Australia in 1970-99, and followed them up until 2016 via data linkage to registries for hospital admissions, congenital anomalies, cancer, and assisted reproductive technologies (ART). Study factors were hypospadias or undescended testes, and study outcomes were testicular cancer, paternity, and use of ART for male infertility. Cox regression was used to calculate hazard ratios (HRs) for the associations between genital anomalies and testicular cancer or paternity, and log-binomial regression was used to calculate relative risks (RRs) for the associations between genital anomalies and use of ART.

Finding(s): The cohort comprised 350 835 boys, of whom 2484 (0.7%) had been diagnosed with hypospadias and 7499 (2.1%) with undescended testes. There were 505 (0.1%) cases of testicular cancer, 109 471 (31.2%) men had fathered children, and 2682 (0.8%) had undergone fertility treatment with ART. Undescended testes was associated with a more than two times increase in risk of testicular cancer (HR 2.43, 95% CI 1.65-3.58) and hypospadias with an almost 40% increase (1.37, 0.51-3.67), although this increase was not significant. Both hypospadias and undescended testes were associated with a 21% reduction in paternity (adjusted HR 0.79 [95%

CI 0.71-0.89] for hypospadias and 0.79 [0.74-0.85] for undescended testes). Undescended testes was associated with a two times increase in use of ART (adjusted RR 2.26, 95% CI 1.58-3.25). For every 6 months' delay in orchidopexy, there was a 6% increase in risk of testicular cancer (HR 1.06, 95% CI 1.03-1.08), a 5% increase in risk of future use of ART (1.05, 1.03-1.08), and a 1% reduction in paternity (RR 0.99, 95% CI 0.98-0.99).

Interpretation(s): Undescended testes is associated with an increased risk of testicular cancer and male infertility, and decreased paternity. We provide new evidence to support current guidelines for orchidopexy before age 18 months to decrease the risk of future testicular cancer and infertility.

Funding(s): National Health and Medical Research Council and Sydney Medical School Foundation.

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Status

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Institution

(Schneuer, Nassar) Child Population and Translational Health Research, The Children's Hospital at Westmead Clinical School, The University of Sydney, NSW, Australia (Holland) Discipline of Child and Adolescent Health, The Children's Hospital at Westmead Clinical School, The University of Sydney, NSW, Australia

(Milne, Jamieson, Pereira, Hansen, Bower) Telethon Kids Institute, University of Western Australia, Perth, WA, Australia

(Pereira) School of Public Health, Curtin University, Perth, WA, Australia

(Barker) Department of Paediatric Surgery, Princess Margaret Hospital for Children, Perth, WA, Australia

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2018

Acquired undescended testis and possibly associated testicular torsion in children with cerebral palsy or neuromuscular disease.

Ito T., Matsui F., Fujimoto K., Matsuyama S., Yazawa K., Matsumoto F., Shimada K.

Embase

Journal of Pediatric Urology. 14 (5) (pp 402-406), 2018. Date of Publication: October 2018.

[Article]

AN: 2001094189

Introduction: Torsion of an undescended testis (UDT) associated with cerebral palsy (CP) and neuromuscular disease (NMD) is an uncommon condition that is not well recognized by primary care physicians or healthcare providers.

Objective(s): The objective of this study was to highlight the clinical importance of torsion of a UDT in children with CP and NMD.

Material(s) and Method(s): Eleven children with testicular torsion of a UDT operated on at the study institute between 1991 and 2015 were identified. The records of seven children (63.6%) associated with CP or NMD were retrospectively reviewed. Clinical findings of testicular torsion were assessed along with the treatment outcome and testicular salvageability.

Result(s): All seven children were not identified with a UDT by public health checkup for infant and young children. No children with CP or NMD had torsion of a descended testis during the present study period. Median age at surgery was 15 years (range, 1-20 years). The testis location was at the external inguinal ring in five patients, in the inguinal canal in one, and in the superficial inguinal pouch in one. Of the contralateral testes, four were a UDT, one was a retractile testis, and two were descended testes. Orchiectomy was performed in six patients (85.7%). In the remaining patients, the testis was preserved but became atrophic.

Discussion(s): This study demonstrated that children with CP or NMD may be affected with torsion of a UDT with peak at around puberty with the poor salvage rate, even if the testes appear descended in infancy and young children. Shortcomings of this study were the retrospective design and a small series of children undergoing surgery for torsion of a UDT.

Conclusion(s): Pediatric urologists need to educate primary care physicians and healthcare providers in the recognition of acquired UDTs and possibly associated testicular torsion in children with CP and NMD. Genital examination should be continued regularly until adolescence in these children to detect acquired UDT. These children should be referred to pediatric urologists to promote surgery as soon as the diagnosis of acquired UDT is carried out. It is believed that it is perhaps the best approach to prevent loss of the testis in children with CP and NMD.[Table presented]

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Author NameID

Matsui F.; ORCID: <http://orcid.org/0000-0002-7102-6115> Yazawa K.; ORCID:

<http://orcid.org/0000-0002-0408-717X>

Institution

(Ito, Matsui, Fujimoto, Matsuyama, Yazawa, Matsumoto, Shimada) Department of Urology,
Osaka Women's and Children's Hospital, Osaka, Japan

Publisher

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2018

195.

Description of mutation spectrum and polymorphism of Wilms' tumor 1 (WT1) gene in
hypospadias patients in the Indonesian population.

Diposarosa R., Pamungkas K.O., Sribudiani Y., Herman H., Suciati L.P., Rahayu N.S., Effendy
S.H.

Embase

Journal of Pediatric Urology. 14 (3) (pp 237.e1-237.e7), 2018. Date of Publication: June 2018.

[Article]

AN: 2000887745

Introduction: Hypospadias is one of the most common congenital anomalies of the penis.
Previous studies reported mutation of the Wilms' tumor 1 (WT1) gene as a cause of hypospadias.
The aim of this study is to describe the WT1 mutation spectrum and polymorphism in
hypospadias patients in Indonesia.

Material(s) and Method(s): DNA was isolated from 74 hypospadias patients at the Division of
Pediatric Surgery, Department of Surgery Hasan Sadikin Hospital. All exons in the WT1 gene
were amplified by a PCR method, followed by Sanger sequencing. Mutation analysis was
performed using BioEdit software and in silico analysis using Mutation Taster, Polymorphism
Phenotyping-2 (PolyPhen-2), and Sorting Intolerant from Tolerant (SIFT).

Result(s): DNA analysis results showed two types of heterozygous mutations in five subjects (Table), hence the frequency of WT1 mutations was 6.7% (10/148 allele). The first mutation was a missense mutation identified in twin boys. The second was a novel heterozygous alteration in the non-coding region nine bp upstream of exon 6 (c.366-9T>C), which was identified in three patients. One heterozygous polymorphism in the coding region of exon 7 (c.471A>G/rs16754) was identified in 10 subjects. This variant did not cause any change in amino acid products (silence polymorphism). Allele frequency for the G allele (mutant allele) and A allele (wild type) was 13.5% and 86.5%, respectively.

Discussion(s): WT1 is one of the best known hypospadias genes. The WT1 gene is involved in male genital development in the early and late periods of sex determination, and hence is known as a long-term expression gene in genitalia development. Mutation analysis of WT1 in a Chinese population identified that the WT1 mutation frequency was 4.4%. The WT1 mutation frequency identified in the present study was higher, at 6.7%. Coincidentally, research subjects with p.R158H variants were monozygotic twin siblings with midshaft hypospadias accompanied by undescended testis in one and penoscrotal hypospadias with micropenis in the other. The incidence of familial hypospadias in male siblings suffering from hypospadias was reported to be 9.6% in a study conducted by Sorensen et al. Moreover, in the present study polymorphism c.471A>G(rs16754) at exon 7 was identified heterozygously in 10 research subjects (minor allele frequency 13.5%).

Conclusion(s): WT1 mutations were identified in only a few cases of hypospadias and most of these were syndromic. This result implies that mutation of WT1 is not a common cause of hypospadias in the Indonesian population. [Table presented]

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Institution

(Diposarosa, Pamungkas) Department of Surgery, Pediatric Surgery Division, Hasan Sadikin Hospital, Faculty of Medicine, Padjadjaran University, Bandung, Indonesia (Sribudiani)
Department of Biochemistry and Molecular Biology, Faculty of Medicine, Padjadjaran University, Bandung, Indonesia

(Sribudiani, Effendy) Clinical Genetics Working Group, Faculty of Medicine, Padjadjaran University, Bandung, Indonesia

(Herman) Department of Surgery, Orthopedic Surgery Division, Hasan Sadikin Hospital, Faculty of Medicine, Padjadjaran University, Bandung, Indonesia

(Suciati, Rahayu) Laboratory of Genetic and Biology Molecular, Faculty of Medicine, Padjadjaran University, Bandung, Indonesia

(Effendy) Department of Pediatric, Hasan Sadikin Hospital, Faculty of Medicine, Padjadjaran University, Bandung, Indonesia

Publisher

Elsevier Ltd

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2018

196.

A novel technique to construct a neo-scrotum out of preputial skin for agenesis and underdeveloped scrotum.

Benson M., Hanna M.K.

Embase

Journal of Pediatric Urology. 14 (5) (pp 449.e1-449.e4), 2018. Date of Publication: October 2018.

[Article]

AN: 2000855090

Introduction: Congenital scrotal agenesis (CSA) is an extremely rare condition with fewer than 10 cases reported in the literature. These patients are often further complicated by undescended testicles as well as systemic developmental disorders. Herein, we report our experience and an innovative surgical technique for creation of a neo-scrotum in three children.

Material(s) and Method(s): We evaluated the records of three children in our database who were labeled as having an absent (1 patient) or severely underdeveloped scrotum (2 patients). Patients were aged 6, 14, and 16 months. All patients were uncircumcised, and the decision was made to create a neo-scrotum using the foreskin. The prepuce was harvested on a pedicle of dartos and transposed over the perineal cleft to create a neo-scrotal pouch. The flap was allowed to heal for 12-14 weeks, at which time the orchidopexy was performed.

Result(s): The follow-up to the surgery was 5 years, 3 years, and 6 months, respectively. There were no instances of flap necrosis, dehiscence, or wound infection. The three children had a capacious, aesthetically pleasing scrotal sac, which in all cases accommodated both testicles.

Conclusion(s): In our experience, a well-vascularized preputial skin flap rotated to the perineum based on its ventral dartos pedicle provides an excellent source of tissue for creation of a neo-

scrotum. We believe that the esthetic outcome of our technique rivals that of other reported techniques.

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Status

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Author NameID

Benson, Michael; ORCID: <http://orcid.org/0000-0002-8181-3083>

Institution

(Benson) Division of Urology, Rutgers New Jersey Medical School, Newark, NJ, United States

(Hanna) Department of Urology, New York-Presbyterian Weill Cornell Medical Center (MKH), New York, NY, United States

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197.

Tunica vaginalis flap for urethrocutaneous fistula repair after proximal and mid-shaft hypospadias surgery: A 12-year experience.

Pescheloché P., Parmentier B., Hor T., Chamond O., Chabaud M., Irtan S., Audry G.

Embase

Journal of Pediatric Urology. 14 (5) (pp 421.e1-421.e6), 2018. Date of Publication: October 2018.

[Article]

AN: 2000810910

Introduction: Fistulas are a common complication of hypospadias surgery; they are more frequent after mid-shaft and posterior hypospadias repair. Surgical treatment of fistula still remains challenging with a significant failure rate. The basic principle is to add layers between skin and neourethra in order to decrease the incidence of recurrent urethrocutaneous fistula (UCF). We report our experience of UCF repair using a vascularized tunica vaginalis flap (TVF) after posterior and mid-shaft hypospadias surgery.

Material(s) and Method(s): A retrospective review of all patients operated on using TVF for UCF in our institution between December 2005 and July 2017 was performed.

Result(s): Among 36 cases, TVF was used at a first attempt in 22 patients; 14 children had a prior attempt to close the fistula, and four of them had two surgeries before TVF repair. UCF was respectively penoscrotal (n = 3, 8%), posterior (n = 19, 53%), midshaft (n = 9, 25%) and anterior (n = 5, 14%). The size of the fistula was more than 5 mm in 26 patients. The UCF was treated successfully in every case after one single procedure. In the three children with two fistulas, both fistulas were successfully treated by the same TVF. After an average follow-up time of 45 months there was no recurrence of the initial UCF. In four cases of undescended testis, it was possible to dissect the flap through an inguinal incision and perform an orchidopexy in the same time. One patient presented a testicular atrophy after undescended testis surgery.

Discussion(s): Area review of published series shows excellent results in UCF repair including recurrent fistula (Table). TVF can aspire to some advantages with regard to a dartos flap (DF). First of all, a nearly 2.5-fold lower incidence of fistula after fistula repair with TVF than with DF (5.1% vs. 12.2%) has been shown. Secondly, TVF allows treating multiple fistulas, and can also be brought to the anterior part of the penis until the balano preputial furrow, allowing curing anterior fistula. Furthermore, it doesn't lead to aesthetic complications such as penile rotation or distal skin necrosis, which can occur during DF procedures.

Conclusion(s): TVF is a simple and reproductive technique for UCF repair, with a high success rate. The risk of testicular atrophy has to be considered in case of associated undescended testis surgery, and careful attention must be given to the TVF dissection. This technique should be considered as first choice treatment for any UCF. [Table presented]

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Embase

Institution

(Pescheloche, Parmentier, Hor, Chamond, Chabaud, Irtan, Audry) Department of Pediatric Surgery, APHP Hopital Armand Trousseau, Sorbonne Universites, Paris, France

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198.

Shear wave elastography evaluation of testes with non-communicating hydrocele in infants and toddlers: A preliminary study.

Kocaoglu C., Durmaz M.S., Sivri M.

Embase

Journal of Pediatric Urology. 14 (5) (pp 445.e1-445.e6), 2018. Date of Publication: October 2018.

[Article]

AN: 2000720028

Introduction: Shear wave elastography (SWE) is a new technology and non-invasive ultrasound device that can measure tissue rigidity and elasticity.

Objective(s): This controlled prospective study aimed to demonstrate using SWE whether there was a difference between the elasticity of testes with non-communicating (NC) hydrocele in infants and toddlers and elasticity of controls' testes without hydrocele, and to reveal quantitative values of elasticity reflecting histological findings.

Material(s) and Method(s): Testes of 37 cases at an average age of 6.32 months and diagnosed with NC hydrocele between December 2016 and April 2017 were evaluated for hydrocele and testicular volumes, and testes elasticity through ultrasonography and SWE.

Result(s): Of all cases, 15 had bilateral hydroceles, while 22 were diagnosed with unilateral hydrocele. Testes with NC hydrocele (n = 52) in infants and toddlers were compared with testes without hydrocele (n = 36) in controls. Median hydrocele volume of 52 testes with NC hydrocele was 5.0 cm³ (0.2-37). Median volume of testes with hydrocele was 0.6 cm³ (0.2-1.5) in the study group, and 0.5 cm³ (0.3-1) in controls (P = 0.577). Although median elastography values were measured as 1.67 m/s (1.29-2.59) and 10.0 kPa (2.1-23) in patients, those of controls were found to be 1.61 m/s (1.27-2.34) and 8.25 kPa (5.1-18.9) (P = 0.03, P = 0.005, respectively). While there was no between-group difference in testes volumes, a statistically significant difference was observed in SWE-derived quantitative data (Summary Table).

Discussion(s): As a novel elastographic method, SWE is used to track shear waves passing through tissues by quantifying the elasticity of structures and nodules, such as liver fibrosis, and to improve the characterization of breast and thyroid nodules. Shear wave elastography was assessed to be a beneficial ultrasonography tool to predict the histologic features of undescended testicles, which might replace testicular biopsy in the modern management of undescended testes. This study also quantitatively measured whether there was a change in testicular tissues with NC hydrocele through SWE, and found that SWE values of testes with NC hydrocele were

significantly higher compared with those of controls, despite the absence of a significant difference in testes volumes.

Conclusion(s): The present study confirmed that quantitative changes in testes elasticity can reliably be evaluated through SWE. Non-communicating hydrocele may be damaging to testicular tissues. More definitive results will be achieved with further comprehensive studies including larger patient populations. It is believed that the operation age of children with NC hydrocele can be re-evaluated in the future.[Table presented]

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Status

Embase

Author NameID

Kocaoglu C.; ORCID: <http://orcid.org/0000-0001-7111-3318>

Institution

(Kocaoglu) Department of Pediatric Surgery, University of Health Sciences Konya Education and Research Hospital, Meram Yeni Yol, Konya, Turkey (Durmaz, Sivri) Department of Radiology, University of Health Sciences Konya Education and Research Hospital, Meram Yeni Yol, Konya, Turkey

Publisher

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2018

199.

Association of in Utero Persistent Organic Pollutant Exposure with Placental Thyroid Hormones.

Li Z.-M., Hernandez-Moreno D., Main K.M., Skakkebaek N.E., Kiviranta H., Toppari J., Feldt-Rasmussen U., Shen H., Schramm K.-W., De Angelis M.

Embase

Endocrinology. 159 (10) (pp 3473-3481), 2018. Date of Publication: 01 Oct 2018.

[Article]

AN: 624189560

In utero exposure to persistent organic pollutants (POPs) can result in thyroid function disorder, leading to concerns about their impact on fetal and neonatal development. The associations between placental levels of various POPs and thyroid hormones (THs) were investigated. In a prospective Danish study initially established for assessing congenital cryptorchidism, 58 placenta samples were collected from mothers of boys born with (n =28) and without (n =30) cryptorchidism. The concentrations of polybrominated diphenyl ethers (PBDEs), polychlorinated biphenyls (PCBs), polychlorinated dibenzo-p-dioxins/furans (PCDD/Fs), organotin chemicals (OTCs), organochlorine pesticides (OCPs), T 4, T 3, and rT 3 were measured. The associations between placental THs and various POPs were analyzed using multiple linear regression. Five PBDEs, 35 PCBs, 14 PCDD/Fs, 3 OTCs, 25 OCPs, T 4, T 3, and rT 3 were measured. No correlation between THs and the odds of cryptorchidism was found. Several POPs were significantly associated with THs: (1) T 4 was inversely associated with BDEs 99, 100, I PBDE, and 2378-TeCDD, and positively associated with 1234678-HpCDF; (2) T 3 was positively associated with 2378-TeCDF and 12378-PeCDF; and (3) rT 3 was positively associated with PCB 81, 12378-PeCDF, and 234678-HxCDF, and inversely associated with tributyltin, I OTC, and methoxychlor. These results revealed that POP exposures were associated with TH levels in placenta, which may be a possible mechanism for the impacts of POP exposures on children's growth and development. This study provides new insight into the complexity of thyroid-disrupting properties of POPs. More research is needed to elucidate the biological consequences of POP exposures.

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Embase

Institution

(Li, Hernandez-Moreno, Schramm, De Angelis) Helmholtz Zentrum Munchen-German Research Center for Environmental Health, Molecular EXposomics, Ingolstadter Landstrasse 1, Neuherberg D-85764, Germany (Li) School of Life Sciences Weihenstephan (Nutrition), Technische Universitat Munchen, Freising, Germany

(Main, Skakkebaek, Toppari) Department of Growth and Reproduction, EDMaRC, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark

(Kiviranta) National Institute for Health and Welfare, Department of Health Security, Kuopio, Finland

(Toppari) Institute of Biomedicine, University of Turku, Turku, Finland

(Toppari) Department of Pediatrics, Turku University Hospital, Turku, Finland

(Feldt-Rasmussen) Department of Medical Endocrinology PE, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark

(Shen) Key Laboratory of Urban Environment and Health, Institute of Urban Environment, Chinese Academy of Sciences, Xiamen, China

(Schramm) Department für Biowissenschaftliche Grundlagen, Technische Universität München, Freising, Germany

Publisher

Oxford University Press

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2018

200.

Shortcomings in the management of undescended testis: guideline intention vs reality and the underlying causes - insights from the biggest German cohort.

Boehme P., Geis B., Doerner J., Wirth S., Hensel K.O.

Embase

BJU International. 122 (4) (pp 644-653), 2018. Date of Publication: October 2018.

[Article]

AN: 624064154

Objectives: To assess the implementation of the current guideline and identify potential underlying causes for late surgery in children with undescended testis (UDT) in Germany. UDT is the most common surgical issue in paediatric urology and to avoid malignant degeneration and subfertility current guidelines recommend orchidopexy during the first year of life; however, this seems not to be implemented in practice.

Patients and Methods: In all, 5 547 patients with cryptorchidism at 16 hospitals nationwide were studied regarding age at orchidopexy between 2003 and 2016. Multivariate analysis was performed to identify factors influencing timing of surgery. Additionally, a survey on knowledge of UDT management was conducted amongst physicians treating boys and final-year medical students.

Result(s): Between 2003 and 2008 only 4% of boys with UDT underwent surgery before the age of 1 year. After the guideline update from 2009, this figure was 5% from 2010 to 2012, and 8% from 2013 to 2016. The presence of a specialised department for paediatric surgery, as well as a

high UDT case-to-year ratio positively influenced the timing of orchidopexy. The survey revealed discipline-specific differences in the levels of knowledge about UDT management. One-third of respondents did not know the guideline recommendations and 61% felt insufficiently informed. International comparisons revealed significant differences in the age at surgery of boys with UDT, with Germany and Great Britain ranging in the middle of the field.

Conclusion(s): Currently, only a small proportion of boys with UDT are operated upon during their first year of life. The level of knowledge in attending physicians remains in need of improvement. This should be actively addressed, i.e. by campaigns and educational programmes. Further studies are needed to investigate the underlying causes of late orchidopexy in UDT.

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Status

Embase

Author NameID

Hensel, Kai O.; ORCID: <http://orcid.org/0000-0001-5826-6133>

Institution

(Boehme, Wirth, Hensel) Witten/Herdecke University, Department of Paediatrics, Centre for Clinical and Translational Research (CCTR), Wuppertal University Hospital, Wuppertal, Germany

(Boehme) Cardiovascular Research, Bayer Pharma AG, Wuppertal, Germany

(Geis) Institute of Medical Biometry and Epidemiology (IMBE), Faculty of Health, Witten/Herdecke University, Witten, Germany

(Doerner) Witten/Herdecke University, Department of Surgery, Centre for General and Visceral Surgery, HELIOS University Hospital Wuppertal, Wuppertal, Germany

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Year of Publication

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201.

The Role of the Appendix Testis in Normal Testicular Descent: Is There a Connection?.

Zvizdic Z., Zivkovic D., Sabanovic J., Milisic E.

Embase

BioMed Research International. 2018 (no pagination), 2018. Article Number: 3078031. Date of Publication: 23 Apr 2018.

[Article]

AN: 622060544

Objective. The presence of testicular appendices was prospectively evaluated in 89 boys with 96 undescended testes who underwent orchidopexy over the period of 4 years. Results. The patients were divided into two groups. Group A included 42 boys with 49 undescended testes positioned close to the internal inguinal ring, and Group B included 47 boys with 47 undescended testes close to the external inguinal ring. The incidence of appendix testis (AT) in Group A was 57.1% (28 in 49) and 78.7% (37 in 47) in Group B. The results of our study showed significantly decreased incidence of testicular appendices in undescended testes positioned close to the internal inguinal ring compared with undescended testes positioned close to the external inguinal ring ($p < 0.05$). Conclusion. AT may play a role in normal testicular descent and the undescended testis positioned close to the external inguinal ring can be considered as a separate entity of the true congenital undescended testis.

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Author NameID

Zvizdic, Zlatan; ORCID: <http://orcid.org/0000-0003-1238-880X> Zivkovic, Dragana; ORCID: <http://orcid.org/0000-0001-9391-2938>

Institution

(Zvizdic, Sabanovic, Milisic) Clinic of Pediatric Surgery, Clinical Center University of Sarajevo, Sarajevo, Bosnia and Herzegovina (Zvizdic) Medical Faculty, University of Sarajevo, Sarajevo, Bosnia and Herzegovina

(Zivkovic) Department of Urology, Institute for Children and Youth Health Care of Vojvodina, Novi Sad, Serbia

Publisher

Hindawi Limited (410 Park Avenue, 15th Floor, 287 pmb, New York NY 10022, United States)

Year of Publication

2018

202.

Congenital Abnormalities of the Male Reproductive System and Risk of Autism Spectrum Disorders.

Rotem R.S., Chodick G., Davidovitch M., Hauser R., Coull B.A., Weisskopf M.G.

Embase

American Journal of Epidemiology. 187 (4) (pp 656-663), 2018. Date of Publication: 01 Apr 2018.

[Article]

AN: 621707034

Androgens have an extensive influence on brain development in regions of the brain that are relevant for autism spectrum disorder (ASD), yet their etiological involvement remains unclear. Hypospadias (abnormal positioning of the urethral opening) and cryptorchidism (undescended testes) are 2 relatively common male birth defects that are strongly associated with prenatal androgen deficiencies. Having either disorder is a proxy indicator of atypical gestational androgen exposure, yet the association between these disorders and autism has not been extensively studied. We analyzed male singleton live births ($n = 224,598$) occurring from January 1, 1999, through December 31, 2013, in a large Israeli health-care organization. Boys with autism, cryptorchidism, and hypospadias were identified via International Classification of Diseases, Ninth Revision, codes, with further verification of autism case status by review of medical records. In multivariable-adjusted analyses, the odds ratio for ASD among boys with either condition was 1.62 (95% confidence interval (CI): 1.44, 1.82). The odds ratio for boys with cryptorchidism only was 1.55 (95% CI: 1.34, 1.78), and that for boys with hypospadias only was 1.65 (95% CI: 1.38, 1.98). ASD risk was not elevated among unaffected brothers of hypospadias or cryptorchidism cases, despite familial aggregation of all 3 conditions, providing some indication for the possibility of pregnancy-specific risk factors driving the observed associations. Results suggest that in-utero hypoandrogenicity could play a role in ASD etiology.

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Embase

Institution

(Rotem, Hauser, Coull, Weisskopf) Department of Environmental Health, Harvard T.H. Chan School of Public Health, 401 Park Drive, Landmark Center, P.O. Box 15697, Boston, MA 02215, United States (Hauser, Weisskopf) Department of Epidemiology, Harvard T.H. Chan School of Public Health, Boston, MA, United States

(Coull) Department of Biostatistics, Harvard T.H. Chan School of Public Health, Boston, MA, United States

(Chodick) Maccabi Institute for Research and Innovation, Maccabi Healthcare Services, Tel Aviv, Israel

(Davidovitch) Child Development Department, Maccabi Healthcare Services, Tel Aviv, Israel

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Oxford University Press

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2018

203.

Nonpalpable testes: Ultrasound and contralateral testicular hypertrophy predict the surgical access, avoiding unnecessary laparoscopy.

Berger C., Haid B., Becker T., Koen M., Roesch J., Oswald J.

Embase

Journal of Pediatric Urology. 14 (2) (pp 163.e1-163.e7), 2018. Date of Publication: April 2018.

[Article]

AN: 619494036

Introduction: In up to 20% of patients presenting with undescended testes, one or both are non-palpable. Whereas the most reliable means to exclude an abdominal testis is laparoscopy, there has been a lot of debate about the role of inguinal ultrasound (US) in detecting non-palpable inguinal testis. While we do not aim to add another paper claiming the benefits of US, we wanted to determine the excess capability of US to determine the correct surgical approach - inguinal or laparoscopy. In the light of avoiding unnecessary diagnostic laparoscopies, even the cost-effectiveness raised in many current papers might be called into question.

Patients and Methods: Of a total of 684 boys who underwent surgery for undescended testes at our department between 2011 and 2014, in 58 (8.5%), one or both testes were neither palpable preoperatively nor under general anesthesia. These boys were examined by two experienced

pediatric urologists clinically as well as by US. Besides the size of the contralateral testis, the presence of a testis in the inguinal channel was investigated. The additional impact of US over clinical exam and consideration of the size of the contralateral testis was assessed by means of intra-individual comparisons using Cochran-Q as well as McNemar tests.

Result(s): Clinical exam without considering the size of the contralateral testis had a sensitivity of 9% (95% CI 2-24%) and a specificity of 100% (95% CI 86-100%) to accurately predict the surgical approach deemed appropriate postoperatively. The consideration of the size of the contralateral testis - taken as an isolated factor - accurately predicted the surgical approach with a sensitivity of 21% (95% CI 9-38%) and a specificity of 88% (95% CI 68-97%). Ultrasound accounted for a sensitivity of 53% (95% CI 35-70%) and a specificity of 100% (95% CI 86-100%). The addition of US increased the sensitivity to correctly predict an inguinal incision from 29% to 71% and specificity slightly increased from 88% to 92%. This difference is significant ($p = 0.008$) in the bilateral McNemar test (Figure).

Conclusion(s): Inguinal US of non-palpable testes and measurement of the contralateral testis are synergistic in predicting the surgical approach. The addition of ultrasound to a clinical exam, performed also under general anesthesia and by an experienced pediatric urologist significantly increases the prediction of the correct surgical approach. Our results translate into five boys needing an US of the NPT to prevent one laparoscopy. Whereas cost-effectiveness of US might be debatable in regard to different healthcare systems, it is proven to be an effective, non-harmful tool to avoid unnecessary diagnostic laparoscopies.[Figure presented]

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Institution

(Berger, Haid, Becker, Koen, Roesch, Oswald) Department of Pediatric Urology, Hospital of the Sisters of Charity, Linz, Austria

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204.

Orchidopexy in children with Prader-Willi syndrome: Results of a long-term follow-up study.

Pacilli M., Heloury Y., O'Brien M., Lioni T., Rowell M., Hutson J.

Embase

Journal of Pediatric Urology. 14 (1) (pp 63.e1-63.e6), 2018. Date of Publication: February 2018.

[Article]

AN: 619197769

Introduction: Prader-Willi syndrome (PWS) is a rare (1:20.000) genetic condition affecting both males and females. Among other features, in boys, the syndrome is characterized by cryptorchidism in 86-100% of cases, hypogonadism, delayed puberty and infertility. The aim of the present study is to appraise the results of orchidopexy in this selected population of children. Study design: A follow-up study of children with PWS treated for undescended testes at a single institution over a 20-year period was performed. Patients were identified from a National PWS registry and reviewed at a special follow-up clinic. Data were collected from electronic and hard copies records and reported as median (range).

Result(s): Thirty-three children (1-17 years) were identified. Co-morbidities were present in 22 (66%) and 15 (45%) were on growth-hormone therapy. Six patients (19%) had normal testes palpable in the scrotum; twenty-seven (81%) had undescended testes and required orchidopexy. Thirteen (48%) underwent a bilateral procedure for a total of 40 procedures. A 2-stage Fowler-Stephens orchidopexy was required in 2 (7%) testes. At surgery hypotrophic testes were documented in 6 (22%) patients. Age at orchidopexy was 1.4 years (0.5-5.5). Age at F-U was 7.2 years (1.7-17). Length of follow-up is 3.5 years (0.4-14). At follow-up 16 (40%) testes were of normal size and palpable in the scrotum; 7 (17.5%) testes required redo-orchidopexy. All patients (6/33) over 16 years of age that had testosterone levels tested had values below normal limits after successful orchidopexy.

Conclusion(s): This study evaluates the results of orchidopexy in a large population of children with PWS. At follow-up, only 40% of testes were of normal size and in the scrotum. This information should be taken into consideration for patients' management and pre-operative parents' counseling. [Table presented]

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Status

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Author NameID

Pacilli, Maurizio; ORCID: <http://orcid.org/0000-0003-1259-4304>

Institution

(Pacilli) Department of Paediatric Surgery and Department of Paediatrics, Monash University, Monash Children's Hospital, Melbourne, Australia (Pacilli) Department of General Surgery, Monash Children's Hospital and Department of Paediatrics, Monash University, Melbourne, Australia

(Heloury, O'Brien, Hutson) Department of Paediatric Urology, Royal Children's Hospital, Melbourne, Australia

(Lionti) Murdoch Children's Research Institute, Royal Children's Hospital, Melbourne, Australia

(Rowell) Developmental Medicine, Royal Children's Hospital, Melbourne, Australia

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205.

Urinary carbohydrate antigen 19-9/creatinine ratio: A non-invasive marker for follow-up of unilateral ureteropelvic junction obstruction in children.

Alizadeh F., Taefnia A.M., Haghdani S.

Embase

Journal of Pediatric Urology. 14 (1) (pp 62.e1-62.e4), 2018. Date of Publication: February 2018.

[Article]

AN: 619197462

Introduction: Ureteropelvic junction obstruction (UPJO) is the most common cause of hydronephrosis in children. One major challenge in the management of UPJO is to select the patients that must be subjected to early obstruction relief. Currently, there is no gold standard for this assessment. Therefore, the aim of the present study was to evaluate the urinary levels of carbohydrate antigen (CA) 19-9 and normalized CA 19-9 (Ca 19-9/Cr ratio) in UPJO patients before and after surgery and compare them with a control group to assess their potential clinical application as an assisting tool in diagnosis of UPJO patients.

Material(s) and Method(s): From Jan 2013 to Jun 2016, 30 children with history of inguinal hernia, circumcision, hydrocele, and undescended testis as the control group (group 1) and 30 children with unilateral congenital UPJO (group 2) were enrolled in the study. Random CA 19-9 and

random creatinine were measured in the voided urine samples of control group and proven congenital UPJO group preoperatively (group 2A) and at 6 months after dismembered pyeloplasty (group 2B). In addition, the random urinary CA 19-9/CR ratio was evaluated as a marker to normalized urinary CA 19-9.

Discussion(s): The urinary CA 19-9/Cr ratio was significantly greater in the UPJO group than in the control group. The urinary CA 19-9 also was more in group 2A than in group 1; however, it was not statistically significant. The urinary CA 19-9/Cr ratio and renal pelvis anteroposterior diameter decreased significantly in the group 2B 6 month after complete relief of obstruction in comparison with group 2A preoperatively. Urinary CA 19-9 also decreased in 2B group though it was not significant.

Conclusion(s): Urinary CA 19-9/Cr ratio is suggested as a non-invasive marker that can assist in diagnosis and long-term follow-up of UPJO patients. This ratio is superior to urinary CA 19-9 as it is more strongly correlated with UPJ obstruction.[Figure presented]

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Status

Embase

Institution

(Alizadeh, Taefnia) Department of Urology, Isfahan University of Medical Sciences (IUMS), Isfahan, Iran, Islamic Republic of (Haghdani) Department of Urology, Hasheminejad Kidney Research Center (HKRC), Iran University of Medical Science (IUMS), Tehran, Iran, Islamic Republic of

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206.

Incidence and diagnoses of disorders of sex development in proximal hypospadias.

Wong Y.S., Tam Y.H., Pang K.K.Y., Yau H.C.

Embase

Journal of Pediatric Surgery. 53 (12) (pp 2498-2501), 2018. Date of Publication: December 2018.
[Article]

AN: 2001103715

Background: Evidence-based guidelines on evaluation of boys with proximal hypospadias for the possibility of a disorder of sex development (DSD) have yet to be developed. We aimed to investigate the incidence and diagnoses of DSD in patients with proximal hypospadias.

Method(s): We retrospectively reviewed the records of consecutive boys who underwent proximal hypospadias repairs from 2006 to Sept 2017. Data collected included scrotal anomaly, testes position/palpability, micropenis, DSD investigations, and surgical techniques.

Result(s): 165 patients were eligible for the study. 14 (8.5%) were diagnosed to have DSD. The diagnoses were 46,XX testicular DSD [n = 1], 46,XY DSD [n = 7; partial gonadal dysgenesis (PGD) = 3; 5alpha-reductase type 2 deficiency = 3; 17alpha-hydroxylase deficiency = 1], Sex Chromosome DSD [n = 6; 45,X/46,XY PGD = 4; Klinefelter = 2]. 3/7 (43%) patients with PGD had gonadal germ cell neoplasms. Of the DSD patients, 6/14 (43%), 11/14 (79%) and 11/14 (79%) had undescended/impalpable testes, micropenis and penoscrotal transposition/bifid scrotum, respectively, significantly higher prevalence rates than those without DSD diagnosis (p-values < 0.05). 10/14 (71.4%) DSD patients underwent 2-stage repair compared with 57/151 (37.7%) of others without DSD diagnosis (p = 0.01).

Conclusion(s): Patients presenting with proximal hypospadias and one or more of the coexisting anomalies of micropenis, undescended/impalpable testes, and penoscrotal transposition/bifid scrotum should warrant DSD evaluation. Presence of bilaterally descended testes in scrotum does not preclude the possibility of DSD.

Level of Evidence: IV.

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Status

Embase

Institution

(Wong, Tam, Pang) Division of Paediatric Surgery and Paediatric Urology, Department of Surgery, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong (Yau) Department of Paediatrics, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong

Publisher

W.B. Saunders

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207.

Endocrine disruptors and testicular function.

Lymperi S., Giwercman A.

Embase

Metabolism: Clinical and Experimental. 86 (pp 79-90), 2018. Date of Publication: September 2018.

[Article]

AN: 2000719871

Despite concerns of the scientific community regarding the adverse effects of human exposure to exogenous man-made chemical substances or mixtures that interfere with normal hormonal balance, the so called "endocrine disruptors (EDs)", their production has been increased during the last few decades. EDs' extensive use has been implicated in the increasing incidence of male reproductive disorders including poor semen quality, testicular malignancies and congenital developmental defects such as hypospadias and cryptorchidism. Several animal studies have demonstrated that exposure to EDs during fetal, neonatal and adult life has deleterious consequences on male reproductive system; however, the evidence on humans remains ambiguous. The complexity of their mode of action, the differential effect according to the developmental stage that exposure occurs, the latency from exposure and the influence of the genetic background in the manifestation of their toxic effects are all responsible factors for the contradictory outcomes. Furthermore, the heterogeneity in the published human studies has hampered agreement in the field. Interventional studies to establish causality would be desirable, but unfortunately the nature of the field excludes this possibility. Therefore, future studies based on standardized guidelines are necessary, in order to estimate human health risks and implement policies to limit public exposure.

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Institution

(Lymeri) Unit of Reproductive Endocrinology, First Department of Obstetrics and Gynecology, Aristotle University of Thessaloniki, Thessaloniki, Greece (Giwerzman) Department of Translational Medicine, Lund University, Malmo, Sweden

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208.

Adolescent testicular microlithiasis: A case-based, multinational survey of clinical management practices.

Brodie K.E., Saltzman A.F., Cost N.G.

Embase

Journal of Pediatric Urology. 14 (2) (pp 151.e1-151.e8), 2018. Date of Publication: April 2018.

[Article]

AN: 2000599740

Introduction: Testicular microlithiasis (TM) is a condition characterized by calcium deposits within the testis, usually detected incidentally during ultrasonography of the scrotum. TM has been associated with the presence of, and possibly the development of, testicular malignancy. Our aim was to document international clinical management practices for TM and to analyze what factors and perception of risk influence conservative versus active management and follow-up.

Method(s): European Society for Paediatric Urology (ESPU) and Society for Pediatric Urology (SPU) members were invited to complete an online case-based survey of clinical management practices of TM. Eight cases had a single variable changed each time (classic versus limited TM, unilateral versus bilateral, prior cryptorchidism versus no cryptorchidism) to ascertain the provider's perception of risk. The respondents completed multiple choice questions on initial management, follow-up plan, length and interval of follow-up. Multivariate logistic regression was performed to determine factors associated with decisions on management and follow-up.

Result(s): There were 265 respondents to the survey from 35 countries (Table). Median time in practice was 13 years. Factors that were significantly associated with more aggressive initial management (more than counseling on self-examination) included: not yet in independent practice, low volume TM cases per year, those practicing pediatric and adult urology, classic

appearance of TM and cryptorchidism. Factors that were significantly associated with urologist follow-up and active investigation included: European practitioners, low TM case volume per year, those practicing both pediatric urology and pediatric surgery, classic TM appearance and a case history of cryptorchidism. Interval and length of follow-up was wide-ranging, with most respondents favoring annual follow-up.

Conclusion(s): Management of TM varies and a mix of surgeon and case factors significantly influences management strategies. This baseline understanding of the lack of systematic management suggests the need for the development of consensus guidelines and prospective study. [Table presented]

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PMC Identifier

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Status

Embase

Institution

(Brodie, Saltzman, Cost) Department of Surgery, Division of Urology, University of Colorado School of Medicine, Aurora, CO, United States

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Year of Publication

2018

209.

Birth outcomes after inadvertent use of category x drugs contraindicated in pregnancy: Where is the real risk?.

Ozturk Z., Olmez E., Gurpinar T., Vural K.

Embase

Turkish Journal of Pediatrics. 60 (3) (pp 298-305), 2018. Date of Publication: 2018.

[Article]

AN: 625238066

Drugs contraindicated in pregnancy are medicines that should be avoided by pregnant women, since they carry a concern for teratogenicity or there is no indication for their use during

pregnancy. It does not mean that exposures to these drugs always cause harm. The aim of the present study was to investigate the risk of adverse outcomes following maternal exposure to the drugs contraindicated in pregnancy. We retrospectively analyzed prenatal drug exposure records of the pregnant patients referred to the clinical pharmacology consultation service in a tertiary-level university hospital from January 2007 until December 2012. Exposures to category X drugs (CXD) contraindicated in pregnancy were evaluated. After the expected date of delivery, we collected data about pregnancy complications and the outcomes. For comparison the women in the exposed group (N=52) were matched with a control group (N=162) of pregnant women without teratogenic exposure. We observed only one baby born with a birth defect (congenital cryptorchidism) in CXD group (2.6%) and four in control group (RR 0.91; 95% CI 0.10-7.94). The rates of adverse pregnancy outcomes including miscarriage, preterm birth and congenital abnormality were not significantly different from controls. However, the rate of elective termination of pregnancy was higher in women exposed to CXD while pregnant (RR 2.54; 95% CI 1.11-5.80, $p = 0.027$). Contraceptive failure and unintended pregnancy are the reasons for inadvertent drug exposure and choosing abortion. The high perception of teratogenic risk among pregnant women may cause terminations of pregnancies. Individual risk assessment and avoiding the phrase 'CXD' or 'contraindicated in pregnancy' in counseling may help to reduce maternal concerns about medication use in pregnancy.

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Embase

Institution

(Ozturk) Department of Clinical Pharmacology and Toxicology, Izmir Ataturk Research Hospital, Izmir, Turkey (Ozturk) Department of Healthcare Management, Izmir University of Economics, Izmir, Turkey

(Olmez, Gurpinar, Vural) Department of Pharmacology, Celal Bayar University, Manisa, Turkey

Publisher

Turkish Journal of Pediatrics (E-mail: deryakaraduman@gmail.com)

Year of Publication

2018

210.

Anogenital distance is associated with genital measures and seminal parameters but not anthropometrics in a large cohort of young adult men.

Foresta C., Valente U., Di Nisio A., Cacco N., Magagna S., Cosci I., Presciutti A., Garolla A.
Embase

Human Reproduction. 33 (9) (pp 1628-1635), 2018. Date of Publication: 2018.

[Article]

AN: 624416397

STUDY QUESTION: Is the anogenital distance (AGD) correlated to anthropometric, genital and sperm parameters in young adult men? SUMMARY ANSWER: We observed that reduced AGD is strongly associated with altered semen parameters and reduced testicular volume. WHAT IS KNOWN ALREADY: Abnormalities in the foetal development of the testis have been suggested as causative of common male reproductive disorders, such as cryptorchidism, hypospadias, reduced semen quality and testicular germ cell tumour, collectively defined as 'testicular dysgenesis syndrome'. In human epidemiological studies, alterations in AGD have been frequently associated with clinically relevant outcomes of reproductive health, suggesting AGD as a marker of foetal testicular development. STUDY DESIGN, SIZE, DURATION: This study was performed within the annual screening protocol to evaluate male reproductive health in the high schools of Padua and surroundings (Veneto Region, the North-East of Italy). Here we report the findings of 794 subjects who completed the study protocol between October 2016 and May 2017. PARTICIPANTS/MATERIALS, SETTING, METHODS: We evaluated 794 students aged 18-19 years recording the following parameters: height, weight, BMI, waist circumference, arm span, pubis-to-floor and crown-to-pubis length, penile length and circumference, testicular volumes, semen parameters and AGD (measured from the posterior base of the scrotum to the centre of the anus). MAIN RESULTS AND THE ROLE OF CHANCE: Of the subjects, 49% had an abnormal arm span-height difference (>3 cm) and 63.4% had an altered ratio of crown-to-pubis/pubis-to-floor length (≤ 0.92). The rate of subjects with reduced testicular volume was 23%. Median sperm concentration was $51.0 \times 10^6/\text{ml}$ and total sperm count was 122.5×10^6 . AGD showed a direct positive relation with testicular volume and penile length and circumference ($R = 0.265, 0.176$ and 0.095 , respectively, all $P < 0.05$). No significant relation was observed between AGD and anthropometric parameters. Sperm concentration, total sperm count, progressive motility and normal morphology showed a significant and positive correlation with AGD ($R = 0.205, 0.210, 0.216$ and 0.117 , respectively, all $P < 0.05$). LIMITATIONS, REASONS FOR CAUTION: Our cohort of young adults is not representative of the general population. Hormonal evaluation was missing. WIDER IMPLICATIONS OF THE FINDINGS: Our findings show that AGD is associated with testicular volumes, penile measures and seminal parameters in

young adult men. Because AGD is hormonally determined during foetal life, the reported high incidence of reduced semen quality and reduced testicular volume could be related to a reduced androgenic exposure in utero. AGD could represent a simple and useful method to evaluate testicular and penile development in adult men. STUDY FUNDING/COMPETING INTEREST(S): The authors have no potential conflict of interest to declare. No external funding was obtained for this study. TRIAL REGISTRATION NUMBER: N/A.

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PMC Identifier

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Embase

Institution

(Foresta, Valente, Di Nisio, Cacco, Magagna, Cosci, Presciutti, Garolla) Department of Medicine, Operative Unit of Andrology and Medicine of Human Reproduction, University of Padova, Via Giustiniani, 2, Padova 35128, Italy

Publisher

Oxford University Press

Year of Publication

2018

211.

Gonadotropin-Releasing Hormone Agonist Corrects Defective Mini-Puberty in Boys with Cryptorchidism: A Prospective Randomized Study.

Vincel B., Verkauskas G., Bilius V., Dasevicius D., Malcius D., Jones B., Hadziselimovic F.

Embase

BioMed Research International. 2018 (no pagination), 2018. Article Number: 4651218. Date of Publication: 2018.

[Article]

AN: 623081805

Introduction. This prospective study investigated the efficacy of a gonadotropin-releasing hormone agonist (LH-RHa) in restoring defective mini-puberty. Materials and Methods. Boys with isolated bilateral cryptorchidism and defective mini-puberty were randomly divided into two

groups. The "surgery only" group underwent a second orchidopexy without hormonal treatment (control). The "LH-RHa" group received LH-RHa therapy followed by a second orchidopexy. The number of Ad spermatogonia and the total germ cell count per tubule (S/T) were analyzed. Results. Five boys were included in each arm. In the LH-RHa group, the median S/T increased from 0.11 to 0.42, $p=0.04$. In the surgery only group, the median S/T did not change. In the surgery only group, none of the testes had Ad spermatogonia. In contrast, in the LH-RHa group, all testes completed the transition from gonocytes to Ad spermatogonia ($p=0.008$). Conclusions. Treatment with LH-RHa was effective in rescuing defective mini-puberty in boys with bilateral cryptorchidism.

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Author NameID

Vincel, Beata; ORCID: <http://orcid.org/0000-0002-3751-1352> Verkauskas, Gilvydas; ORCID: <http://orcid.org/0000-0001-7152-0802>

Bilius, Vytautas; ORCID: <http://orcid.org/0000-0003-4309-7343>

Institution

(Vincel) Children's Surgery Centre, Clinic of Gastroenterology, Nephrourology and Surgery, Institute of Clinical Medicine, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

(Verkauskas, Bilius) Children's Surgery Centre, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

(Dasevicius) National Centre of Pathology, Affiliate of Vilnius University Hospital Santaros Klinikos, Vilnius, Lithuania

(Malcius) Department of Pediatric Surgery, Lithuanian University of Health Sciences, Kaunas, Lithuania

(Jones) Children's Hospital, Affiliate of Vilnius University Hospital Santaros Klinikos, Vilnius, Lithuania

(Hadziselimovic) Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, Liestal, Switzerland

Publisher

Hindawi Limited (410 Park Avenue, 15th Floor, 287 pmb, New York NY 10022, United States)

Year of Publication

2018

212.

Inhibin B in healthy and cryptorchid boys.

Esposito S., Cofini M., Rigante D., Leonardi A., Lucchetti L., Cipolla C., Lanciotti L., Penta L.

Embase

Italian Journal of Pediatrics. 44 (1) (no pagination), 2018. Article Number: 81. Date of Publication: 16 Jul 2018.

[Review]

AN: 623030646

Background: Cryptorchidism, the most common male genital abnormality observed in paediatrics, might often be associated with long-term functional consequences and can even reoccur after a successful orchidopexy. Serum markers that identify cryptorchid boys with gonadal dysfunction early should be useful in a decision-making process. Inhibin B, produced during all of childhood but altered in cryptorchid subjects, appears strictly related to Sertoli cells, and its levels directly reflect the status of the testis germinative epithelium. Unfortunately, its precise roles in bilateral and unilateral cryptorchidism are still debated and being unravelled. Herein, we report the most current knowledge about inhibin B in both healthy boys and those with cryptorchidism to discuss and clarify its potential clinical applications.

Discussion(s): Inhibin B represents a simple and repeatable serum marker and it seems to well asses the presence and function of the testicular tissue. Testicular tissue in prepubertal age is largely made up of Sertoli cells; inhibin B, coming from working Sertoli cells, allows to indirectly evaluate their function. Besides, inhibin B is produced throughout childhood, even before puberty, in contrast with central hormones, and it is not influenced by androgens during puberty, in contrast with other testicular hormones. Although further studies are needed, low levels of inhibin B have been related with low testicular score and/or with consistent alterations of testicular parameters at histological examination. This means that inhibin B could be an indirect marker of testicular functions that could even replace testicular biopsies, but current data are inconsistent to confirm this potential role of inhibin B in cryptorchidism.

Conclusion(s): Inhibin B represents an effective candidate for early identification of testicular dysfunction after orchidopexy for cryptorchidism. Unfortunately, current data cannot exactly clarify the real role of inhibin B as a predictor of future testicular function in cryptorchidism and future long-term follow-up studies, with repeated inhibin B checks both in cryptorchid and in formerly cryptorchid children and adolescents, will permit to assess if previous normal levels of inhibin B would match with future normal pubertal development and fertility potential.

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Author NameID

Esposito, Susanna; ORCID: <http://orcid.org/0000-0003-4103-2837>

Institution

(Esposito, Cofini, Leonardi, Lucchetti, Lanciotti, Penta) Pediatric Clinic, Department of Surgical and Biomedical Sciences, Universita Degli Studi di Perugia, Piazza Menghini 1, Perugia 06129, Italy (Rigante, Cipolla) Institute of Pediatrics, Universita Cattolica Sacro Cuore, Fondazione Policlinico, Universitario A. Gemelli, Rome, Italy

Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2018

213.

Maternal Diabetes Mellitus and Genital Anomalies in Male Offspring: A Nationwide Cohort Study in 2 Nordic Countries.

Arendt L.H., Lindhard M.S., Henriksen T.B., Olsen J., Cnattingius S., Petersson G., Parner E.T., Ramlau-Hansen C.H.

Embase

Epidemiology. 29 (2) (pp 280-289), 2018. Date of Publication: 01 Mar 2018.

[Article]

AN: 622578379

Background: Pre-existing diabetes has been associated with an increased risk of congenital malformations overall, but studies on genital anomalies in boys are conflicting and possible causal mechanisms are not well understood. Previous studies have mainly assessed pregestational and gestational diabetes in combination. Yet considering the vulnerable time windows for the genital anomalies, associations could well differ between types of diabetes and between the 2 genital anomalies and we therefore aimed to study this further.

Method(s): A population-based cohort study of 2,416,246 singleton live-born boys from Denmark (1978-2012) and Sweden (1987-2012) was carried out using Danish and Swedish register-based data. Using Cox regression models, we estimated hazard ratios for hypospadias and cryptorchidism according to maternal diabetes. We considered type and severity of diabetes, as well as timing of diagnosis in relation to birth.

Result(s): Pregestational type 1 diabetes was associated with a higher risk of both genital anomalies. The highest risks were seen for boys of mothers with diabetic complications (hazard ratio for hypospadias = 2.33 [95% confidence interval, 1.48, 3.66] and hazard ratio for cryptorchidism = 1.92 [95% confidence interval, 1.39, 2.65]). Gestational diabetes was associated with slightly increased risks of both genital anomalies.

Conclusion(s): These results are consistent with the hypothesis that poor glycemic control may interfere with fetal genital development in the critical early period of organogenesis. Given the widespread and increasing occurrence of diabetes, these results are of public health importance.

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Institution

(Arendt, Ramlau-Hansen) Department of Public Health, Section for Epidemiology, Aarhus University, Bartholins Alle 2, Aarhus C 8000, Denmark (Arendt, Lindhard, Henriksen) Perinatal Epidemiology Research Unit, Department of Paediatric and Adolescent Medicine, Aarhus University Hospital, Aarhus, Denmark

(Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

(Cnattingius, Petersson) Clinical Epidemiology Unit, Department of Medicine, Karolinska University Hospital, Karolinska Institute, Stockholm, Sweden

(Parner) Department of Public Health, Section for Biostatistics, Aarhus University, Aarhus, Denmark

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

Year of Publication

2018

214.

Prevalence and risk factors of testicular microlithiasis in patients with hypospadias: A retrospective study.

Nakamura M., Moriya K., Nishimura Y., Nishida M., Kudo Y., Kanno Y., Kitta T., Kon M., Shinohara N.

Embase

BMC Pediatrics. 18 (1) (no pagination), 2018. Article Number: 179. Date of Publication: 29 May 2018.

[Article]

AN: 622334304

Background: It has been described that the incidence of testicular microlithiasis is high in several congenital disorders which may be associated with testicular impairment and infertility. Several reports have shown that a prepubertal or pubertal hormonal abnormality in the pituitary-gonadal axis was identified in some patients with hypospadias that is one of the most common disorders of sex development. However, exact prevalence or risk factors of testicular microlithiasis in patients with hypospadias have not reported so far. In the present study, to clarify the prevalence and risk factors of testicular microlithiasis in patients with hypospadias, a retrospective chart review was performed.

Method(s): Children with hypospadias who underwent testicular ultrasonography between January 2010 and April 2016 were enrolled in the present study. Severity of hypospadias was divided into mild and severe. The prevalence and risk factors of testicular microlithiasis or classic testicular microlithiasis were examined.

Result(s): Of 121 children, mild and severe hypospadias were identified in 66 and 55, respectively. Sixteen children had undescended testis. Median age at ultrasonography evaluation was 1.7 years old. Testicular microlithiasis and classic testicular microlithiasis were documented in 17 children (14.0%) and 8 (6.6%), respectively. Logistic regression analysis revealed that presence of undescended testis was only a significant factor for testicular microlithiasis and classic testicular microlithiasis. The prevalence of testicular microlithiasis or classic testicular microlithiasis was significantly higher in children with undescended testis compared to those without undescended testis (testicular microlithiasis; 43.8% versus 9.5% ($p=0.002$), classic testicular microlithiasis; 37.5% versus 1.9% ($p<0.001$)).

Conclusion(s): The current study demonstrated that the presence of undescended testis was only a significant risk factor for testicular microlithiasis or classic testicular microlithiasis in patients with hypospadias. As co-existing undescended testis has been reported as a risk factor for testicular dysfunction among patients with hypospadias, the current findings suggest that testicular microlithiasis in children with hypospadias may be associated with impaired testicular

function. Conversely, patients with isolated HS seem to have lower risks for testicular impairment. Further investigation with longer follow-up will be needed to clarify these findings.

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Embase

Institution

(Nakamura, Moriya, Nishimura, Kanno, Kitta, Kon, Shinohara) Hokkaido University Graduate School of Medicine, Department of Renal and Genitourinary Surgery, North-15, West-7, Kita-Ku, Sapporo 060-8638, Japan (Nishida, Kudo) Hokkaido University Hospital, Diagnostic Center for Sonography, Sapporo, Japan

(Nishida, Kudo) Hokkaido University Hospital, Division of Laboratory and Transfusion Medicine, Sapporo, Japan

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Year of Publication

2018

215.

Subphenotype meta-analysis of testicular cancer genome-wide association study data suggests a role for RBFOX family genes in cryptorchidism susceptibility.

Wang Y., Gray D.R., Robbins A.K., Crowgey E.L., Chanock S.J., Greene M.H., McGlynn K.A., Nathanson K., Turnbull C., Wang Z., Devoto M., Barthold J.S.

Embase

Human Reproduction. 33 (5) (pp 967-977), 2018. Date of Publication: 01 May 2018.

[Article]

AN: 622230155

Study Question: Can subphenotype analysis of genome-wide association study (GWAS) data from subjects with testicular germ cell tumor (TGCT) provide insight into cryptorchidism (undescended testis, UDT) susceptibility? Summary Answer: Suggestive intragenic GWAS signals common to UDT, TGCT case-case and TGCT case-control analyses occur in genes

encoding RBFOX RNA-binding proteins (RBPs) and their neurodevelopmental targets. What is Known Already: UDT is a strong risk factor for TGCT, but while genetic risk factors for TGCT are well-known, genetic susceptibility to UDT is poorly understood and appears to be more complex.

STUDY DESIGN, SIZE, DURATION We performed a secondary subphenotype analysis of existing GWAS data from the Testicular Cancer Consortium (TECAC) and compared these results with our previously published UDT GWAS data, and with data previously acquired from studies of the fetal rat gubernaculum. **Participants/Materials, Setting, Methods:** Studies from the National Cancer Institute (NCI), United Kingdom (UK) and University of Pennsylvania (Penn) that enrolled white subjects were the source of the TGCT GWAS data. We completed UDT subphenotype case-case (TGCT/UDT vs TGCT/non-UDT) and case-control (TGCT/UDT vs control), collectively referred to as 'TECAC' analyses, followed by a meta-analysis comprising 129 TGCT/UDT cases, 1771 TGCT/non-UDT cases, and 3967 unaffected controls. We reanalyzed our UDT GWAS results comprising 844 cases and 2718 controls by mapping suggestive UDT and TECAC signals (defined as $P < 0.001$) to genes using Ingenuity Pathway Analysis (IPA). We compared associated pathways and enriched gene categories common to all analyses after Benjamini-Hochberg multiple testing correction, and analyzed transcript levels and protein expression using qRT-PCR and rat fetal gubernaculum confocal imaging, respectively.

Main Results and the Role of Chance: We found suggestive signals within 19 genes common to all three analyses, including RBFOX1 and RBFOX3, neurodevelopmental paralogs that encode RBPs targeting (U)GCATG-containing transcripts. Ten of the 19 genes participate in neurodevelopment and/or contribute to risk of neurodevelopmental disorders. Experimentally predicted RBFOX gene targets were strongly overrepresented among suggestive intragenic signals for the UDT (117 of 628 (19%), $P = 3.5 \times 10^{-24}$), TECAC case-case (129 of 711 (18%), $P = 2.5 \times 10^{-27}$) and TECAC case-control (117 of 679 (17%), $P = 2 \times 10^{-21}$) analyses, and a majority of the genes common to all three analyses (12 of 19 (63%), $P = 3 \times 10^{-9}$) are predicted RBFOX targets. Rbfox1, Rbfox2 and their encoded proteins are expressed in the rat fetal gubernaculum. Predicted RBFOX targets are also enriched among transcripts differentially regulated in the fetal gubernaculum during normal development ($P = 3 \times 10^{-31}$), in response to in vitro hormonal stimulation ($P = 5 \times 10^{-45}$) and in the cryptorchid LE/orl rat ($P = 2 \times 10^{-42}$).

Large Scale Data: GWAS data included in this study are available in the database of Genotypes and Phenotypes (dbGaP accession numbers phs000986.v1.p1 and phs001349.v1p1). **Limitations, Reasons for Caution:** These GWAS data did not reach genome-wide significance for any individual analysis. UDT appears to have a complex etiology that also includes environmental factors, and such complexity may require much larger sample sizes than are currently available. The current methodology may also introduce bias that favors false discovery of larger genes.

Wider Implications of the Findings: Common suggestive intragenic GWAS signals suggest that RBFOX paralogs and other neurodevelopmental genes are potential UDT risk candidates, and

potential TGCT susceptibility modifiers. Enrichment of predicted RBFOX targets among differentially expressed transcripts in the fetal gubernaculum additionally suggests a role for this RBP family in regulation of testicular descent. As RBFOX proteins regulate alternative splicing of Calca to generate calcitonin gene-related peptide, a protein linked to development and function of the gubernaculum, additional studies that address the role of these proteins in UDT are warranted. Study Funding/Competing Interest(S): The Eunice Kennedy Shriver National Institute for Child Health and Human Development (R01HD060769); National Center for Research Resources (P20RR20173), National Institute of General Medical Sciences (P20GM103464), Nemours Biomedical Research, the Testicular Cancer Consortium (U01CA164947), the Intramural Research Program of the NCI, a support services contract HHSN26120130003C with IMS, Inc., the Abramson Cancer Center at Penn, National Cancer Institute (CA114478), the Institute of Cancer Research, UK and the Wellcome Trust Case-Control Consortium (WTCCC) 2. None of the authors reports a conflict of interest.

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Status

Embase

Institution

(Wang, Gray, Robbins, Crowgey, Barthold) Nemours Biomedical Research, Alfred I. DuPont Hospital for Children, Wilmington, DE, United States (Chanock, Greene, McGlynn) Division of Cancer Epidemiology and Genetics, Department of Health and Human Services, National Cancer Institute, National Institutes of Health, Rockville, MD, United States

(Nathanson) Department of Medicine, Division of Translational Medicine and Human Genetics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, United States

(Turnbull) Division of Genetics and Epidemiology, Institute of Cancer Research, London, United Kingdom

(Wang) St. Jude Children's Research Hospital, Department of Computational Biology, Memphis, TN, United States

(Devoto) Division of Genetics, Children's Hospital of Philadelphia, Departments of Biostatistics and Epidemiology, Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, United States

(Devoto) Department of Molecular Medicine, Sapienza University, Rome, Italy

Publisher

Oxford University Press

Year of Publication

2018

216.

A diagnostic germ cell score for immature testicular tissue at risk of germ cell loss.

Heckmann L., Langenstroth-Rower D., Pock T., Wistuba J., Stukenborg J.-B., Zitzmann M., Kliesch S., Schlatt S., Neuhaus N.

Embase

Human Reproduction. 33 (4) (pp 636-645), 2018. Date of Publication: 01 Apr 2018.

[Article]

AN: 621532149

STUDY QUESTION: Can a systematic scoring procedure provide crucial information on the status of highly heterogeneous immature human testicular tissues in the context of cryopreservation for fertility preservation? SUMMARY ANSWER: We developed a systematic histological score as a novel diagnostic tool which differentiates the patient cohort according to the status of germ cell differentiation and number of spermatogonia (normal, diminished and absent), and which could be relevant in the fertility clinic. WHAT IS KNOWN ALREADY: Cryopreservation of testicular tissue of immature boys is currently considered the option for future fertility restoration. However, experimental techniques for the derivation of sperm as well as valid diagnostic scoring of these immature testis tissues are not yet reported. STUDY DESIGN, SIZE, DURATION: Testicular tissues of 39 patients (aged 220 years) who attended our clinic for cryopreservation between 2010 and 2015 were analyzed to determine the variability of testicular tissue composition, germ cell numbers and differentiation status. PARTICIPANTS/MATERIALS, SETTING, METHODS: Human testicular tissue samples were divided into three groups. Group NT included patients suffering from diseases which do not directly affect the testes (n = 6; aged 614 years), group AT included patients suffering from diseases that directly affect the testes (n = 14; 217 years), and group KS (Klinefelter patients, n = 19; 1220 years). Based on immunohistochemical stainings for MAGEA4, the differentiation status as well as the numbers of gonocytes, spermatogonia and spermatocytes were determined. MAIN RESULTS AND THE ROLE OF CHANCE: Testicular tissue samples from the NT group contained a mean of 100.3 spermatogonia/ mm³ (103). Highly heterogeneous and significantly lower mean numbers of spermatogonia were scored in testes from boys after cytotoxic exposures or with pre-existing disease (AT group: 35.7 spermatogonia/mm³ (103); KS group: 1.8 spermatogonia/mm³ (103)). In

addition, the germ cell differentiation status was determined and revealed tissues with either spermatogonia and gonocytes, only spermatogonia, spermatogonia and spermatocytes, or all three germ cell types were present. Based on spermatogonial numbers and differentiation status, we developed a germ cell score which we applied to each individual patient sample.

LIMITATIONS REASONS FOR CAUTION: Normal human testicular tissue samples are difficult to obtain for ethical reasons and the sample numbers were small. However, six such samples provide a valid baseline for the normal situation. WIDER IMPLICATIONS OF THE FINDINGS: Fertility preservation of immature male tissues is an emerging field and is currently offered in many specialized centers worldwide. Our diagnostic germ cell score delivers an easily applicable tool, facilitating patient counseling and thus ensuring comparability between the centers with regard to future studies.

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Embase

Author NameID

Stukenborg J.-B.; ORCID: <http://orcid.org/0000-0002-2839-1870>

Institution

(Heckmann, Langenstroth-Rower, Pock, Wistuba, Schlatt, Neuhaus) Centre of Reproductive Medicine and Andrology, Institute of Reproductive and Regenerative Biology, Albert-Schweitzer-Campus 1, Munster 48149, Germany (Stukenborg) Department of Women's and Children's Health, NORDFERTIL Research Lab Stockholm, Paediatric Endocrinology Unit, Karolinska Institutet and Karolinska University Hospital, Stockholm SE-17176, Sweden

(Zitzmann, Kliesch) Centre of Reproductive Medicine and Andrology, Department of Clinical and Surgical Andrology, Albert-Schweitzer-Campus 1, Munster 48149, Germany

Publisher

Oxford University Press

Year of Publication

2018

20S proteasome in the blood plasma of boys with cryptorchidism.

Toliczenko-Bernatowicz D., Matuszczak E., Tylicka M., Sankiewicz A., Komarowska M., Gorodkiewicz E., Debek W., Hermanowicz A.

Embase

Journal of Endocrinological Investigation. 41 (9) (pp 1103-1106), 2018. Date of Publication: 01 Sep 2018.

[Article]

AN: 620739081

Purpose: To evaluate the concentration of 20S proteasome in the blood plasma of boys with cryptorchidism.

Method(s): Patients-50 boys aged 1-4 years (median = 2.4 years) with unilateral cryptorchidism. The control group-50 healthy, age-matched boys (aged 1-4 years, median = 2.1 years), admitted for planned herniotomy. In our study, we used a novel technique Surface PLASMON RESONANCE Imaging.

Result(s): The median concentration of 20S proteasome in the blood plasma of boys with cryptorchidism was 2.5-fold higher than in boys with inguinal hernia. We noticed statistically significant difference between 20S proteasome levels in boys with cryptorchidism up to 2 years old and above 2 years old.

Conclusion(s): We believe that the 20S proteasome concentrations in the blood plasma of boys with cryptorchidism reflect the heat-induced apoptosis of germ cells.

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PMC Identifier

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Status

Embase

Institution

(Toliczenko-Bernatowicz, Matuszczak, Komarowska, Debek, Hermanowicz) Paediatric Surgery Department, Medical University of Bialystok, Waszyngtona 17, Bialystok 15-274, Poland

(Tylicka) Biophysics Department, Medical University of Bialystok, Bialystok, Poland

(Sankiewicz, Gorodkiewicz) Electrochemistry Department, University of Bialystok, Bialystok, Poland

Publisher

Springer International Publishing

Year of Publication

2018

218.

Testicular tumours in children: Indications for testis-sparing surgery. Tumores testiculares en la edad pediátrica: indicaciones de la cirugía conservadora <Tumores testiculares en la edad pediátrica: indicaciones de la cirugía conservadora.>

Romo Muñoz M.I., Nunez Cerezo V., Dore Reyes M., Vilanova Sanchez A., Gonzalez-Peramato P., Lopez Pereira P., Martinez Urrutia M.J.

Embase

Anales de Pediatría. 88 (5) (pp 253-258), 2018. Date of Publication: May 2018.

[Article]

AN: 617375804

Introduction and objectives: Although standard surgical treatment of a testicular tumour is orchiectomy, use can be made of testis-sparing surgery in selected cases, based on tumour markers, tumour size, and histopathological findings. Our objective is to become acquainted with the indications of testis-sparing surgery as a treatment for the incidental finding of a palpable and non-palpable testicular mass.

Material(s) and Method(s): A retrospective study was conducted on 22 patients younger than 18 years diagnosed with a testicular tumour between 2000 and 2014. An assessment was made of the condition, the history, ultrasound, histopathology, tumour markers (BHCG, AFP), therapeutic approach, and outcome.

Result(s): Of the 22 patients (10 prepubertal age) studied, 82% had palpable mass, and 18% were incidental findings. Two had cryptorchidism. The BHCG was increased in 27% and AFP in 45% of cases. There were 18 tumorectomies and 4 orchiectomies performed. The histopathology found 72% germ cell, 14 orchiectomy, and 2 tumorectomies (2 teratomas), with 27% non-germ cell tumours in 4 orchiectomies and 2 tumorectomies (2 cells of Leydig). Six patients received post-surgical chemotherapy (mixed tumours). The median tumour size was 1 (0.4-1.5) cm in tumorectomies, and 2.5 (0.5-14) cm in orchiectomies. The mean follow-up was 5 (1-15) years. One patient died due to metastatic disease. There was no local recurrence in the follow up of the tumorectomies.

Conclusion(s): A change in the trend of our therapeutic approach is demonstrated. We propose that testis-sparing surgery is indicated in prepubertal patients who meet the benignity criteria of the testicular mass (small size and negative tumour markers).

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Status

Embase

Institution

(Romo Munoz, Nunez Cerezo, Dore Reyes, Vilanova Sanchez) Departamento de Cirugia Pediatrica, Hospital La Paz, Madrid, Spain (Gonzalez-Peramato) Departamento de Patologia, Hospital La Paz, Madrid, Spain

(Lopez Pereira, Martinez Urrutia) Departamento de Cirugia Pediatrica, Servicio de Urologia infantil, Hospital La Paz, Madrid, Spain

Publisher

Elsevier Doyma

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2018

219.

Phthalate exposure and male reproductive outcomes: A systematic review of the human epidemiological evidence.

Radke E.G., Braun J.M., Meeker J.D., Cooper G.S.

Embase

Environment International. Part 1. 121 (pp 764-793), 2018. Date of Publication: December 2018.

[Review]

AN: 2001188162

Objective: We performed a systematic review of the epidemiology literature to identify the male reproductive effects associated with phthalate exposure. Data sources and study eligibility criteria: Six phthalates were included in the review: di(2-ethylhexyl) phthalate (DEHP), diisononyl phthalate (DINP), dibutyl phthalate (DBP), diisobutyl phthalate (DIBP), butyl benzyl phthalate (BBP), and diethyl phthalate (DEP). The initial literature search (of PubMed, Web of Science, and Toxline) included all studies of male reproductive effects in humans, and outcomes were selected for full systematic review based on data availability. Study evaluation and synthesis methods: For each outcome, studies were evaluated using criteria defined a priori for risk of bias and sensitivity by two reviewers using a domain-based approach. Evidence was synthesized by outcome and phthalate and strength of evidence was summarized using a structured framework.

Result(s): The primary outcomes reviewed here are (number of included/excluded studies in parentheses): anogenital distance (6/1), semen parameters (15/9), time to pregnancy (3/5), testosterone (13/8), timing of pubertal development (5/15), and hypospadias/cryptorchidism (4/10). Looking at the overall hazard, there was robust evidence of an association between DEHP and DBP exposure and male reproductive outcomes; this was based primarily on studies of anogenital distance, semen parameters, and testosterone for DEHP and semen parameters and time to pregnancy for DBP. There was moderate evidence of an association between DINP and BBP exposure and male reproductive outcomes based on testosterone and semen parameters for DINP and semen parameters and time to pregnancy for BBP. DIBP and DEP were considered to have slight evidence of an association. For DIBP, the less conclusive evidence was attributed to a more limited literature base (i.e., fewer studies) and lower exposure levels in the population, decreasing the ability to observe an effect. For DEP, the findings were consistent with experimental animal data that suggest DEP does not have as strong an anti-androgenic effect as other phthalates. Conclusions and implications of key findings: Overall, despite some inconsistencies across phthalates in the specific outcomes associated with exposure, these results support that phthalate exposure at levels seen in human populations may have male reproductive effects, particularly DEHP and DBP. The relative strength of the evidence reflects differing levels of toxicity as well as differences in the range of exposures studied and the number of available studies. The views expressed are those of the authors and do not necessarily represent the views or policies of the U.S. EPA.

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Status

Embase

Institution

(Radke, Cooper) U.S. Environmental Protection Agency, National Center for Environmental Assessment, United States (Braun) Brown University, School of Public Health, United States

(Meeker) University of Michigan, School of Public Health, United States

(Cooper) The Innocence Project, United States

Publisher

Elsevier Ltd

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2018

220.

Secular trends in the incidence and timing of surgical intervention for congenital undescended testis and surgically treated hypospadias in Ontario, Canada between 1997 and 2007.

Dave S., Liu K., Garg A.X., Shariff S.Z.

Embase

Journal of Pediatric Urology. 14 (6) (pp 552.e1-552.e7), 2018. Date of Publication: December 2018.

[Article]

AN: 2000986583

Introduction: Recent studies have suggested contradictory trends in the incidence of undescended testis (UDT) and hypospadias (HYP), partly because of methodological issues and ascertainment bias. The recently described association of "testicular dysgenesis syndrome" links concomitant UDT and HYP, with decreasing sperm counts and testicular cancer. Current guidelines suggest that orchidopexy for UDT should be performed by 18 months of age.

Objective(s): We conducted a retrospective population-based cohort study to estimate the incidence of UDT, HYP, and concomitant UDT and HYP in Ontario, based on a surgical procedure performed in the 5 years after birth. We hypothesized that the incidence of UDT and HYP are stable in the province of Ontario, Canada, over an 11-year time period. Study design: Linked administrative databases held at the Institute of Clinical Evaluative Sciences (ICES) in the province of Ontario, were used to identify all live male newborns between 1997 and 2007. Incidence rates of UDT, HYP and concomitant UDT and HYP were calculated by identifying a surgical procedure for these anomalies, within 5 years of birth. Incidence trends were analyzed using the Cochran Armitage test for trend. Age at surgery for surgical intervention for an orchidopexy or HYP repair was determined.

Result(s): The incidence of UDT, defined by an orchidopexy within 5 years of birth, has remained stable in Ontario, Canada (8.2/1000 male live births, p-value for trend 0.9, 95% CI 8.0-8.4). The incidence of hypospadias has similarly remained stable (3.8/1000 male live births, p-value for trend 0.8, 95% CI 3.7-3.9). The incidence of concomitant UDT and HYP repair showed a significant increase over the 11-year period (0.2/1000 male live births, p-value for trend 0.03, 95% CI 0.2-0.3). The median age at orchidopexy (23 months, IQR 16-34 months) was beyond guideline recommendations, with earlier orchidopexy in recent years. The median age at hypospadias repair was 17 months (IQR 12-26 months).

Discussion(s): The variable rates of incidence for UDT and HYP can be explained by variations in study methodology and differing data sources utilized. The current study uses a surgical procedure to minimize information bias to correctly identify index cases of UDT and HYP.

Conclusion(s): The incidence of undescended testis and hypospadias, over 5 years after birth, has remained stable in the province of Ontario between 1997 and 2007 (Summary Table).

Concomitant UDT and HYP incidence showed a significant increase over this time period. Most boys in Ontario, Canada, undergo orchidopexy beyond 18 months of age. [Table presented]

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Author NameID

Dave, Sumit; ORCID: <http://orcid.org/0000-0003-1462-647X>

Institution

(Dave) Department of Surgery and Paediatrics, Division of Urology, Western University, London, Ontario, Canada (Dave, Liu, Garg, Shariff) Institute for Clinical Evaluative Sciences, London, Ontario, Canada

(Garg, Shariff) Arthur Labatt Family School of Nursing, Western University, London, Ontario, Canada

(Garg, Shariff) Lawson Health Research Institute, London, Ontario, Canada

(Garg, Shariff) Department of Medicine, Division of Nephrology, Western University, London, Ontario, Canada

(Garg, Shariff) Department of Epidemiology and Biostatistics, Western University, London, Ontario, Canada

Publisher

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Year of Publication

2018

Isolated hypospadias: The impact of prenatal exposure to pesticides, as determined by meconium analysis.

Haraux E., Tourneux P., Kouakam C., Stephan-Blanchard E., Boudailliez B., Leke A., Klein C., Chardon K.

Embase

Environment International. 119 (pp 20-25), 2018. Date of Publication: October 2018.

[Article]

AN: 2000869422

Although endocrine-disrupting chemicals (EDCs, including pesticides) are thought to increase the risk of hypospadias, no compounds have been formally identified in this context. Human studies may now be possible via the assessment of meconium as a marker of chronic prenatal exposure. The objective of the present study was to determine whether or not prenatal exposure to pesticides (as detected in meconium) constitutes a risk factor for isolated hypospadias. In a case-control study performed between 2011 and 2014 in northern France, male newborns with isolated hypospadias (n = 25) were matched at birth with controls (n = 58). Newborns with obvious genetic or hormonal anomalies, undescended testis, micropenis, a congenital syndrome or a family history of hypospadias were not included. Neonatal and parental data were collected. Foetal exposure was assessed by determining the meconium concentrations of the pesticides or metabolites (organophosphates, carbamates, phenylurea, and phenoxyherbicides) most commonly used in the region. Risk factors were assessed in a multivariate analysis. The pesticides most commonly detected in meconium were organophosphates (in up to 98.6% of samples, depending on the substance) and phenylurea (>85.5%). A multivariate analysis revealed an association between isolated hypospadias and the presence in meconium of the phenylurea herbicide isoproturon and of the phenoxyherbicide 2-methyl-4-chlorophenoxyacetic acid (odds ratio [95% confidence interval]: 5.94 [1.03-34.11] and 4.75 [1.20-18.76]) respectively). We conclude that prenatal exposure to these two herbicides (as assessed by meconium analysis) was correlated with the occurrence of isolated hypospadias. The results of our case-control study (i) suggest that prenatal exposure to pesticides interferes with the development of the male genitalia, and (ii) emphasize the importance of preventing pregnant women from being exposed to EDCs in general and pesticides in particular.

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Institution

(Haraux, Klein) Paediatric Surgery Unit, Amiens University Hospital, Amiens F-80054, France
(Haraux, Tourneux, Kouakam, Stephan-Blanchard, Leke, Chardon) PeriTox - UMI 01, UFR de
Medecine, Jules Verne University of Picardy, Amiens F-80054, France
(Tourneux) Paediatric Intensive Care Unit, Amiens University Hospital, Amiens F-80054, France
(Boudailliez) Department of Paediatrics, Amiens University Hospital, Amiens F-80054, France
(Leke) Neonatal Intensive Care Unit, Amiens University Hospital, Amiens F-80054, France
Publisher
Elsevier Ltd
Year of Publication
2018

222.

Nationwide Increase in Cryptorchidism After the Fukushima Nuclear Accident.

Murase K., Murase J., Machidori K., Mizuno K., Hayashi Y., Kohri K.

Embase

Urology. 118 (pp 65-70), 2018. Date of Publication: August 2018.

[Article]

AN: 2000811106

Objective: To estimate the change of discharge rate after cryptorchidism surgery between pre- and postdisaster in Japan. Cryptorchidism cannot be diagnosed before birth and is not a factor that would influence a woman's decision to seek an abortion. Therefore, this disease is considered suitable for assessing how the Great East Japan Earthquake and the subsequent Fukushima Daiichi nuclear accident (2011) influenced congenital diseases.

Material(s) and Method(s): We obtained cryptorchidism discharge data collected over 6 years from hospitals that were included in an impact assessment survey of the Diagnosis Procedure Combination survey database in Japan and used these data to estimate the discharge rate after cryptorchidism surgery before and after the disaster. The 94 hospitals in Japan that participated in Diagnosis Procedure Combination system and had 10 or more discharges after cryptorchidism surgery within successive 6 years covering pre- and postdisaster period (FY2010-FY2015) were involved. The change in discharge rate between pre- and postdisaster was analyzed using a Bayesian generalized linear mixed model.

Result(s): Nationwide, a 13.4% (95% credible interval 4.7%-23.0%) increase in discharge rates was estimated. The results of all sensitivity analyses were similar to the reported main results.
Conclusion(s): The discharge rate of cryptorchidism was increased nationwide. The rates of low-weight babies or preterm births, risk factors of cryptorchidism, were almost constant during the study period, and age distribution of the surgery was also not changed, which suggested that the other factors that associated with the disaster increased the incidence of cryptorchidism.

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Status

Embase

Institution

(Murase) Nagoya City University Graduate School of Natural Sciences, Nagoya, Japan

(Murase) Goshawk Protection Fund, Tochigi, Japan

(Machidori) Machidori Clinic, Tokyo, Japan

(Mizuno, Hayashi) Department of Pediatric Urology, Nagoya City University Graduate School of Medical Sciences, Nagoya, Japan

(Kohri) Department of Nephro-Urology, Nagoya City University Graduate School of Medical Sciences, Nagoya, Japan

Publisher

Elsevier Inc. (E-mail: usjcs@elsevier.com)

Year of Publication

2018

223.

Birth defects in children of men exposed in utero to diethylstilbestrol (DES).

Tournaire M., Devouche E., Epelboin S., Cabau A., Dunbavand A., Levadou A.

Embase

Therapie. 73 (5) (pp 399-407), 2018. Date of Publication: October 2018.

[Article]

AN: 2000614968

Objective: Prenatal exposure to diethylstilbestrol (DES) is associated with adverse effects, including genital anomalies and cancers in men and women. Animal studies showed birth defects and tumors in the offspring of mice prenatally exposed to DES. In humans, birth defects, such as hypospadias were observed in children of prenatally exposed women. The aim of this research was to assess the birth defects in children of prenatally exposed men.

Method(s): In a retrospective study conceived by a patients' association (Reseau DES France), the reports of men prenatally exposed to DES on adverse health effects in their children were compared with those of unexposed controls and general population.

Result(s): An increased incidence of two genital anomalies, cryptorchidism (OR = 5.72; 95% CI 1.51-21.71), and hypoplasia of the penis (OR = 22.92; 95% CI 3.81-137.90), was observed in the 209 sons of prenatally exposed men compared with controls, but hypospadias incidence was not increased in comparison with either the controls or the general population. No increase of genital anomalies was observed in daughters.

Conclusion(s): With caution due to the methods and to the small numbers of defects observed, this work suggests an increased incidence of two male genital tract defects in sons of men prenatally exposed to DES. This transgenerational effect, already observed in animals and in the offspring of women prenatally exposed to DES, could be the result of epigenetic changes transmitted to the subsequent generation through men.

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Embase

Author NameID

Devouche, Emmanuel; ORCID: <http://orcid.org/0000-0003-3987-1842> Dunbavand, Annabel;

ORCID: <http://orcid.org/0000-0002-2879-0820>

Institution

(Tournaire, Cabau) Assistance publique-Hopitaux de Paris, Paris 75014, France (Tournaire, Devouche) Universite Paris Descartes, Paris 75006, France

(Devouche) Laboratoire psychopathologie et processus de sante EA4057, Boulogne-Billancourt 92100, France

(Epelboin) Assistance publique-Hopitaux de Paris, Hopital Bichat-Claude Bernard, Paris 75018, France

(Epelboin) Universite Paris 7 Diderot, Paris 75013, France

(Dunbavand) Mutualite francaise, Paris 75015, France

(Levadou) Association reseau DES France, Mont-de-Marsan 40000, France

Publisher

Elsevier Masson SAS (62 rue Camille Desmoulins, Issy les Moulineaux Cedex 92442, France)

Year of Publication

2018

224.

The volume of unilaterally undescended testis after hCG therapy compared to orchidopexy and combined methods.

Niedzielski J., Kucharski P., Slowikowska-Hilczer J.

Embase

Andrology. 6 (5) (pp 742-747), 2018. Date of Publication: September 2018.

[Article]

AN: 624541754

The aim of the study was to compare the effects of human chorionic gonadotropin (hCG) therapy with those of surgical or combined therapy on testicular volume (TV) in boys at different ages with unilateral canalicular undescended testis (UDT). In total, 155 boys aged 1 to 12 years were treated: either surgically (ST), or by 50 IU/kg body weight hCG administration every three days for five weeks (HT), or by a combination of the two. The patients underwent ultrasound examination of TV before the treatment, 9-12 (median 10) and 24-39 months (median 32) after therapy. The testicular atrophy index (TAI) of the affected testicle was calculated. The success rate was 94.7% for ST, 39.2% for HT and 98% for HST patients. The atrophy rate was 5.3% for ST, 0% for HT and 2% for HST. Neither treatment type nor patient age significantly influenced gonadal atrophy. No significant differences in TV of the affected testis were observed after treatment between the groups. The TAI values were significantly the lowest in HT group ($p = 0.0006$). Both TV and TAI changes from the baseline values did not differ between the treatment groups. At the 24- to 39-month follow-up, no significant differences were observed in the change in baseline TV and baseline TAI between age groups. TV of the affected testis increased significantly ($p = 0.0000$), and TAI decreased significantly over time ($p = 0.01$), with no significant differences depending on the age group, treatment type or the interaction of the two factors. The hCG therapy did not impair the development of affected and healthy testes, neither as single nor as neoadjuvant therapy, both during early assessment and after 2-3 years. Patients' age at the initiation of treatment seems irrelevant.

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Embase

Institution

(Niedzielski, Kucharski) Department of Pediatric Surgery and Urology, the Maria Konopnicka Paediatric Centre, Medical University of Lodz, Lodz, Poland (Slowikowska-Hilczer) Department of Andrology and Reproductive Endocrinology, Medical University of Lodz, Lodz, Poland

Publisher

Blackwell Publishing Ltd

Year of Publication

2018

225.

Adolescence and andrologist: An imperfect couple.

Olana S., Mazzilli R., Delfino M., Zamponi V., Iorio C., Mazzilli F.

Embase

Archivio Italiano di Urologia e Andrologia. 90 (3) (pp 208-211), 2018. Date of Publication: 30 Sep 2018.

[Article]

AN: 625184266

Objective: The aims of this research were to study: a) the prevalence of male adolescents, aged between 10 and 19 years of age, referred to our Unit for an andrological assessment; b) the reasons (stated and subsequently modified) for referral; c) the prevalence of clinically diagnosed diseases.

Material(s) and Method(s): A total of 2.855 subjects, referred to the Andrology Unit for a first examination, were retrospectively studied. For each adolescent, a medical history was taken and an andrological physical examination was carried out.

Result(s): Prevalence was found to be 6.9% (197/2855). Subjects were divided into two groups according to age (A: ≤ 14 and B: ≥ 15 years). The original reason stated for their consultation was corrected by 11.7% of the subjects (23/197); this correction concerned almost all the Group B subjects (21/23 (91.3%) vs 2/23 (8.7%) of Group A; $p < 0.01$). Regarding sexual dysfunctions, a

simple explanation of certain conditions reassured the subject in about 15% of the cases. Furthermore, the physical examination proved extremely useful, revealing clinical alterations in more than 60% of subjects.

Conclusion(s): In conclusion, to date in Italy, the prevalence of adolescents among males referred to an Andrology Unit for assessment is very low. It is important to encourage adolescents to undergo andrological examination to enable identification of reproductive function and psycho-sexual disorders.

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Institution

(Olana, Mazzilli, Delfino, Zamponi, Iorio, Mazzilli) Andrology Unit, Department of Clinical and Molecular Medicine, Sant'Andrea Hospital, University of Rome "Sapienza", Rome, Italy

Publisher

Edizioni Scripta Manent s.n.c. (E-mail: scriman@tin.it)

Year of Publication

2018

226.

Gonadal function and pubertal development in patients with Silver-Russell syndrome.

Goedegebuure W.J., Smeets C.C.J., Renes J.S., De Rijke Y.B., Hokken-Koelega A.C.S.

Embase

Human Reproduction. 33 (11) (pp 2122-2130), 2018. Date of Publication: 2018.

[Article]

AN: 625128439

STUDY QUESTION: Is gonadal function affected in males and females with Silver-Russell Syndrome (SRS)? SUMMARY ANSWER: Sertoli cell dysfunction is more common in males with SRS, with 11p15 LOM, but gonadal function seems to be unaffected in females with SRS. WHAT IS KNOWN ALREADY: Males with SRS have an increased risk for genital abnormalities such as cryptorchidism and hypospadias, which could be associated with reproductive problems in later

life. In SRS females, an association has been described with Mayer-Rokitansky-Kuster-Hauser syndrome, which might compromise their reproductive function. STUDY DESIGN, SIZE, DURATION: Longitudinal follow-up study, involving 154 subjects, over a time period of 20 years. PARTICIPANTS/MATERIALS, SETTING, METHODS: Thirty-one SRS patients (14 males) and 123 non-SRS patients born at same gestational age (SGA; 65 males). All received growth hormone and 27.3% received additional gonadotropin-releasing hormone analog treatment (GnRHa). MAIN RESULTS AND THE ROLE OF CHANCE: Mean age at onset of puberty was 11.5 years in SRS males versus 11.6 years in non-SRS males ($P = 0.51$), and 10.5 years in SRS females versus 10.7 years in non-SRS females ($P = 0.50$). Four of the 14 SRS males had a post-pubertal inhibin-B level below the fifth percentile compared to healthy controls, and two of them an FSH above the 95th percentile, indicating Sertoli cell dysfunction. One of them had a history of bilateral cryptorchidism and orchiopexy. All SRS females had AMH, LH and FSH levels within the reference range. Pubertal duration to Tanner stage five was similar in SRS and non-SRS. Pubertal height gain was better in SRS patients who additionally received GnRHa ($P < 0.01$). Mean age at menarche was 13.1 years in SRS versus 13.3 years in non-SRS ($P = 0.62$). One SRS female had primary amenorrhea due to Mullerian agenesis. LIMITATIONS, REASONS FOR CAUTION: As this is a rare syndrome, the SRS group had a small size. WIDER IMPLICATIONS OF THE FINDINGS: As gonadal function is not affected in females with SRS, it is likely that reproductive function is also not affected. Sertoli cell dysfunction in males with SRS could cause impaired reproductive function and should be assessed during pubertal development. STUDY FUNDING/COMPETING INTEREST(S): No external funding was used for the study. The authors have no conflicts of interest.

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Institution

(Goedegebuure, Hokken-Koelega) Dutch Growth Research Foundation, Rotterdam 3016AH, Netherlands (De Rijke) Department of Clinical Chemistry, Erasmus University Medical Centre, Rotterdam 3015GD, Netherlands

(Goedegebuure, Smeets, Renes, Hokken-Koelega) Department of Pediatrics, Subdivision of Endocrinology, Erasmus University Medical Centre, Wytemaweg 80, Rotterdam 3015 CN, Netherlands

Publisher

Oxford University Press

Year of Publication

2018

227.

Modified 2-port laparoscopic herniorrhaphy with Kirschner wire in children A retrospective review.

Cao Z., Chen J., Li Z., Li G.

Embase

Medicine (United States). 97 (42) (no pagination), 2018. Article Number: e12790. Date of Publication: 01 Oct 2018.

[Review]

AN: 627460077

Background: Pediatric inguinal hernia is one of the most common diseases in children, and laparoscopy is the main surgical method. This study aims to evaluate the efficacy of a new modified 2-port laparoscopic herniorrhaphy with Kirschner wire (TLHK) for inguinal hernia in children.

Method(s): A total of 5304 children with inguinal hernia hospitalized at the Jiangmen Center Hospital from June 2003 to May 2016 were enrolled in this retrospective study. Four thousand one hundred thirty-five children underwent TLHK that comprised the observation group, while 1169 received single incision laparoscopy (SIL) as the control group (CG). A propensity score matched cohort study was conducted between these groups. We included all patients who were diagnosed as inguinal hernia and matched comparators with a proportion of 1:1. The propensity score was calculated using logistic regression with forward stepwise selection in 4 variables. The patients' operative details, intra- and postoperative complications, and postoperative hospital stay were analyzed. The follow-up lasted from 1 month to 2 years.

Result(s): Among 5304 potential patients, the propensity score identified 270 (135 TLHK cases and 135 comparators) patients. The age, sex, body mass index, and the hernia type and location did not differ between CG and TLHK. TLHK group had a shorter operative time (unilateral: 17.4 +/- 3.35 minutes vs 20.7 +/- 3.71 minutes; bilateral: 20.4 +/- 5.17 minutes vs 25.2 +/- 5.43 minutes), less complications (2.10% vs 2.65%), lower recurrence rate (0% vs 4.44%), and similar hospital stay (2.3 +/- 1.1 vs 2.1 +/- 1.3) as compared with CG. No iliac vessel injury, spermatic cord vessels injury, vas deferens injury, or iatrogenic cryptorchidism occurred in either of the groups.

Conclusion(s): TLHK is a safe and feasible treatment for inguinal hernia in children due to less invasion and less recurrence rate than SIL.

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Status

Embase

Institution

(Cao, Chen, Li, Li) Department of General Surgery, Nanfang Hospital, Southern Medical University, Guangzhou, China (Cao) Department of Pediatric Surgery, Huangshi Maternity and Children's Health Hospital of Edong Healthcare Group, Huangshi, Hubei, China

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

Year of Publication

2018

228.

Experience of Using Shear Wave Elastography Imaging in Evaluation of Undescended Testes in Children: Feasibility, Reproducibility, and Clinical Potential.

Durmaz M.S., Sivri M., Sekmenli T., Kocaoglu C., Ciftci I.

Embase

Ultrasound Quarterly. 34 (4) (pp 206-212), 2018. Date of Publication: 01 Dec 2018.

[Article]

AN: 625359693

We aimed to determine the difference in tissue stiffness, which might reflect histologic damage, by comparing the potential of the shear wave elastography (SWE) values of operated undescended testes (OUT) with those of undescended testes (UT) and normal testes. A total of 120 patients (235 testes) were enrolled in the current study. Quantitative SWE values were measured by manually drawing contours of the entire testis structure with a free region of interest. A group of 66 OUT were classified as group A. Operation age and the period passed over the operation time and SWE values were compared among the testes assigned to group A. The 50 testes having inguinal canal placement were classified as group B, and the 119 testes whose

sonography findings were normal were classified as group C. These 3 groups were compared in terms of the SWE values and volume. The SWE values of group A were significantly higher than those of groups B and C ($P < 0.001$). The SWE values of group B were significantly higher than those of group C ($P < 0.001$). The SWE values of OUT were significantly higher than those of the contralateral normal testes and UT ($P < 0.001$). There were no significant differences among operation age and period passed over the operation time and SWE values in the OUT ($P > 0.05$). There was no significant correlation between SWE values and testes' volume ($P > 0.05$). The SWE technique can be used effectively as a new parameter to assess stiffness of the OUT and UT to predict interstitial fibrosis and the severity of histologic damage.

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Status

Embase

Institution

(Durmaz, Sivri) Department of Radiology, Konya Health Sciences University Teaching and Research Hospital, Fatih Cad No 4/1, Meram, Konya 42090, Turkey (Sekmenli, Ciftci) Faculty of Medicine, Department of Pediatric Surgery, Selcuk University, Konya, Turkey (Kocaoglu) Department of Pediatric Surgery, Konya Health Sciences University Teaching and Research Hospital, Konya, Turkey

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

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229.

Initial laparoscopy and optimized approach for unilateral nonpalpable testis: review of 8-year single-center experience.

Anwar A.Z.M., Fathelbab T.K., Abdelhamid A.M., Galal E.M., Ali M.M., Tawfik E.R.

Embase

International Urology and Nephrology. 50 (12) (pp 2139-2144), 2018. Date of Publication: 01 Dec 2018.

[Article]

AN: 624457913

Purpose: We evaluated the role of initial laparoscopy and optimized approach in cases of unilateral nonpalpable testis.

Method(s): Seventy-four patients with nonpalpable testes were presented. We excluded 9 patients, with palpable testes under anesthesia. Laparoscopy was offered to 65 patients. Contralateral testis hypertrophy with length ≥ 1.8 cm was confirmed in 47 patients. Ultrasound results were available for 35 patients.

Result(s): Age ranged from 1 to 10 years. Of 65 nonpalpable testes, right side comprised 23 (35.4%) and the left 42 (64.6%). Laparoscopy revealed intra-abdominal testis in 18 patients (27.7%), blind-ending vessels and vas in 8 (12.3%), and vas and vessels traversing the internal ring in 39 (60%). Treatment of intra-abdominal testes included Fowler-Stephens orchiopexy in 7 patients, laparoscopic orchiopexy in 9, and laparoscopic orchiectomy in 2. In 8 patients with blind-ending vas and vessels, laparoscopy was terminated. In 39 patients with vas and vessels traversing the internal ring, scrotal exploration was performed in 36 patients with closed internal ring and inguinal exploration in 3 with open internal ring. Vanished testes were present in 43/47(91.5%) of patients with contralateral testis hypertrophy ≥ 1.8 cm. Ultrasound detected the presence of a testis in only 4/11 (36.3%) of patients, although it could not identify vanished testis.

Conclusion(s): Initial laparoscopy should be retained as one of the standard treatment for nonpalpable testis. It was the only required modality in 26 patients (40%) and optimized further treatment in 39 patients (60%) by evaluation of the condition of the internal ring.

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Institution

(Anwar, Fathelbab, Abdelhamid, Galal, Ali, Tawfiek) Urology Department, School of Medicine, University Hospital, Minia 61111, Egypt

Publisher

Springer Netherlands (E-mail: rbk@louisiana.edu)

Year of Publication

2018

230.

Use of ultrasound for the palpable undescended testis: A wasteful practice.

Wayne C., Guerra L.A., Yao J., Keays M.A., Leonard M.P.

Embase

Family Practice. 35 (4) (pp 452-454), 2018. Date of Publication: 2018.

[Article]

AN: 624420897

Background. Many primary care physicians order an ultrasound (US) before referral to specialist care for suspected undescended testis; however, the value of this practice is questionable.

Objective. To determine the proportion of boys referred for suspected undescended testis who had accompanying US, the cost of this practice and the accuracy of US for testis localization when compared with physical examination by a pediatric urologist. Methods. This was a retrospective chart review at a pediatric urology service, including all patients referred for suspected undescended testis from 2008 to 2012. We determined the cost of US ordered, and calculated Cohen's kappa, sensitivity and specificity, and positive and negative predictive value. Results. We identified 894 eligible patients; 32% (289/894) were accompanied by US. In 77% (223/289), the urologist was able to palpate the testis: 51% (147/289) had a normal/retractile testis and 26% (76/289) had a palpable undescended testis. At a cost of 71.10 CAD per US, 20 547.90 CAD was expended on this practice. Of the 223 patients with palpable testes, we were able to gather detailed US and physical examination results for 214 patients. Cohen's kappa was 0.06 (95% CI -0.005, 0.11; P = 0.10). US had 92.8% sensitivity (95% CI 84.1, 96.9%) and 15.2% specificity (95% CI 10.2, 21.9%) to detect an undescended testis. The positive predictive value was 34.2% (95% CI 27.8, 41.3%), while the negative predictive value was 81.5% (95% CI 63.3, 91.8%). Conclusions. Referral of patients for suspected undescended testis should not be accompanied by an US study as US is not useful in these cases.

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Institution

(Wayne, Guerra, Yao, Keays, Leonard) Division of Urology, Department of Surgery, Children's Hospital of Eastern Ontario, 401 Smyth Rd, Ottawa K1H 8L1, Canada (Guerra, Yao, Keays, Leonard) Department of Surgery, Faculty of Medicine, University of Ottawa, Ottawa, Canada

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Oxford University Press

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231.

Associations between hypospadias, cryptorchidism and anogenital distance: Systematic review and meta-analysis.

Hua X.-G., Hu R., Hu C.-Y., Li F.-L., Jiang W., Zhang X.-J.

Embase

Andrologia. 50 (10) (no pagination), 2018. Article Number: e13152. Date of Publication: December 2018.

[Review]

AN: 624023859

Hypospadias and cryptorchidism are potential manifestations of testicular dysgenesis syndrome (TDS) at birth. Anogenital distance (AGD) has been presumed as an indicator related to endocrine disruptors proposed as one of the pathogenetic mechanisms underlying male reproductive disorders. In humans, recent studies have correlated AGD in boys to testicular anomalies. However, the associations between hypospadias, cryptorchidism and AGD remain inconsistent and have not been combined. Hence, we conducted a meta-analysis to assess gradations in the severity of the endocrine disruption in cryptorchidism or hypospadias by using AGD. A total of 2,119 boys from five birth cohort studies and two cross-sectional studies were subjected to meta-analysis. Random-effect model was used to calculate the standardised mean difference (SMD) of AGD. Our results reveal that boys with hypospadias or cryptorchidism have shorter AGD ([SMD, -2.63; 95% CI, -4.65 to -0.62] and [SMD, -0.69; 95% CI, -1.36 to -0.02]) respectively. There was no indication of a publication bias either from the result of Egger's test or Begg's test for hypospadias and cryptorchidism.

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Author NameID

Zhang, Xiu-Jun; ORCID: <http://orcid.org/0000-0002-8123-185X>

Institution

(Hua, Hu, Li, Jiang, Zhang) Department of Epidemiology and Biostatistics, School of Public Health, Anhui Medical University, Hefei, Anhui, China (Hu) School of Health Services Management, Anhui Medical University, Hefei, Anhui, China

Publisher

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2018

232.

A history of undescended testes in young men with Klinefelter syndrome does not reduce the chances for successful microsurgical testicular sperm extraction.

Ragab M.W., Cremers J.-F., Zitzmann M., Nieschlag E., Kliesch S., Rohayem J.

Embase

Andrology. 6 (4) (pp 525-531), 2018. Date of Publication: July 2018.

[Article]

AN: 623344806

Klinefelter syndrome (KS) and undescended testes (UDT) are known etiologies for non-obstructive azoospermia (NOA), and coexistence of both etiologies is not uncommon. Patients with both KS and a history of UDT are therefore considered to have extremely reduced chances for paternity. We aimed to analyze the impact of previous surgically corrected unilateral or bilateral UDT on sperm retrieval rates (SRRs) by microsurgical testicular sperm extraction (mTESE) in azoospermic men with KS. Age, testicular volumes, and hypothalamo-pituitary-gonadal axis function were investigated in relation to SRRs in 29 non-mosaic KS patients (47,XXY) with a history of UDT (group 1) who underwent mTESE between 2008 and 2016 in our center and compared to the data of age- and serum testosterone-matched non-mosaic KS controls with eutopic testes at birth (group 2), and to those of 51 men with NOA and a normal male karyotype (46,XY), but previous UDT (group 3). SRRs in KS patients with surgically corrected UDT during childhood were comparable to SRRs of KS patients with eutopic testes at birth: 31% (35% in unilateral and 22% in bilateral UDT) vs. 38% ($p = 0.581$). SRRs and Leydig cell function in group 1 were negatively correlated with age. Significantly higher SRRs (66%) were

found in euploid azoospermic men with surgically corrected UDT ($p < 0.001$). A history of UDT does not preclude chances for future fatherhood in young azoospermic males with KS. In one of three men with previous unilateral UDT and in one of 4-5 in those with previous bilateral UDT, spermatozoa can be harvested by mTESE during late adolescence or young adulthood for immediate or future use in assisted reproduction.

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Author NameID

Ragab M.W.; ORCID: <http://orcid.org/0000-0001-6139-7513>

Institution

(Ragab, Cremers, Zitzmann, Nieschlag, Kliesch, Rohayem) Department of Clinical and Surgical Andrology, Center of Reproductive Medicine and Andrology, University of Munster, Munster, Germany (Ragab) Department of Clinical and Surgical Andrology, Cairo University, Cairo, Egypt

Publisher

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Year of Publication

2018

233.

Obstetric outcomes and effects on babies born to women treated for epilepsy during pregnancy in a resource limited setting: A comparative cohort study.

Galappatthy P., Liyanage C.K., Lucas M.N., Jayasekara D.T.L.M., Abhayaratna S.A., Weeraratne C., De Abrew K., Gunaratne P.S., Gamage R., Wijeyaratne C.N.

Embase

BMC Pregnancy and Childbirth. 18 (1) (no pagination), 2018. Article Number: 230. Date of Publication: 14 Jun 2018.

[Article]

AN: 622563037

Background: Management of epilepsy during pregnancy in a resource-limited setting (RLS) is challenging. This study aimed to assess obstetric outcomes and effects on babies of women with epilepsy (WWE) exposed to Anti-epileptic drugs (AEDs) compared to non-exposed controls in a RLS.

Method(s): Pregnant WWE were recruited from antenatal and neurology clinics of a tertiary care hospitals in Sri Lanka. Patients were reviewed in each trimester and post-partum. Medication adherence, adverse effects, seizure control and carbamazepine blood levels were monitored. Post-partum, measurements for anthropometric and dysmorphic features of the babies and congenital abnormalities were recorded. Age and sex matched babies not exposed to AED recruited as controls were also examined.

Result(s): Ninety-six pregnant WWE were recruited (mean period of gestation 22.9 weeks). Mean age was 28 years and 48(50%) were primigravidae. Fifty percent (48) were on monotherapy, while 23.8, 15.9 and 4.1% were on two, three and four AEDs respectively. AEDs in first trimester (TM1) were carbamazepine (71%), valproate (25.8%) clobazam (29.5%), lamotrigine (7%) topiramate (5%) and others (3.4%). Sodium valproate use reduced significantly from T1 to T2($p < 0.05$). Sub-therapeutic carbamazepine levels correlated positively ($r = 0.547$) with poor medication adherence ($p = 0.009$) and negatively ($r = 0.306$) with adverse effects ($p = 0.002$).

Seventy-six WWE completed follow-up reporting w 75 (98.6%) live births and one T1 miscarriage (1.3%). Three (4.3%) were preterm. Majority (73.33%) were normal vaginal deliveries. Cesarean sections were not increased in WWE. Fifty-nine (61.45%) babies were examined. For those examined during infancy, 53 age and sex matched controls were recruited and examined.

Congenital abnormalities occurred in 5 (9.43%) babies of WWE [atrio-ventricular septal defect (2), renal hypoplasia (1), cryptorchidism (1), microcephaly (1)] compared to 2 (3.77%) in controls (2 microcephaly; $p = 0.24$). Fetal exposure to AEDs increased a risk of low birth weight (RR 2.8; $p = 0.049$). Anthropometric parameters of AED exposed babies were lower at birth but not statistically significant between the two groups (weight $p = 0.263$, length $p = 0.363$, occipito-frontal circumference (OFC) $p = 0.307$). However, weight ($p = 0.009$), length ($p = 0.016$) and OFC ($p = 0.002$) were significantly lower compared to controls at an average of 3.52 months.

Conclusion(s): Most pregnancies are unplanned in the RLS studied, and AEDs were altered during pregnancy. Congenital anomalies occurred at rates comparable to previous reports. Fetal exposure to AED had growth retardation in infancy compared to non-exposed babies.

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Institution

(Galappatthy, Liyanage, Jayasekara, Abhayaratna, Weeraratne, De Abrew) University of Colombo, Department of Pharmacology, Faculty of Medicine, Colombo, Sri Lanka (Lucas) University of Colombo, Department of Paediatrics, Faculty of Medicine, Colombo, Sri Lanka (Gunaratne, Gamage) National Hospital of Sri Lanka, Colombo, Sri Lanka (Wijeyaratne) Faculty of Medicine, University of Colombo, Department of Obstetrics and Gynecology, Colombo, Sri Lanka

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Year of Publication

2018

234.

Study of Ras/MAPK pathway gene variants in Chilean patients with Cryptorchidism.

Rodriguez F., Vallejos C., Ponce D., Unanue N., Hernandez M.I., Celis S., Arcos K., Belmar F., Lopez M.T., Cassorla F.

Embase

Andrology. 6 (4) (pp 579-584), 2018. Date of Publication: July 2018.

[Article]

AN: 622125390

Cryptorchidism is one of the most common congenital disorders in boys, and several genetic, hormonal, and environmental factors have been proposed as possible causes for this genitourinary defect. Genetic factors have been intensively searched, but relatively few pathogenic variants have been described. Cryptorchidism is a frequent finding in patients with RASopathies, a group of syndrome caused by mutations in genes of the Ras/MAPK pathway. Our aim was to determine whether patients with isolated cryptorchidism (IC) exhibit Ras/MAPK pathway gene variants associated with RASopathies. Two hundred thirty-nine patients with IC were recruited after orchidopexy. Determination of Ras/MAPK pathway gene variants was performed by high-resolution melting (HRM) analysis followed by sequencing. Restriction or allele-specific amplification assay was applied to (i) variant confirmation; (ii) search in healthy controls; and (iii) segregation analysis. Controls correspond to 100 healthy Chilean adults without a history of cryptorchidism. Molecular analysis showed one synonymous substitution (BRAF_p.Q456Q) in two patients and four missense substitutions (SOS1_p.R497Q, BRAF_

p.F595L, NRAS_ p.T50I, and MAP2K2_ p.Y134C) in five patients. Our results suggest that some patients with isolated cryptorchidism, but with no evidence of dysmorphic features suggestive of RASopathies, may harbor Ras/MAPK pathway gene alterations.

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Author NameID

Rodriguez F.; ORCID: <http://orcid.org/0000-0003-0461-7393>

Institution

(Rodriguez, Vallejos, Ponce, Unanue, Hernandez, Arcos, Belmar, Cassorla) Institute of Maternal and Child Research, School of Medicine, University of Chile, Santiago, Chile (Celis, Lopez)

Pediatric Urology Department, Hospital Clinico San Borja - Arriaran, Santiago, Chile

Publisher

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2018

235.

THE EFFECT OF A HYPNOTIC-BASED ANIMATED VIDEO ON STRESS AND PAIN REDUCTION IN PEDIATRIC SURGERY.

Arnon Z., Hanan H., Mogilner J.

Embase

International Journal of Clinical and Experimental Hypnosis. 66 (2) (pp 123-133), 2018. Date of Publication: 03 Apr 2018.

[Article]

AN: 621544933

Presurgical stress and its negative influences on postsurgical recovery and pain are well documented in the medical literature. Hence, the reduction of stress is advisable. The present study aimed to reduce stress using a hypnotic-based animated video. Thirty children aged 3 to 16 years hospitalized for ambulatory surgery for undescended testes or umbilical/inguinal hernia

were recruited for the study. They watched the video 1 time prior to surgery in the presence of their parents and reported their anxiety and pain pre- and postvideo watching on a visual analogue scale. The results show a statistically significant reduction in both anxiety and pain. The article describes the structuring of the animated video and includes links to English, Hebrew, and Arabic versions of it.

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Institution

(Arnon, Mogilner) Bnai Zion Medical Center, Haifa, Israel (Arnon, Hanan) the Max Stern Yezreel Valley College, Israel

Publisher

Routledge (E-mail: info@tandf.co.uk)

Year of Publication

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236.

Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinstein-Taybi syndrome.

Menke L.A., Gardeitchik T., Hammond P., Heimdal K.R., Houge G., Hufnagel S.B., Ji J., Johansson S., Kant S.G., Kinning E., Leon E.L., Newbury-Ecob R., Paolacci S., Pfundt R., Ragge N.K., Rinne T., Ruivenkamp C., Saitta S.C., Sun Y., Tartaglia M., Terhal P.A., van Essen A.J., Vigeland M.D., Xiao B., Hennekam R.C.

Embase

American Journal of Medical Genetics, Part A. 176 (4) (pp 862-876), 2018. Date of Publication: April 2018.

[Article]

AN: 620750217

In 2016, we described that missense variants in parts of exons 30 and 31 of CREBBP can cause a phenotype that differs from Rubinstein-Taybi syndrome (RSTS). Here we report on another 11

patients with variants in this region of CREBBP (between bp 5,128 and 5,614) and two with variants in the homologous region of EP300. None of the patients show characteristics typical for RSTS. The variants were detected by exome sequencing using a panel for intellectual disability in all but one individual, in whom Sanger sequencing was performed upon clinical recognition of the entity. The main characteristics of the patients are developmental delay (90%), autistic behavior (65%), short stature (42%), and microcephaly (43%). Medical problems include feeding problems (75%), vision (50%), and hearing (54%) impairments, recurrent upper airway infections (42%), and epilepsy (21%). Major malformations are less common except for cryptorchidism (46% of males), and cerebral anomalies (70%). Individuals with variants between bp 5,595 and 5,614 of CREBBP show a specific phenotype (ptosis, telecanthi, short and upslanted palpebral fissures, depressed nasal ridge, short nose, anteverted nares, short columella, and long philtrum). 3D face shape demonstrated resemblance to individuals with a duplication of 16p13.3 (the region that includes CREBBP), possibly indicating a gain of function. The other affected individuals show a less specific phenotype. We conclude that there is now more firm evidence that variants in these specific regions of CREBBP and EP300 result in a phenotype that differs from RSTS, and that this phenotype may be heterogeneous.

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Author NameID

Menke, Leonie A.; ORCID: <http://orcid.org/0000-0002-8312-7730> Ji, Jianling; ORCID: <http://orcid.org/0000-0003-4941-3538>

Paolacci, Stefano; ORCID: <http://orcid.org/0000-0002-0946-0543>

Xiao, Bing; ORCID: <http://orcid.org/0000-0002-3736-6714>

Terhal, Paulien A.; ORCID: <http://orcid.org/0000-0002-4417-4307>

Institution

(Menke, Hennekam) Department of Pediatrics, Academic Medical Center, Amsterdam, Netherlands Wellcome Trust Sanger Institute, Wellcome Genome, Campus, United Kingdom

(Gardeitchik, Pfundt, Rinne) Department of Human Genetics, Radboud University Medical Center, Nijmegen, Netherlands

(Hammond) Big Data Institute and Nuffield Department of Obstetrics and Gynaecology, University of Oxford, Oxford, United Kingdom

(Heimdal, Vigeland) Department of Medical genetics, Oslo University Hospital, Oslo, Norway

(Houge, Johansson) Center for medical genetics and molecular medicine, Haukeland University Hospital, Bergen, Norway

(Hufnagel, Leon) Division of Genetics and Metabolism, Children's National Health System, Columbia, WA, United States
(Ji, Saitta) Division of Genomic Medicine, Department of Pathology, Children's Hospital Los Angeles and Keck USC School of Medicine, Los Angeles, CA, United States
(Johansson) K.G. Jebsen Centre for Neuropsychiatric Disorders, The Department of Clinical Science, University of Bergen, Bergen, Norway
(Kant, Ruivenkamp) Department of Clinical Genetics, Leiden University Medical Center, Leiden, Netherlands
(Kinning) West of Scotland Genetics Service, Queen Elizabeth University Hospitals, Glasgow, United Kingdom
(Newbury-Ecob) Department of Clinical Genetics, University Hospitals Bristol, Bristol, United Kingdom
(Paolacci) Department of Experimental Medicine, Sapienza, University of Rome Rome, Italy
(Ragge) Birmingham Women's Hospital, Birmingham, United Kingdom
(Sun, Xiao) Department of Pediatric Endocrinology/Genetics, Xinhua Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai Institute for Pediatric Research, Shanghai, China
(Tartaglia) Genetics and Rare Diseases Research Division, Ospedale Pediatrico Bambino Gesù, Rome, Italy
(Terhal) Department of Genetics, Wilhelmina Children's Hospital, Utrecht, Netherlands
(van Essen) University of Groningen, University Medical Center Groningen, Department of Genetics, Groningen, Netherlands

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237.

Comparison of penile length at 6-24 months between children with unilateral cryptorchidism and a healthy normal cohort.

Ryu D.S., Cho W.Y., Chung J.M., Kang D.I., Lee S.D., Park S.

Embase

Investigative and Clinical Urology. 59 (1) (pp 55-60), 2018. Date of Publication: January 2018.

[Article]

AN: 620138314

Purpose: Urologic diseases affected by testosterone can be associated with smaller penis size compared to the normal population. We sought to compare penile length in children with unilateral cryptorchidism and normative data from a cohort of healthy Korean boys.

Material(s) and Method(s): This study was performed in 259 Korean boys (212, normal cohort; 47, cryptorchidism) aged 6-24 months, each of whom had been brought to an outpatient clinic at one of five tertiary hospitals (Gyeongsangnam-do Province) between April 2014 and June 2015. Penile length was measured via stretched penile length (SPL) and testicular size was measured using orchidometry (mL).

Result(s): SPL in children with cryptorchidism was significantly shorter compared to a cohort of healthy Korean boys aged 6-24 months (3.7±0.5 cm and 4.3±0.8 cm, p<0.001), although there were no differences with regard to height, body weight and contralateral testicular size between the two groups. According to the stratified ages (6-12, 12-18, and 18-24 months), SPL in children with cryptorchidism was persistently shorter at their ages than those without.

Conclusion(s): It might be that the penile length aged 6-24 months of children with unilateral cryptorchidism is shorter than that of a cohort of healthy Korean boys.

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Author NameID

Park, Sungchan; ORCID: <http://orcid.org/0000-0002-2337-983X>

Institution

(Ryu) Department of Urology, Samsung Changwon Hospital, Sungkyunkwan University School of Medicine, Changwon, South Korea (Cho) Department of Urology, Dong-A University Hospital, Dong-A University College of Medicine, Busan, South Korea

(Chung, Lee) Department of Urology, Pusan National University, Yangsan Hospital, Research Institute for Convergence of Biomedical Science and Technology, Yangsan, South Korea

(Kang) Department of Urology, Inje University Busan Paik Hospital, Inje University College of Medicine, Busan, South Korea

(Park) Department of Urology, Ulsan University Hospital, University of Ulsan College of Medicine, Ulsan, South Korea

Publisher

Korean Urological Association (E-mail: uro-edit@urology.or.kr)

Year of Publication

2018

238.

Parental subfertility and hypospadias and cryptorchidism in boys: results from two Danish birth cohorts.

Arendt L.H., Lindhard M.S., Kjersgaard C., Henriksen T.B., Olsen J., Ramlau-Hansen C.H.

Embase

Fertility and Sterility. 110 (5) (pp 826-832), 2018. Date of Publication: October 2018.

[Article]

AN: 2001168398

Objective: To study if parental subfertility is related to the occurrence of the male genital anomalies, cryptorchidism and hypospadias.

Design(s): Population-based cohort study.

Setting(s): Not applicable. Patient(s): A total of 80,220 singleton boys and their mothers from the Danish National Birth Cohort and the Aarhus Birth Cohort. Intervention(s): None. Main Outcome Measure(s): The two congenital anomalies; cryptorchidism and hypospadias, registered within the Danish National Patient Register up until December 31, 2012. Result(s): By means of Cox regression analyses, we found no associations between waiting time-to-pregnancy (TTP) and cryptorchidism or hypospadias among those who conceived spontaneously. The highest hazard ratio for cryptorchidism was seen among boys of couples with a TTP>12 months who conceived after fertility treatment (adjusted hazard ratio [aHR] 1.19, 95% confidence interval 0.92-1.55). For hypospadias, we found that boys of couples with a TTP>12 months who conceived after fertility treatment, had a 71% higher risk of hypospadias (aHR 1.71, [95% confidence interval 1.24-3.36]) as compared with boys of couples with a TTP<5 months. Conclusion(s): The findings from this study showed that boys of couples with TTP>12 months who conceived after fertility treatment, had a higher occurrence of hypospadias than boys conceived spontaneously of couples with a short TTP. Among those who conceived spontaneously, TTP was not associated with hypospadias or cryptorchidism. These findings indicate that fertility treatment or severity of subfertility is related to hypospadias.

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Institution

(Arendt, Kjersgaard, Ramlau-Hansen) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus University Hospital, Aarhus, Denmark (Arendt, Lindhard, Henriksen) Perinatal Epidemiology Research Unit, Department of Paediatric and Adolescent Medicine, Aarhus University Hospital, Aarhus, Denmark

(Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

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Year of Publication

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239.

Transscrotal orchidopexy for palpable cryptorchid testis: Follow-up and outcomes.

Papparella A., Cobellis G., Rosa L.D., Noviello C.

Embase

Pediatrica Medica e Chirurgica. 40 (2) (pp 34-37), 2018. Article Number: 191. Date of Publication: 2018.

[Article]

AN: 625819429

We retrospectively reviewed the results of transscrotal orchidopexy in the surgical management of palpable testis. From January 2014 to June 2017, 130 male children with a total of 140 palpable undescended testes (UDT) underwent transscrotal orchidopexy. The charts were retrospectively reviewed for demographic data, preoperative position and mobility of the testis, patency of the peritoneal vaginal duct (PVD), and post-operative complications. The resting position of the testis and its traction towards the scrotum were assessed before surgery and under anaesthesia. The mean age of the patients was 4.6 years. The position of the testis assessed at surgery was in most cases at the external inguinal ring (62.8%), at the neck of the scrotum (15.7%), in the inguinal canal (12.8%), or in an ectopic position (8,5%). A PVD was found in 66 testes (47.1%). Two surgical cases required an inguinal incision. In each patient, the postoperative course was unremarkable. The testicle at 1-year follow-up was in a scrotal position

in 134 cases, but 6 patients required a second surgical intervention for re-ascent of the testis. No testicular atrophy or inguinal hernias were observed. Transscrotal orchidopexy is a simple and effective procedure for the treatment of palpable UDT. The incidence of complications is low and manageable, with rapid postoperative recovery and early resumption of normal activities.

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PMC Identifier

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Status

Embase

Institution

(Papparella, Rosa) Department of Women and Children, General and Specialist Surgery, Campania University "Luigi Vanvitelli", Naples 80131, Italy (Cobellis, Noviello) Salesi Children's Hospital, Marche Polytechnic University, Ancona, Italy

Publisher

Page Press Publications

Year of Publication

2018

240.

Investigation of Azoospermia Factor (AZF) microdeletion of hypospadias patients in Indonesian population.

Juniarto A.Z., Yasari N.A.L., Santosa A., Faradz S.M.H.

Embase

Pakistan Journal of Medical and Health Sciences. 12 (3) (pp 1350-1353), 2018. Date of Publication: July-September 2018.

[Article]

AN: 625436554

Background: Hypospadias, a midline fusion defect of the male ventral urethra, is disorder of male external genital development occurring 0.7 - 4.5 per 10.000 live births. Hypospadias patients might have fertility problem and genetic factor could be involved on this aspect. Microdeletion of the Y chromosome, notably in Azoospermia factor region (AZF) have been observed in some patients with cryptorchidism and severe defects of spermatogenesis.

Aim(s): This study aimed to investigate microdeletions of AZF region in patients with hypospadias as a potential predictor factor for infertility. AZF amicrodeletion was associated to sertoli cell syndrome, while AZF bmicrodeletion lead to maturation arrest at the spermatocyte stage and AZF cmicrodeletion caused defect in sperm production.

Method(s): Total of 60 isolated hypospadias patients who admitted to CEBIOR were analyzed for AZF microdeletions during period of 2008 - 2016. DNA samples were analyzed by PCR-screening using several sequences-tagged-site (STS) markers from different region of the AZFa, AZFb, AZFc on Yq chromosome and SRY on Yp as internal control.

Result(s): Out of 60 analyzed cases (mean age 5.66 years), 3 (5%) patients showed microdeletion of AZF regions and only detected in AZFa region. No deletion was observed in AZFb and AZFc region. In addition, used as internal control, there no SRY gene microdeletion was found.

Conclusion(s): AZF microdeletions analysis canbe used as an infertility potential prognostic predictor in hypospadias patients and become important leading of genetic counseling related to possibility of infertility in the future.

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Status

Embase

Institution

(Juniarto, Faradz) Division of Human Genetics Center for Biomedical Research (CEBIOR), Faculty of Medicine, Diponegoro University, Sexual Adjustment Team, Dr. Kariadi Hospital, Semarang, Indonesia (Yasari) Faculty of Medicine, Diponegoro University, Indonesia (Santosa) Division of Urology, Sexual Adjustment Team, Dr. Kariadi Hospital, Semarang, Indonesia

Publisher

Lahore Medical And Dental College (Lahore Medical & Dental College, Lahore x, Pakistan. E-mail: nayyar_salam@yahoo.com)

Year of Publication

2018

Hypertensive disorders of pregnancy and genital anomalies in boys: A danish nationwide cohort study.

Arendt L.H., Henriksen T.B., Lindhard M.S., Parner E.T., Olsen J., Ramlau-Hansen C.H.

Embase

Epidemiology. 29 (5) (pp 739-748), 2018. Date of Publication: 2018.

[Article]

AN: 624836760

Background: Although congenital abnormalities in the male reproductive tract are common, their causes remain poorly understood. We studied associations between hypertensive disorders of pregnancy (pregestational hypertension, gestational hypertension, and preeclampsia) and the genital anomalies, cryptorchidism (undescended testes), and hypospadias (ventrally displaced urethral meatus).

Method(s): We established a population of 1,073,026 Danish boys born alive between 1 January 1978 and 31 December 2012. By means of Cox regression analyses, we estimated hazard ratios with 95% confidence intervals for cryptorchidism and hypospadias according to type and severity of hypertensive disorder. Further, we used restricted cubic spline analyses to investigate the association between gestational age at onset of severe and moderate preeclampsia and the two genital anomalies.

Result(s): We found associations between pregestational hypertension and cryptorchidism (HR: 1.3; 95% CI = 1.1, 1.6) and hypospadias (HR: 1.7; 95% CI = 1.3, 2.3), whereas gestational hypertension was only associated with cryptorchidism (HR: 1.2; 95% CI = 1.1, 1.4). Boys of mothers with preeclampsia had the highest occurrence of cryptorchidism and hypospadias, increasing with preeclampsia severity. Women with HELLP syndrome faced the highest risk of having a child with both cryptorchidism (HR: 2.1; 95% CI = 1.4, 3.2) and hypospadias (HR: 3.9; 95% CI = 2.5, 6.1). Further, the occurrence increased with early onset of preeclampsia diagnosis.

Conclusion(s): These findings support the hypotheses that preeclampsia and genital anomalies share common etiologic factors and that placental dysfunction and androgen deficiency in early pregnancy are important in the etiology of male genital anomalies.

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PMC Identifier

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Embase

Institution

(Arendt, Ramlau-Hansen) Department of Public Health, Section for Epidemiology, Aarhus University, Bartholins Alle 2, Aarhus 8000, Denmark (Arendt, Henriksen, Lindhard) Perinatal

Epidemiology Research Unit, Department of Paediatric and Adolescent Medicine, Aarhus University Hospital, Aarhus, Denmark

(Parner) Department of Public Health, Section for Biostatistics, Aarhus University, Aarhus, Denmark

(Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

Year of Publication

2018

242.

Anogenital distance and reproductive outcomes in 9- to 11-year-old boys: the INMA-Granada cohort study.

Freire C., Ocon-Hernandez O., Davila-Arias C., Perez-Lobato R., Calvente I., Ramos R., Olea N., Fernandez M.F.

Embase

Andrology. 6 (6) (pp 874-881), 2018. Date of Publication: November 2018.

[Article]

AN: 623776305

Background: Studies examining the association of anogenital distance (AGD), a biomarker of prenatal androgen exposure, with sexual development in children are lacking.

Objective(s): To assess the association between AGD measures and reproductive outcomes, including puberty onset, testicular volume, reproductive hormone levels, and urogenital malformations in boys aged 9-11 years.

Material(s) and Method(s): A cross-sectional study was conducted among children belonging to the Spanish Environment and Childhood (INMA) Project, a population-based birth cohort study. The present sample included 279 boys for whom data were available on AGD, pubertal stage, testicular volume, and relevant covariates. Out of the boys with AGD data, 187 provided a blood sample for hormone analysis. AGD was measured from the center of the anus to the base of the scrotum. Pubertal development was assessed according to Tanner stage of genital development (G1-G5), and testicular volume was measured with an orchidometer.

Result(s): After adjusting for potential confounders, logistic regression analysis showed that AGD was positively associated with testicular volume but not with Tanner stage (>G1 vs. G1), serum hormone levels, or undescended testis. Regardless of their age, body mass index, and Tanner stage (G1 or >G1), boys with longer AGD showed increased odds of a testicular volume >3 mL (OR = 1.06, 95%CI = 1.00-1.19 per 10% increment in AGD; and OR = 3.14, 95%CI = 0.99-9.94 for AGD >42 mm vs. <33 mm).

Discussion(s): Longer AGD was associated with testicular growth, an indicator of gonadarche, but not with other reproductive outcomes.

Conclusion(s): Although AGD was positively associated with testicular volume, it remains unclear whether AGD predicts testis size at puberty or is related to puberty onset.

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Author NameID

Freire C.; ORCID: <http://orcid.org/0000-0001-7370-1842>

Institution

(Freire, Ocon-Hernandez, Perez-Lobato, Calvente, Ramos, Olea, Fernandez) Health Research Institute of Granada (ibs.GRANADA), Granada, Spain (Freire, Olea, Fernandez) Spanish Consortium for Research on Epidemiology and Public Health (CIBERESP), Granada, Spain (Ocon-Hernandez) Obstetrics and Gynecology Service, San Cecilio University Hospital, Granada, Spain

(Davila-Arias) Radiology Unit, Health Science Technological Park University Hospital, Granada, Spain

(Perez-Lobato) Psychology Clinic ISEP, Granada, Spain

(Olea, Fernandez) Department of Radiology, School of Medicine, Center for Biomedical Research, University of Granada, Granada, Spain

Publisher

Blackwell Publishing Ltd

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2018

243.

Altered Integrins Expression of Patients Affected by Cryptorchidism.

Arena S., Favalaro A., La Face A., Romeo P., Scalfari G., Turiaco N., Antonuccio P., Rizzo G., Anastasi G., Romeo C.

Embase

Urologia Internationalis. 101 (2) (pp 219-223), 2018. Date of Publication: 01 Aug 2018.

[Article]

AN: 623657125

Objectives: The study aimed to investigate the expression of the integrin isoforms alpha7A and beta1A, expressed by myogenic precursor cells, and alpha7B and beta1D, expressed by mature muscle cells in the cremaster of patients affected by an undescended testis.

Method(s): Fifteen samples of cremaster were obtained from patients undergoing surgery for an undescended testis. Thirty control specimens of cremaster were harvested from patients with congenital hydrocele or inguinal hernia. Immunofluorescent analysis was carried out using anti-alpha7A, beta1A, alpha7B, and beta1D integrin antibodies. Sections were observed using confocal laser scanning microscopy.

Result(s): As compared with controls, a significant loss of alpha7B ($p = 0.0355$) and beta1D ($p = 0.0069$) integrins and a higher expression of alpha7A ($p = 0.0003$) and beta1A ($p = 0.0150$) was detected in the cremaster of patients affected by an undescended testis.

Conclusion(s): Our data document a critical alteration of the cytoskeleton of cremasteric smooth muscle cells in patients with an undescended testis. This might explain the altered function in smooth muscle cells in cremaster implied during testicular descent. We therefore speculate that the postnatal splicing of alpha7A to alpha7B and of beta1A to beta1D integrins is delayed. This could account for the common clinical scenario of spontaneous descent of the testes in the first months of life.

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PMC Identifier

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Embase

Institution

(Arena, La Face, Scalfari, Turiaco, Antonuccio, Romeo) Department of Human Pathology in Adult and Developmental Age Gaetano Barresi, Unit of Paediatric Surgery, University of Messina, Messina IT-98125, Italy (Favalaro, Rizzo, Anastasi) Department of Biomorphology and

Biotechnologies, University of Messina, Messina, Italy

(Romeo) Catholic University of Rome, Rome, Italy

Publisher
S. Karger AG
Year of Publication
2018

244.

Neonatal features of the Prader-Willi syndrome; the case for making the diagnosis during the first week of life.

Cizmecioglu F.M., Jones J.H., Paterson W.F., Kherra S., Kourime M., McGowan R., Shaikh M.G., Donaldson M.

Embase

JCRPE Journal of Clinical Research in Pediatric Endocrinology. 10 (3) (pp 264-273), 2018. Date of Publication: September 2018.

[Article]

AN: 623401131

Objective: Early diagnosis is of proven benefit in Prader-Willi syndrome (PWS). We therefore examined key perinatal features to aid early recognition.

Method(s): Data were collected from case records of subjects attending a multi-disciplinary clinic and from a retrospective birth questionnaire.

Result(s): Ninety patients (54 male-36 female) were seen between 1991-2015, most with paternal deletion (n=56) or maternal isodisomy (n=26). Features included cryptorchidism in 94% males, preterm birth (26%), birthweight <2500 g (24%), polyhydramnios (23%), breech presentation (23%) and need for nasogastric feeding (83%). Reduced fetal movements (FM) were reported in 82.5% patients compared with 4% healthy siblings. Of 35 children born since 1999, 23 were diagnosed clinically within 28 days while diagnosis in 12 was >28 days: 1-12 months in seven; and 3.75-10.5 years in five. Typical PWS features in these 12 infants included hypotonia (100%), feeding difficulties (75%), cryptorchidism (83% males) and reduced FM (66%). Causes other than PWS including neuromuscular disease were considered in nine patients.

Conclusion(s): Neonatal hypotonia, reduced FM, feeding difficulties and cryptorchidism should immediately suggest PWS, yet late diagnosis continues in some cases. Awareness of the typical features of PWS in newborn units is required to allow prompt detection even in the presence of confounding factors such as prematurity.

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Status

Embase

Author NameID

Cizmecioglu, Filiz Mine; ORCID: <http://orcid.org/0000-0001-7340-6368>

Institution

(Cizmecioglu) Kocaeli University, Faculty of Medicine, Department of Paediatric Endocrinology and Diabetes, Kocaeli, Turkey (Jones, Paterson, Shaikh) Royal Hospital for Children, Clinic of Endocrinology, Glasgow, United Kingdom

(Kherra) CHU Parnet, Clinic of Pediatrics, Algiers, Algeria

(Kourime) University Hospital Abderrahim Harouchi, Casablanca, Morocco

(McGowan) Southern Glasgow University Hospital, West of Scotland Genetic Services, Glasgow, United Kingdom

(Donaldson) University of Glasgow, Faculty of Medicine, Royal Hospital for Children, Clinic of Child Health, Glasgow, United Kingdom

Publisher

Galenos Yayıncılık, (Molla Gurani Cad. 21, Fındıkzade-Istanul 34093, Turkey)

Year of Publication

2018

245.

Timing of orchidopexy at a tertiary center in Saudi Arabia: Reasons for late surgery.

Alhazmi H., Junejo N.N., Albeaiti M., Alshammari A., Aljallad H., Almathami A., Vallasciani S.

Embase

Annals of Saudi Medicine. 38 (4) (pp 284-287), 2018. Date of Publication: July-August 2018.

[Article]

AN: 623355156

BACKGROUND: Orchidopexy should be performed during the first 18 months of life to decrease the risk of infertility and tumor formation. In our center, the timing of surgical correction varies depending on the availability of an operating room.

OBJECTIVE(S): Evaluate whether orchidopexy performed for patients referred to our center is done within the recommended time period and to determine causes for delay. DESIGN: Retrospective descriptive study. SETTING: Pediatric urology department of a tertiary care center. SUBJECTS AND METHODS: We retrospectively reviewed the charts of patients charts who underwent orchidopexy at our center from 2000 to 2010. We assessed referral time and waiting list time, which were subdivided as follows: from referral to first visit and from first visit to surgery. We included patients younger than 14 years and excluded patients with comorbidities that affected the timing of referral and surgical treatment. MAIN OUTCOME MEASURES: Referral time period and waiting list time for surgical correction of patients presented with undescended testis. SAMPLE SIZE: 128 RESULTS: After exclusion of 32 patients because of comorbidities, we describe 128 who underwent surgery for cryptorchidism at our center. The median (interquartile range, minimum-maximum) for age at surgery was 46.7 months (24.4-83.4, 3.1-248.6)]. The median (IQR) referral occurred at an age of 25.3 months (4.1-65.5). The median (IQR) waiting list time was 15.2 months (8.1-23.3). The median (IQR) waiting time from referral to the first visit was 4.1 months (1.0-8.2). The median waiting time from the first visit to surgery was 8.1 months (3.8-17.5).

CONCLUSION(S): The age at the time of surgery at our center was far from ideal because of late referrals. A structured program offered by our National Health Service to educate referring physicians is necessary. Community health initiatives must emphasize prompt referral to reduce the impact of delayed surgery. LIMITATIONS: Lack of data on the type of referring physician (i.e., general practitioner, pediatrician, surgeon, urologist).

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Embase

Author NameID

Junejo, Noor Nabi; ORCID: <http://orcid.org/0000-0002-8711-0792>

Institution

(Alhazmi) Pediatric Urology Division, Department of Surgery, King Saud University Medical City, King Saud University, Riyadh, Saudi Arabia (Junejo, Albeaiti, Alshammari, Aljallad, Almathami, Vallasciani) Department of Pediatric Urology, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia

(Junejo) Department of Urology, King Faisal Specialist Hospital and Research Centre, PO Box 3354, Riyadh 11211, Saudi Arabia

Publisher

King Faisal Specialist Hospital and Research Centre (E-mail: asm@kfshrc.edu.sa)

Year of Publication
2018

246.

Management of undescended testis may be improved with educational updates and new transferring model.

Yi W., Sheng-De W., Lian-Ju S., Tao L., Da-Wei H., Guang-Hui W.

Embase

Italian Journal of Pediatrics. 44 (1) (no pagination), 2018. Article Number: 58. Date of Publication: 24 May 2018.

[Article]

AN: 622263133

Background: To investigate whether management of undescended testis (UDT) may be improved with educational updates and new transferring model among referring providers (RPs).

Method(s): The age of orchidopexies performed in Children's Hospital of Chongqing Medical University were reviewed. We then proposed educational updates and new transferring model among RPs. The age of orchidopexies performed after our intervention were collected. Data were represented graphically and statistical analysis Chi-square for trend were used.

Result(s): A total of 1543 orchidopexies were performed. The median age of orchidopexy did not matched the target age of 6-12 months in any subsequent year. Survey of the RPs showed that 48.85% of their recommended age was below 12 months. However, only 25.50% of them would directly make a surgical referral to pediatric surgery specifically at this point. After we proposed educational updates, tracking the age of orchidopexy revealed a statistically significant trend downward.

Conclusion(s): The management of undescended testis may be improved with educational updates and new transferring model among primary healthcare practitioners.

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Institution

(Yi, Sheng-De, Lian-Ju, Tao, Da-Wei, Guang-Hui) Department of Urology, Children's Hospital of Chongqing Medical University, Kejiao Building (NO.6 Building), No.136, 2nd Zhongshan Road, Yuzhong District, Chongqing City 400014, China (Yi, Sheng-De, Lian-Ju, Guang-Hui) Chongqing Key Laboratory of Child Urogenital Development and Tissue Engineering, Chongqing, China

(Tao, Guang-Hui) Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing, China

(Da-Wei, Guang-Hui) China Intl. Science and Technology Cooperation Base of Child Development and Critical Disorders, Chongqing, China

(Yi, Lian-Ju, Guang-Hui) Chongqing Key Laboratory of Pediatrics Chongqing, Chongqing, China
Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2018

247.

Discordance in the Dependence on Kisspeptin Signaling in Mini Puberty vs Adolescent Puberty: Human Genetic Evidence.

Shahab M., Lippincott M., Chan Y.-M., Davies A., Merino P.M., Plummer L., Mericq V., Seminara S.

Embase

Journal of Clinical Endocrinology and Metabolism. 103 (4) (pp 1273-1276), 2018. Date of Publication: 01 Apr 2018.

[Article]

AN: 621707488

Context Hypothalamic kisspeptin signaling plays a critical role in the initiation and maintenance of reproductive function. Biallelic mutations in the coding sequence of KISS1R (GPR54) have been identified in patients with idiopathic hypogonadotropic hypogonadism, but it is unknown whether biallelic variants can also be associated with related reproductive disorders. Case Description A missense homozygous variant (c.890G>T p.R297L) in KISS1R was identified in a child who presented with micropallus and bilateral cryptorchidism. This variant has been reported to reduce, but not abolish, postreceptor signaling in vitro. Biochemical evaluation during the

neonatal period revealed low testosterone levels. By 11 years and 8 months, the boy began demonstrating increases in testicular volume. By 17 years and 3 months, his testicular volume was 20 mL; his penile length was 7.3 cm; and he had adult levels of circulating gonadotropins and testosterone. Conclusion This case report associates biallelic loss-of-function mutations in KISS1R with normal timing of adolescent puberty. Because these coding sequence variants occurred in a patient with microphallus and cryptorchidism, they demonstrate different levels of dependence of the hypothalamic-pituitary-gonadal cascade on kisspeptin signaling at distinct times in the reproductive life span. The suppression of the hypothalamic-pituitary-gonadal cascade during early life but not adolescence suggests that the mini puberty of infancy depends more on kisspeptin-induced, gonadotropin-releasing hormone-induced luteinizing hormone secretion than does adolescent puberty.

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Institution

(Shahab, Lippincott, Davies, Plummer, Seminara) Harvard Reproductive Sciences Center and Reproductive Endocrine Unit, Department of Medicine, Massachusetts General Hospital, Reproductive Endocrine Unit, 55 Fruit Street, Boston, MA 02114, United States (Chan) Division of Endocrinology, Department of Medicine, Boston Children's Hospital, Boston, MA, United States (Merino, Mericq) Institute of Maternal and Child Research, University of Chile, Santiago, Chile

Publisher

Oxford University Press (E-mail: mzendell@endo-society.org)

Year of Publication

2018

248.

Gonocyte transformation in a congenitally cryptorchid rat is normal and may be similar to the situation reported in human acquired cryptorchidism.

Loebenstein M., Hutson J., Li R.

Embase

Journal of Pediatric Surgery. 53 (9) (pp 1770-1775), 2018. Date of Publication: September 2018.
[Article]

AN: 620647148

Background: In congenital undescended testis (UDT) in humans, thermal insult damages early germ cell development during mini-puberty (3-6 months) causing increased risk of both cancer and infertility. In rodents however, UDT causes infertility but not cancer. In the TS rat with congenital UDT we hypothesized that early germ cell development would be normal as UDT only becomes manifest at 3-4 weeks (and the germ cells only become sensitive to thermal injury) after minipuberty is complete at 1 week.

Method(s): Normal testis and potential UDT from unilateral cryptorchid TS rats were collected at week 1 and 4 and processed into paraffin sections labeled for Sertoli cells (AMH), early germ cells (MVH) and spermatogonial stem cells (PLZF). Confocal microscopic images and Fiji Image J were used to count cells in testicular tubules with paired T-test statistical analysis.

Result(s): Total germ cells/tubule, basement membrane-bound germ cells/tubule, and Sertoli cells/tubule were unchanged between normally descending and future UDT at 1-4 weeks old ($P > 0.05$) Total germ cells/tubule and spermatogonial stem cells/tubule increased dramatically between weeks 1 and 4.

Conclusion(s): Rat gonocyte transformation is normal in both normally descending and future UDT. This suggests that congenitally cryptorchid rats may not develop testicular cancer because gonocytes (the putative origin of malignant degeneration) normally transform into spermatogonial stem cells before UDT occurs and the risk of thermal injury develops. This suggests the TS rat may be a good model for acquired UDT in human where the abnormal testicular position develops after gonocyte transformation is completed in the first year.

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Status

Embase

Institution

(Loebenstein, Hutson, Li) FD Stephens Surgical Research Group, Murdoch Children's Research Institute, Parkville, Victoria 3052, Australia (Hutson) Department of Urology, Royal Children's Hospital, Parkville, Victoria 3052, Australia

(Loebenstein, Hutson, Li) Department of Paediatrics, University of Melbourne, Parkville, Victoria 3052, Australia

Publisher

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Year of Publication

2018

249.

A critical review of recent clinical practice guidelines on management of cryptorchidism.

Kim J.K., Chua M.E., Ming J.M., Santos J.D., Zani-Ruttenstock E., Marson A., Bayley M., Koyle M.A.

Embase

Journal of Pediatric Surgery. 53 (10) (pp 2041-2047), 2018. Date of Publication: October 2018.

[Review]

AN: 619850435

Background/purpose: Limited efforts have been made in assessing the qualities of clinical practice guidelines (CPGs) on cryptorchidism (UDT). This appraisal aims to determine the quality of recent CPGs on the management of UDT.

Method(s): After systematic literature search, all English-based CPGs providing recommendations for the management of UDT from 2012 to 2017 were reviewed. Using the AGREE II (Appraisal of Guidelines and Research Evaluation) instrument, eligible CPGs were independently appraised by 5 reviewers. Domain scores were calculated and summarized. Intraclass coefficient (ICC) was used to assess for interrater reliability.

Result(s): Five CPGs from Agency for Healthcare Research and Quality (AHRQ), American Urological Association (AUA), British Association of Pediatric Surgeons/British Association of Urologic Surgeons (BAPS/BAUS), Canadian Urological Association (CUA), and European Association of Urology/European Society for Pediatric Urology (EAU/ESPU) were assessed. There was a solid agreement (ICC: 0.749) among the 5 reviewers ($p < 0.001$). Most recommendations for diagnostic and treatment approaches were consistent across CPGs. For most guidelines, the domains of 'clarity of presentation,' 'scope and purpose,' 'stakeholder involvement,' and 'rigor of development' were high, while 'applicability' was low.

Conclusion(s): Most guidelines on UDT score high in the AGREE II domains and have consistent recommendations. To improve the 'applicability' domain, future guidelines should improve on aspects that facilitate implementation of the recommendations.

Type of Study: Systematic review.

Level of Evidence: V (based on the lowest level of evidence utilized by the assessed guidelines).

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Status

Embase

Institution

(Kim) Faculty of Medicine, University of Toronto, Toronto, ON, Canada (Kim, Chua, Ming,

Santos, Koyle) Division of Urology, The Hospital for Sick Children, Toronto, ON, Canada

(Zani-Ruttenstock) Division of General and Thoracic Surgery, The Hospital for Sick Children,
Toronto, ON, Canada

(Marson) Hospital Library and Archives, The Hospital for Sick Children, Toronto, ON, Canada

(Bayley) Brain and Spinal Cord Rehabilitation Program, Toronto Rehabilitation Institute, Toronto,
ON, Canada

(Santos, Koyle) Department of Surgery, University of Toronto, Toronto, ON, Canada

Publisher

W.B. Saunders

Year of Publication

2018

250.

Postnatal Germ Cell Development in the Cryptorchid Testis: The Key to Explain Why Early
Surgery Decreases the Risk of Malignancy.

Thorup J., Clasen-Linde E., Li R., Reinhardt S., Kvist K., Vikraman J., Southwell B.R., Hutson
J.M., Cortes D.

Embase

European Journal of Pediatric Surgery. 28 (6) (pp 469-476), 2018. Date of Publication: 04 Jul
2018.

[Article]

AN: 618003820

Purpose Cryptorchidism is a risk factor for testicular malignancy and surgical treatment lowers
this risk. This study aimed to investigate the germ cell behavior in prepubertal cryptorchid testes
using immunohistochemical markers for germ cell malignancy to understand how early
orchiopexy may possibly prevent cancer developing. Materials and Methods Histology sections

from 1,521 consecutive testicular biopsies from 1,134 boys aged 1 month to 16.5 years operated for cryptorchidism were incubated with antibodies including antiplacental-like alkaline phosphatase (PLAP), anti-Oct3/4, anti-C-kit, and anti-D2-40. Results Oct3/4 and D2-40-positive germ cells are found throughout the first 2 years of life, with declining frequency thereafter. After 2 years, they should have disappeared and may indicate neoplasia. PLAP-positive cells were seen in 57 to 82% and C-kit-positive cells in 5 to 21% of cryptorchid testes between 4 and 13 years. Not until puberty did PLAP and C-kit-positive undifferentiated spermatogonial stem cells vanish. Only 0.3% of the present material had obvious prepubertal intratubular germ cell neoplasia (ITGCN) and they all had syndromic cryptorchidism. An additional three boys (0.3%) older than 2 years had weak Oct3/4 expression in undescended testes, but all cases were D2-40 negative. Conclusion Prepubertal ITGCN was rare and mostly seen in syndromic cryptorchidism. In nonsyndromic cryptorchidism PLAP-positive undifferentiated spermatogonial stem cells persisted in a significant proportion of nontreated undescended testes and they will be especially sensitive to long-lasting abnormally high temperature that may be the single most important cause facilitating the accumulation of mutations during cell replication and the development of ITGCN to be prevented by orchiopexy.

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Embase

Institution

(Thorup, Reinhardt, Kvist) Department of Pediatric Surgery, Rigshospitalet, 9 Blegdamsvej, Copenhagen DK-2100, Denmark (Thorup, Cortes) Institute for Clinical Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Clasen-Linde) Department of Pathology, Rigshospitalet, Copenhagen, Denmark

(Li, Vikraman, Southwell, Hutson) Douglas Stephens Surgical Research Laboratory, Murdoch Children's Research Institute, Melbourne, Australia

(Li, Southwell, Hutson) Department of Paediatrics, University of Melbourne, Melbourne, Australia

(Hutson) Department of Paediatric Urology, Royal Children's Hospital, Melbourne, Australia

(Cortes) Section of Endocrinology, Department of Pediatrics, Copenhagen University Hospital Hvidovre, Hvidovre, Denmark

Publisher

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251.

Guideline implementation for the treatment of undescended testes: Still room for improvement.

Ellerkamp V., Schmid A., Blumenstock G., Hrivataki G., Astfalk W., Loff S., Fuchs J.J., Zundel S.

Embase

Journal of Pediatric Surgery. 53 (11) (pp 2219-2224), 2018. Date of Publication: November 2018.

[Article]

AN: 2000853621

Background: Early orchidopexy (OP) around the age of 1 year is recommended in boys with congenital undescended testis (UDT) worldwide since decades. Former retrospective studies did not distinguish congenital from acquired UDT with a consecutive negative bias concerning the age at surgery.

Method(s): In a retrospective analysis, data of all boys who underwent OP in eight pediatric surgery institutions from 2009 to 2015 were analyzed. Congenital or acquired UDT were differentiated. Patients were categorized into 3 groups of age at surgery: (1) < 12 months, (2) 12-24 months, (3) > 24 months. Data of one institution were analyzed in detail: exact age of first referral, exact age at surgery, intraoperative findings.

Result(s): Out of 4448 boys, 3270 boys had congenital UDT. In 81% (2656 cases) surgery was performed beyond the age of 1 year, in 54.4% (1780) beyond the age of 2 years. chi-Square statistics showed a higher rate of early operations in hospitals compared to outpatient services and in Germany compared to Switzerland. In 694 congenital detailed cases, median age at referral was 13 months [range 0-196], median age at surgery was 15 months [range 0-202].

Conclusion(s): Delayed referral is the main reason for guideline non-conform delayed surgery in UDT.

Type of Study: Clinical Research paper.

Level of Evidence: Level III: Treatment Study.

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Institution

(Ellerkamp, Schmid, Fuchs) University Hospital Tuebingen, Department for Pediatric Surgery and Pediatric Urology, Germany (Blumenstock) Eberhard Karls University of Tuebingen, Department of Clinical Epidemiology and Applied Biostatistics, Germany

(Hrivatakis) Outpatient Clinic for Pediatric and Adolescent Surgery, Stuttgart, Germany

(Astfalk) Outpatient Clinic for Surgery, Reutlingen, Germany

(Loff) Olga hospital Stuttgart, Pediatric Surgery Clinic, Stuttgart, Germany

(Zundel) Kantonsspital Lucerne, Department of Pediatric Surgery, Lucerne, Switzerland

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252.

A Survey of the Current Practice Patterns of Contralateral Testis Fixation in Unilateral Testicular Conditions.

Abdelhalim A., Chamberlin J.D., McAleer I.M.

Embase

Urology. 116 (pp 156-160), 2018. Date of Publication: June 2018.

[Article]

AN: 2000643824

Objective: To query the current contralateral testis fixation (CTF) practice patterns among pediatric urologists in different clinical situations that could result in monorchism.

Method(s): An online survey was sent to members of the Urology Section of the American Academy of Pediatrics. The survey included questions addressing CTF practice patterns in 14 clinical scenarios. Responses were anonymously submitted, blindly reviewed, and analyzed.

Result(s): Among 53 respondents, 62.3% had academic appointments and 73.6% had an exclusive pediatric urology practice. All participants agreed on CTF necessity in testicular torsion beyond the neonatal period. CTF was advocated by 84.9% in prenatal torsion, 96.2% in postnatal torsion, and 94.3% in delayed torsion presentation. Emergent intervention was favored by 64.4% in prenatal and 98% in postnatal torsion. Only 1 participant (1.9%) preferred CTF with a unilateral testicular tumor and 5 (9.4%) in trauma substantiating an orchiectomy. There was less consensus on CTF in torsed undescended testis (79.3% in prepubertal and 81.13% in postpubertal),

testicular nubbin in a child (40.4%), palpable atrophic undescended testis (13.2%), and unilateral bell-clapper anomaly (47.2%). In situations other than torsion, lack of strong evidence was the commonest reason not to perform CTF.

Conclusion(s): The majority of responding pediatric urologists currently performs CTF in neonatal torsion. Although there is a general consensus on CTF in testicular torsion outside the neonatal period, CTF remains controversial in other clinical situations, warranting further research. The decision for CTF should involve patients, parents, and treating physicians.

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Status

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Author NameID

Abdelhalim, Ahmed; ORCID: <http://orcid.org/0000-0002-4451-0032>

Institution

(Abdelhalim, Chamberlin) The Department of Urology of the University of California, Irvine and the Children's Hospital of Orange County, Orange, CA, United States (Abdelhalim) The Department of Urology, Mansoura Urology and Nephrology Center, Mansoura University, Egypt (McAleer) The Department of Urology of the University of California, Irvine and Children's Hospital of Orange County, Orange, CA, United States

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253.

Erratum: A case-control study of maternal polybrominated diphenyl ether (PBDE) exposure and cryptorchidism in Canadian populations (Environ Health Perspect, (2017) 125(5), 057004, 10.1289/EHP522).

Anonymous

Embase

Environmental Health Perspectives. 126 (3) (no pagination), 2018. Article Number: 039001. Date of Publication: March 2018.

[Erratum]

AN: 621595028

In this article, one of the co-authors was omitted from the author list and affiliations. The correct list of authors and affiliations is as follows: Cynthia G. Goodyer,^{1,2} Shirley Poon,³ Katarina Aleksa^{3,4}, Laura Hou,⁵ Veronica Atehortua,¹ Amanda Carnevale,³ Gideon Koren,⁶ Roman Jednak,⁷ Sherif Emil,⁸ Darius Bagli,⁹ Sumit Dave,¹⁰ Barbara F. Hales,¹¹ and Jonathan Chevrier⁵ ¹Research Institute of McGill University Health Centre, McGill University Health Centre, Montreal, Quebec, Canada ²Department of Pediatrics, McGill University, Montreal, Quebec, Canada ³Department of Pharmacology and Toxicology, Hospital for Sick Children, University of Toronto, Toronto, Ontario, Canada ⁴Leslie Dan Faculty of Pharmacy, University of Toronto, Toronto, Ontario, Canada ⁵Department of Epidemiology, Biostatistics and Occupational Health, McGill University, Montreal, Quebec, Canada ⁶Toronto, Ontario, Canada ⁷Department of Pediatric Urology, McGill University, Montreal, Quebec, Canada ⁸Department of Pediatric General and Thoracic Surgery, McGill University, Montreal, Quebec, Canada ⁹Department of Pediatric Urology, Hospital for Sick Children, University of Toronto, Toronto, Ontario, Canada ¹⁰Division of Paediatric Urology, London Health Sciences Centre, London, Ontario, Canada ¹¹Department of Pharmacology and Therapeutics, McGill University, Montreal, Quebec, Canada
The authors regret the error.

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Erratum: Maternal endometriosis and genital malformations in boys: a Danish register-based study (Fertility and Sterility (2017) 108(4) (687-693)(S0015028217305046)(10.1016/j.fertnstert.2017.07.009)).

Anonymous

Embase

Fertility and Sterility. 109 (2) (pp 367), 2018. Date of Publication: February 2018.

[Erratum]

AN: 620187859

The article by Arendt et al., "Maternal endometriosis and genital malformations in boys: a Danish register-based study" (Fertil Steril 2017;108:687-693), contains the following errors in the confidence intervals reported from the subanalysis stratified by medically assisted reproduction (MAR). In the abstract under the heading "Results," the sixth sentence should read, "When stratified by medically assisted reproduction (MAR) technologies, the association was slightly stronger among boys born to women with endometriosis who had conceived via MAR, yet it remained moderate and statistically insignificant (aHR 1.27; 95% CI, 0.86; 1.88)." Under the heading "Results," the third sentence in the fifth paragraph should read, "We performed analyses stratified by MAR and found a slightly stronger association between endometriosis and cryptorchidism among women who conceived with MAR, yet the association remained statistically insignificant (aHR 1.27; 95% CI, 0.86; 1.88)." The authors apologize for the errors.

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255.

Clinical features of children with multicystic dysplastic kidney.

Kara A., Gurgoze M.K., Aydin M., Koc Z.P.

Embase

Pediatrics International. 60 (8) (pp 750-754), 2018. Date of Publication: August 2018.

[Article]

AN: 623724109

Background: To evaluate the clinical features of patients with multicystic dysplastic kidney (MCDK).

Method(s): The medical files of children diagnosed with MCDK between January 2008 and November 2015 were retrospectively reviewed. The demographic, clinical, laboratory and radiological data were evaluated.

Result(s): Of 128 children with MCDK enrolled in the study, 82 (64.1%) were male, and 46 (35.9%) were female ($P < 0.05$). MCDK were located on left and right sides in 66 (51.6%) and 62 children (48.4%), respectively ($P > 0.05$). Antenatal diagnosis was present in 64 patients (50%). The mean age at diagnosis was 2.8 ± 2.7 years (range, 0-8 years), and follow-up duration was 4.5 years. Fifteen patients (20.8%) had vesicoureteral reflux. Of these, four underwent endoscopic surgical correction. Other associated urological anomalies were ureteropelvic junction obstruction ($n = 6$), hypospadias ($n = 1$), and kidney stones ($n = 1$). On technetium-99 m dimercaptosuccinic acid scintigraphy, which was performed in all patients, no significant association between grade of reflux and presence of scarring was seen. Hypertension was diagnosed only in one child (0.8%) who required antihypertensive treatment. The prevalence of unilateral undescended testicle in children aged <1 year in the 82 male patients was 4.9%. Seventy-six patients (59.4%) developed compensatory hypertrophy in the contralateral kidney during a 1 year follow-up period. Of the total, only seven children (5.5%) had undergone nephrectomy.

Conclusion(s): MCDK follows a benign course with relatively few sequelae, and therefore these patients should be closely followed up and conservatively managed.

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Status

Embase

Author NameID

Kara, Aslihan; ORCID: <http://orcid.org/0000-0003-4410-0444>

Institution

(Kara, Gurgoze) Department of Pediatric Nephrology, Firat University School of Medicine, Elazig, Turkey (Aydin) Department of Neonatology, Firat University School of Medicine, Elazig, Turkey (Koc) Department of Nuclear Medicine, Firat University School of Medicine, Elazig, Turkey

Publisher

Blackwell Publishing

Year of Publication

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256.

Klinefelter syndrome in childhood: Variability in clinical and molecular findings.

Akcan N., Poyrazoglu S., Bas F., Bundak R., Darendeliler F.

Embase

JCRPE Journal of Clinical Research in Pediatric Endocrinology. 10 (2) (pp 100-107), 2018. Date of Publication: June 2018.

[Article]

AN: 622263853

Objective: Klinefelter syndrome (KS) is the most common (1/500-1/1000) chromosomal disorder in males, but only 10% of cases are identified in childhood. This study aimed to review the data of children with KS to assess the age and presenting symptoms for diagnosis, clinical and laboratory findings, together with the presence of comorbidities.

Method(s): Twenty-three KS patients were analyzed retrospectively. Age at admission, presenting symptoms, comorbid problems, height, weight, pubertal status, biochemical findings, hormone profiles, bone mineral density and karyotype were evaluated. Molecular analysis was also conducted in patients with ambiguous genitalia.

Result(s): The median age of patients at presentation was 3.0 (0.04-16.3) years. Most of the cases were diagnosed prenatally (n=15, 65.2%). Other reasons for admission were scrotal hypospadias (n=3, 14.3%), undescended testis (n=2, 9.5%), short stature (n=1, 4.8%), isolated micropenis (n=1, 4.8%) and a speech disorder (n=1, 4.8%). The most frequent clinical findings were neurocognitive disorders, speech impairment, social and behavioral problems and undescended testes. All except two patients were prepubertal at admission. Most of the patients (n=20, 86.9%) showed the classic 47,XXY karyotype. Steroid 5 alpha-reductase 2 gene and androgen receptor gene mutations were detected in two of the three cases with genital ambiguity.

Conclusion(s): Given the large number of underdiagnosed KS patients before adolescence, pediatricians need to be aware of the phenotypic variability of KS in childhood. Genetic analysis in KS patients may reveal mutations associated with other forms of disorders of sex development besides KS.

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Status

Embase

Author NameID

Akcan, Nese; ORCID: <http://orcid.org/0000-0003-2583-5736>

Institution

(Akcan) Near East University Faculty of Medicine, Department of Pediatric Endocrinology,
Nicosia, Cyprus (Poyrazoglu, Bas, Darendeliler) Istanbul University Istanbul Faculty of Medicine,
Department of Pediatric Endocrinology, Istanbul, Turkey

(Bundak) University of Kyrenia Faculty of Medicine, Department of Pediatric Endocrinology,
Kyrenia, Cyprus

Publisher

Galenos Yayincilik, (Molla Gurani Cad. 21, Findikzade-Istanul 34093, Turkey)

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2018

257.

Age at presentation of undescended testicles: A single-center study in Saudi Arabia.

Basalelah J.H., Alzahrani A.M., Alshaibani A.M., Alalyani N.S., Alsubiani T.A., AlMadi M.K.,
Allsowayan O.S.

Embase

Annals of Saudi Medicine. 38 (2) (pp 137-139), 2018. Date of Publication: March-April 2018.

[Article]

AN: 621661023

BACKGROUND: The undescended testis (UDT) is the most common genital anomaly encountered in pediatrics with an estimated incidence of 1% to 4% in full-term and 1% to 45% in preterm newborn boys. Over the years, studies on progressive histological deterioration and cancer risk has led to a change in recommendations for when orchidopexy should be done.

OBJECTIVE(S): Determine age at presentation of patients for UDT to a specialist in Saudi Arabia, age of operation and whether the recommended targeted time frame has been met.

DESIGN: Descriptive retrospective medical record review. SETTINGS: University hospital setting in urban location. SUBJECTS AND METHODS: The records of patients presenting to our center with UDT between the years 1996-2015 were reviewed for data on the age at presentation and age of operation. MAIN OUTCOME MEASURES: Age at time of evaluation and at time of surgical intervention compared with the international standard. SAMPLE SIZE: 331 cases.

RESULT(S): Out of the cases included, 195 met the inclusion criteria. The median age of presentation was 13.7 (range: 0-123.2) months. The median age at time of orchiopexy was 25 (range: 7.5-130.2) months. The median waiting time for elective surgery was 4.8 months (<1 day to 49.4 months).

CONCLUSION(S): Despite the international recommendation of carrying out orchidopexy between the ages of 6-12 months, the targeted recommended time frame is not met in Saudi Arabia. This is mainly related to late referral age and the long waiting time for elective surgery.

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Institution

(Basalelah, Alzahrani, Alshaibani, Alalyani, Alsubiani, AlMadi, Allsowayan) Department of Urology, Imam Abdulrahman Bin Faisal University, Dammam 31311, Saudi Arabia

Publisher

King Faisal Specialist Hospital and Research Centre

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258.

Testicular atrophy following inguinal hernia repair in children.

Sonderman K.A., Wolf L.L., Armstrong L.B., Taylor K., Jiang W., Weil B.R., Koehlmoos T.P., Ricca R.L., Weldon C.B., Haider A.H., Rice-Townsend S.E.

Embase

Pediatric Surgery International. 34 (5) (pp 553-560), 2018. Date of Publication: 01 May 2018.

[Article]

AN: 621447741

Purpose: We sought to determine the incidence and timing of testicular atrophy following inguinal hernia repair in children.

Method(s): We used the TRICARE database, which tracks care delivered to active and retired members of the US Armed Forces and their dependents, including > 3 million children. We abstracted data on male children < 12 years who underwent inguinal hernia repair (2005-2014). We excluded patients with history of testicular atrophy, malignancy or prior related operation. Our primary outcome was the incidence of the diagnosis of testicular atrophy. Among children with atrophy, we calculated median time to diagnosis, stratified by age/undescended testis.

Result(s): 8897 children met inclusion criteria. Median age at hernia repair was 2 years (IQR 1-5). Median follow-up was 3.57 years (IQR 1.69-6.19). Overall incidence of testicular atrophy was 5.1/10,000 person-years, with the highest incidence in those with an undescended testis (13.9/10,000 person-years). All cases occurred in children ≤ 5 years, with 72% in children < 2 years. Median time to atrophy was 2.4 years (IQR 0.64-3), with 30% occurring within 1 year and 75% within 3 years.

Conclusion(s): Testicular atrophy is a rare complication following inguinal hernia repair, with children < 2 years and those with an undescended testis at highest risk. While 30% of cases were diagnosed within a year after repair, atrophy may be diagnosed substantially later.

Level of Evidence: Prognosis Study, Level II.

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Status

Embase

Author NameID

Sonderman, Kristin A.; ORCID: <http://orcid.org/0000-0002-2623-1793>

Institution

(Sonderman, Wolf, Jiang, Haider) Department of Surgery, Center for Surgery and Public Health, Brigham and Women's Hospital, Harvard Medical School, and Harvard T.H. Chan School of Public Health, 1620 Tremont Street, 4th Floor, Suite 4-020, Boston, MA 02120, United States

(Sonderman, Wolf, Haider) Division of Trauma, Burns, and Surgical Critical Care, Department of Surgery, Brigham and Women's Hospital, Boston, MA, United States

(Armstrong, Weil, Weldon, Rice-Townsend) Department of Surgery, Boston Children's Hospital, Boston, MA, United States

(Taylor) Harvard Medical School, Boston, MA, United States

(Koehlmoos, Ricca) Uniformed Services University of the Health Sciences, Bethesda, MD, United States

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259.

Comparison of superb micro-vascular imaging (SMI) and conventional Doppler imaging techniques for evaluating testicular blood flow.

Durmaz M.S., Sivri M.

Embase

Journal of Medical Ultrasonics. 45 (3) (pp 443-452), 2018. Date of Publication: 01 Jul 2018.

[Article]

AN: 619764459

Purpose: Superb micro-vascular imaging (SMI) is a new blood flow (BF) technique recently developed to outface the limitations of conventional Doppler imaging techniques (CDIT). SMI can observe micro-vascular BF and low-velocity BF. SMI is available in two modes as color SMI (cSMI) and monochrome SMI (mSMI). To evaluate testicular BF, we have compared color Doppler (CD), power Doppler (PD), cSMI, and mSMI techniques.

Patients and Methods: A total of 156 patients (310 testes) were included in the study. We evaluated BF in the testes via CD, PD, cSMI, and mSMI techniques in a heterogenous patient group. Doppler examination was performed by observing the whole testis parenchyma within the examination area at the testicular hilus level at all examinations. Spot and linear flow color encoding determined in testis parenchyma were separately counted for every examination.

Result(s): SMI was found to be superior in all age groups and testis volumes for showing the BF. When we sequenced the examinations to show the BF in testis according to their priorities, it was found that mSMI > cSMI > PD > CD. As the testis volume decreases, a significant increase is observed in mSMI when compared to other examinations in showing vascularity in pediatric age groups, in cases diagnosed with undescended testis, and in cases that underwent surgery for undescended testis.

Conclusion(s): SMI renders more detailed vascular information on BF in the testes than CDIT. In particular, as the testis volume decreases, the priority of SMI showing BF increases. SMI should be a part of vascular examination in pediatric patients with small testis volume.

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Institution

(Durmaz, Sivri) Department of Radiology, Konya Health Sciences University Teaching and Research Hospital, Necip Fazil Mahallesi, Fatih Cad. No:4/1, Meram, Konya 42090, Turkey

Publisher

Springer Tokyo (E-mail: orders@springer.jp)

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2018

260.

Inguinal hernia repair by Bianchi incision in boys: a retrospective study.

Lin J., Li D., Chen J., Lin L., Xu Y.

Embase

Pediatric Surgery International. 34 (3) (pp 289-295), 2018. Date of Publication: 01 Mar 2018.

[Article]

AN: 619484530

Objective: Open inguinal hernia repair by Bianchi incision is a potential alternative technique for the treatment of IH. This study aims to retrospectively analyze boys with IH, who underwent open IH repair by Bianchi incision.

Method(s): A total of 3300 boys (1-144 months) with IH from April 2007 to September 2015 were enrolled into this study. An open high scrotal incision (Bianchi incision) to ligate the processus proximal to the internal inguinal ring was performed in patients for IH repair. Then, all patients were followed up after 7 days, 1 month, and 1 year. Operation time, hernia recurrence, hydrocele, testicular atrophy, cosmetic results, and the satisfaction of parents were evaluated.

Result(s): Among these 3300 boys, 1662 (50.36%) and 1349 (40.88%) boys with IH were operated on the right and left side, respectively, while 289 (8.76%) patients underwent bilateral surgery. The average operation time was 13.0 +/- 2.3 min for unilateral cases and 25.2 +/- 4.2 min for bilateral cases. Furthermore, among these 3300 boys, 309 boys (9.36%) were lost to

follow-up, and the remaining 2991 boys underwent a total of 3245 IH repairs. The complications included 20 recurrences who were repaired with the same technique, one wound rupture, and one acquired undescended testis. No infection, obvious scrotal hematoma, testicular atrophy, and vas deferens injury were found during the follow-up. In most instances, the scars were invisible, obtaining an excellent cosmetic effect.

Conclusion(s): Inguinal hernia repair by Bianchi incision is a safe, easy and effective technique with cosmetic benefits, which could be a reliable alternative for the treatment of pediatric inguinal hernia.

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Institution

(Lin, Li, Chen, Lin, Xu) Pediatric Surgery Department of the First Affiliated Hospital of Fujian Medical University, The First Clinical Medical College of Fujian Medical University, 20 Chazhong Road, Fuzhou, Fujian 350004, China

Publisher

Springer Verlag (E-mail: service@springer.de)

Year of Publication

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261.

Rare copy number variants identified in prune belly syndrome.

Boghossian N.S., Sicko R.J., Giannakou A., Dimopoulos A., Caggana M., Tsai M.Y., Yeung E.H., Pankratz N., Cole B.R., Romitti P.A., Browne M.L., Fan R., Liu A., Kay D.M., Mills J.L.

Embase

European Journal of Medical Genetics. 61 (3) (pp 145-151), 2018. Date of Publication: March 2018.

[Article]

AN: 619470902

Prune belly syndrome (PBS), also known as Eagle-Barrett syndrome, is a rare congenital disorder characterized by absence or hypoplasia of the abdominal wall musculature, urinary tract anomalies, and cryptorchidism in males. The etiology of PBS is largely unresolved, but genetic factors are implicated given its recurrence in families. We examined cases of PBS to identify novel pathogenic copy number variants (CNVs). A total of 34 cases (30 males and 4 females) with PBS identified from all live births in New York State (1998-2005) were genotyped using Illumina HumanOmni2.5 microarrays. CNVs were prioritized if they were absent from in-house controls, encompassed ≥ 10 consecutive probes, were ≥ 20 Kb in size, had $\leq 20\%$ overlap with common variants in population reference controls, and had $\leq 20\%$ overlap with any variant previously detected in other birth defect phenotypes screened in our laboratory. We identified 17 candidate autosomal CNVs; 10 cases each had one CNV and four cases each had two CNVs. The CNVs included a 158 Kb duplication at 4q22 that overlaps the BMPR1B gene; duplications of different sizes carried by two cases in the intron of STIM1 gene; a 67 Kb duplication 202 Kb downstream of the NOG gene, and a 1.34 Mb deletion including the MYOCD gene. The identified rare CNVs spanned genes involved in mesodermal, muscle, and urinary tract development and differentiation, which might help in elucidating the genetic contribution to PBS. We did not have parental DNA and cannot identify whether these CNVs were de novo or inherited. Further research on these CNVs, particularly BMP signaling is warranted to elucidate the pathogenesis of PBS.

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Embase

Institution

(Boghossian) Department of Epidemiology and Biostatistics, Arnold School of Public Health, University of South Carolina, Columbia, SC, United States (Boghossian, Giannakou,

Dimopoulos, Yeung, Liu, Mills) Division of Intramural Population Health Research, Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health, Bethesda, MD, United States

(Sicko, Caggana, Kay) Division of Genetics, Wadsworth Center, Department of Health, Albany, NY, United States

(Tsai, Pankratz, Cole) Department of Laboratory Medicine and Pathology, University of Minnesota Medical School, Minneapolis, MN, United States

(Romitti) Department of Epidemiology, College of Public Health, The University of Iowa, Iowa City, IA, United States

(Browne) New York State Department of Health, Congenital Malformations Registry, Albany, NY, United States

(Browne) University at Albany School of Public Health, Rensselaer, NY, United States

(Fan) Department of Biostatistics, Bioinformatics, and Biomathematics, Georgetown University Medical Center (GUMC), Washington, DC, United States

Publisher

Elsevier Masson SAS (62 rue Camille Desmoulins, Issy les Moulineaux Cedex 92442, France)

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262.

Sir Denis Browne, the father of modern pediatric surgery.

Nakayama D.K.

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Journal of Pediatric Surgery. 53 (3) (pp 576-579), 2018. Date of Publication: March 2018.

[Review]

AN: 619431444

Sir Denis John Wolko Browne (1892-1967), while not the first in the British Isles to devote his entire surgical practice to pediatric surgery, is accepted as "the father of pediatric surgery in the United Kingdom." He made contributions to operations as varied as tonsillectomy, pyloromyotomy, and hypospadias repair, and provided fundamental insights into the proper treatment of club foot, congenital dislocation of the hip, and cryptorchidism. He introduced the transverse laparotomy incision, primary repair of congenital intestinal obstruction, and the end-to-back anastomosis for intestinal atresia, techniques so commonly used that it is difficult to think of pediatric surgical operations done any differently. In addition, he invented the elegant Denis Browne retractor that remains in use today, one of the few eponymic instruments known by its originator's first and last name. He was among the founders of the British Association of Pediatric Surgeons, one of the first professional organizations in the field, and served as its first president. His legendary status was enhanced by an acerbic temperament that often surfaced in an outspoken and uncompromising advocacy on the behalf of the proper care of children. A larger-than-life figure in pediatric surgery, Browne's legacy is so wide-ranging and enduring that his unofficial title has been broadened to "the father of modern pediatric surgery."

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PMC Identifier

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Status

Embase

Institution

(Nakayama) Department of Surgery, Florida International University, Sacred Heart Medical Group, Pensacola, FL 32504, United States

Publisher

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2018

263.

Natural History and Conservative Treatment Outcomes for Hydroceles: A Retrospective Review of One Center's Experience.

Acer-Demir T., Ekenci B.Y., Ozer D., Turanoglu M.A., Haberal K.C., Bilgin E.B., Hicsonmez A.

Embase

Urology. 112 (pp 155-160), 2018. Date of Publication: February 2018.

[Article]

AN: 619397446

Objective: To elucidate epidemiological data and hydrocele progression, we reviewed pediatric patients diagnosed with hydroceles in our institution retrospectively.

Material(s) and Method(s): We reviewed data from 355 pediatric patients with hydroceles.

Questionnaires regarding age at diagnosis, time of delivery, presence of hydroceles in the father and brothers, age at recovery, age at surgery, cause of hydrocele (if present), type of hydrocele, associated pathologies, treatments, and posthydrocelectomy complications were completed by reviewing patients' medical records and interviewing their families.

Result(s): Patients with congenital hydroceles were more frequently born prematurely (32.5%) than were patients with noncongenital hydroceles (15.9%; $P = .001$). Fathers of 10 patients (3.7%) and brothers of 21 patients (7.7%) also had hydroceles. Hydroceles were associated with inguinal hernias on the same side (12.2%), cryptorchidism (7.5%), varicoceles (6.0%), and testis torsion

(0.5%). Among patients aged >1 year (n = 185), 27 did not undergo operations and healed spontaneously at an average of 5.30 +/- 3.36 months. For children aged >1 year who did not undergo surgery, the rate of spontaneous recovery within 6 months was 77.8% and that within 1 year was 96.3%.

Conclusion(s): Until strong evidence of hydrocele-induced testicular damage in children arises, we recommend following up congenital hydroceles until at least 1 year and preferably 2 years of age. We recommend following up noncongenital hydroceles for at least 6 months and preferably 1 year if there is no associated pathology indicating the need for earlier surgery such as an inguinal hernia, cryptorchidism, tense hydrocele, testis torsion, or testis mass.

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Institution

(Acer-Demir, Hicsonmez) Department of Pediatric Surgery, Faculty of Medicine, Baskent University, Ankara, Turkey (Ekenci, Ozer, Turanoglu, Haberal, Bilgin) Faculty of Medicine, Baskent University, Ankara, Turkey

Publisher

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Year of Publication

2018

264.

Expression of Xenobiotic Biomarkers CYP1 Family in Preputial Tissue of Patients with Hypospadias and Phimosis and Its Association with DNA Methylation Level of SRD5A2 Minimal Promoter.

Ohsako S., Aiba T., Miyado M., Fukami M., Ogata T., Hayashi Y., Mizuno K., Kojima Y.

Embase

Archives of Environmental Contamination and Toxicology. 74 (2) (pp 240-247), 2018. Date of Publication: 01 Feb 2018.

[Article]

AN: 619021410

Several epidemiological studies have suggested that the incidence of male reproductive organ malformations, including hypospadias or cryptorchidism, has increased due to fetal-stage exposure to environmental pollutants. However, the association of chemical exposure with the expression of target regulatory genes in the tissues of patients has not yet been reported. Because experimental approaches or clinical trials in human studies are limited, especially those using fetal and/or infants, it is difficult to obtain clear physiological evidence of mechanisms underlying male reproductive malformations. Thus, the lack of physiological evidence makes this issue controversial. We analyzed preputial tissues from patients with hypospadias (n = 23) and phimosis (n = 16). The atypical CYP1 family genes, CYP1A1 and CYP1B1, are potential biomarkers of environmental chemical exposure. We then compared the expression levels of CYP1A1 and CYP1B1 between hypospadias and phimosis samples by quantitative RT-PCR analysis. The mRNA expression levels of SRD5A2 and AR also were measured, because the androgen-related genes involved in the onset of disorders of male reproductive system. A significantly higher CYP1B1 expression level and a lower AR expression level were observed in the hypospadias groups than in the phimosis group. Positive correlations ($P < 0.001$) between the mRNA expression levels of the CYP1 family and SRD5A2 were found in patients with hypospadias but not in those with phimosis. Moreover, the methylation levels of the four genes were determined by bisulfite genomic sequencing. Although the SRD5A2 promoter region showed moderate methylation, no methylation was detected in CYP1A1, CYP1B1, or AR. There was no significant difference in SRD5A2 promoter methylation level between hypospadias and phimosis patients. Negative correlations were found between the methylation level of SRD5A2, especially at the - 221 Sp1 site, and the CYP1 family mRNA expression levels (CYP1A1, $p = 0.002$; CYP1B1, $p = 0.007$) in hypospadias patients, but not in phimosis patients. The significant positive association of mRNA expression level and the negative association of methylation level of the SRD5A2 gene with the mRNA expression levels of CYP1 family genes in the preputial tissue seem to indicate the chemical exposure of patients with hypospadias.

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Institution

(Ohsako, Aiba) Laboratory of Environmental Health Sciences, Center for Disease Biology and Integrative Medicine, Graduate School of Medicine, The University of Tokyo, Bunkyo-ku, Tokyo 113-0033, Japan (Miyado, Fukami) Department of Molecular Endocrinology and Metabolism,

National Research Institute for Child Health and Development, Setagaya-ku, Tokyo 157-8535, Japan

(Ogata) Department of Pediatrics, Hamamatsu University School of Medicine, Hamamatsu, Shizuoka 431-3192, Japan

(Hayashi, Mizuno) Department of Nephro-Urology, Nagoya City University Graduate School of Medical Sciences, Nagoya 467-8601, Japan

(Kojima) Department of Urology, Fukushima Medical University School of Medicine, Fukushima 960-1295, Japan

Publisher

Springer New York LLC (E-mail: barbara.b.bertram@gsk.com)

Year of Publication

2018

265.

Cryptorchidism and increased risk of neurodevelopmental disorders.

Chen J., Sorensen H.T., Miao M., Liang H., Ehrenstein V., Wang Z., Yuan W., Li J.

Embase

Journal of Psychiatric Research. 96 (pp 153-161), 2018. Date of Publication: January 2018.

[Article]

AN: 618813684

Male congenital malformations as cryptorchidism may contribute to the development of neurodevelopmental disorders directly or via shared familial genetic and/or environmental factors, but the evidence is sparse. Using population-based health registries, we conducted a cohort study of all liveborn singleton boys in Denmark during 1979-2008. Boys with a diagnosis of cryptorchidism were categorized into the exposed cohort and the other boys into the unexposed comparison cohort. The outcomes were diagnoses of any neurodevelopmental disorders and their subtypes. We used Cox proportional hazards regression to compute hazard ratios (HRs), taking into consideration several potential confounders. Among 884,083 male infants, 27,505 received a diagnosis of cryptorchidism during follow-up. Boys with cryptorchidism were more likely to be diagnosed with intellectual disability (HR = 1.77; 95% confidence interval [CI]:1.59,1.97), autism spectrum disorders (ASD) (HR = 1.24; 95% CI:1.13,1.35), attention-deficit hyperactivity disorder (ADHD) (HR = 1.17; 95% CI: 1.08,1.26), anxiety (HR = 1.09; 95% CI:

1.01,1.17), and other behavioral/emotional disorders (HR = 1.16; 95% CI: 1.08,1.26) compared to boys without cryptorchidism. The observed risks of intellectual disability, ASD, and ADHD were increased further in boys with bilateral cryptorchidism. Except for anxiety, cryptorchid boys had higher risks of neurodevelopmental disorders than their non-cryptorchid full brothers. The observed increased risks were similar among boys who underwent orchiopexy, as well as among those with shorter waiting times for this surgery. Cryptorchidism may be associated with increased risks of intellectual disability, ASD, ADHD, and other behavioral/emotional disorders. Cryptorchidism and neurodevelopmental disorders may have shared genetic or in-utero/early postnatal risk factors, which need to be further investigated.

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Institution

(Chen, Miao, Liang, Wang, Yuan) Key Lab. of Reproduction Regulation of NPFPC, SIPPR, IRD, Fudan University, Shanghai, China (Chen, Sorensen, Ehrenstein, Li) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

Publisher

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Year of Publication

2018

266.

Psychopathological features in Noonan syndrome.

Perrino F., Licchelli S., Serra G., Piccini G., Caciolo C., Pasqualetti P., Cirillo F., Leoni C., Digilio M.C., Zampino G., Tartaglia M., Alfieri P., Vicari S.

Embase

European Journal of Paediatric Neurology. 22 (1) (pp 170-177), 2018. Date of Publication:

January 2018.

[Article]

AN: 618776708

Introduction Noonan syndrome (NS) is an autosomal dominant disorder characterized by short stature, skeletal and haematological/lymphatic defects, distinctive facies, cryptorchidism, and a wide spectrum of congenital heart defects. Recurrent features also include variable cognitive deficits and behavioural problems. Recent research has been focused on the assessment of prevalence, age of onset and characterization of psychiatric features in this disorder. Herein, we evaluated the prevalence of attention deficit and hyperactivity disorder (ADHD), anxiety and depressive symptoms and syndromes in a cohort of individuals with clinical and molecular diagnosis of NS. Methods The Kiddie Schedule for Affective Disorders and Schizophrenia for School-Age Children Present and Lifetime version (K-SADS PL) has been used for the assessment of psychiatric disorders according to Diagnostic and Statistical Manual of Mental Disorders (DSM-IV). Multidimensional Anxiety Scale for Children (MASC) and the Children's Depression Inventory (CDI) have been assessed for the evaluation of anxiety and depressive symptoms and syndromes, whereas Conners Teacher and Parent Rating Scales-long version (CRS-R) have been used to evaluate ADHD. Results The study included 27 individuals (67% males) with an average age of 10.4 years (range 6-18 years) receiving molecular diagnosis of NS or a clinically related condition, evaluated and treated at the Neuropsychiatric Unit of Children's Hospital Bambino Gesù and at the Center for Rare Diseases of Fondazione Policlinico Universitario Agostino Gemelli, in Rome. Twenty individuals showed mutations in PTPN11, five in SOS1 and two in SHOC2. The mean IQ was 94 (Standard Deviation = 17, min = 56, max = 130). Seventy percent of the individuals (n = 19; 95% Confidence Interval = 52-85%) showed ADHD features, with six individuals reaching DSM-IV-TR criteria for ADHD disorder, and thirteen showing subsyndromal traits. Symptoms or syndrome of anxiety were present in 37% of the cohort (n = 10; 95% Confidence Interval = 19-56%), with two individuals showing anxiety disorder and eight cases exhibiting subsyndromal traits. Conclusion Our results show individuals with NS do present a very high risk to develop psychiatric disorders or symptoms during paediatric age. Based on these findings, preschool assessment of inattentive, hyperactivity/impulsivity and anxiety/depressive symptoms is recommended in order to plan a personalized treatment for psychological/psychiatric issues in affected individuals. Dedicated prospective studies are required to confirm the present data and better characterize the psychopathological profile in NS.

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Embase

Institution

(Perrino, Leoni, Zampino) Center for Rare Diseases, Department of Pediatrics, Polo Salute Donna e Bambino, Fondazione Policlinico Universitario A. Gemelli, Catholic University, Rome,

Italy (Licchelli, Serra, Piccini, Caciolo, Cirillo, Alfieri, Vicari) Department of Neuroscience, Unit of Child Neuropsychiatry, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy
(Pasqualetti) Service of Medical Statistics and Information Technology, Fatebenefratelli Foundation for Health Research and Education, Rome, Italy
(Pasqualetti) Language and Communication Across Modalities Laboratory (LaCAM), Institute of Cognitive Sciences and Technologies (ISTC-CNR), Italy
(Digilio, Tartaglia) Genetics and Rare Diseases Research Division, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy
(Serra) NESMOS Department, Sant'Andrea Hospital, Sapienza University of Rome, Italy
(Serra) Department of Psychiatry, Harvard Medical School, Boston, MA, United States
Publisher
W.B. Saunders Ltd
Year of Publication
2018

267.

Structural study of the cremaster muscle in patients with retractile testis.

Favorito L.A., Anderson K.M., Costa S.F., Costa W.S., Sampaio F.J.

Embase

Journal of Pediatric Surgery. 53 (4) (pp 780-783), 2018. Date of Publication: April 2018.

[Article]

AN: 616065925

Objective: To analyze the structure of the cremaster in patients with retractile testis (RT), comparing the distribution of nerves, elastic system and muscles with patients having cryptorchidism and inguinal hernia (IH).

Patients and Methods: We studied 31 patients, 17 with RT (mean age = 5.17years); 9 with IH (mean age = 2.6) and 5 with cryptorchidism (mean age = 3). A cremaster biopsy was performed and submitted to routine histological processing and studied using histochemistry and immunohistochemistry. The samples were photographed under an Olympus BX51 microscope. The images were processed with the Image J software and the cremaster muscle structures were quantified. Means were compared statistically using ANOVA and the unpaired t-test ($p < 0.05$).

Result(s): There were no differences ($p = 0.08$) in diameter of muscle fiber between the groups. The muscle fiber density differed between patients with RT and IH ($p = 0.02$): RT (mean = 17.71%, SD = 16.67), IH (mean = 38.06%, SD = 14) and cryptorchidism (mean = 21.47%, SD = 16.18). There was no difference ($p = 0.07$) in the density of elastic fibers in the three groups. We observed a lower concentration of cremaster nerves of patients with RT compared with IH ($p = 0.0362$): RT (mean = 1.72%, SD = 0.58), IH (mean = 3.28% SD = 0.94) and cryptorchidism (mean = 2.52%, SD = 0.53).

Conclusion(s): Retractable testis is not a normal variant, and presented a similar cremaster muscle structure as in patients with cryptorchidism.

Level of Evidence: II; prospective comparative study.

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Institution

(Favorito, Anderson, Costa, Costa, Sampaio) Urogenital Research Unit, State University of Rio de Janeiro, Brazil

Publisher

W.B. Saunders

Year of Publication

2018

268.

Fertility Issues in Pediatric Urology.

Kieran K., Shnorhavorian M.

Embase

Urologic Clinics of North America. 45 (4) (pp 587-599), 2018. Date of Publication: November 2018.

[Review]

AN: 2001082261

Improved understanding of the pathogenesis and natural history of many urologic disorders, as well as advances in fertility preservation techniques, has increased the awareness of and options for management of fertility threats in pediatric patients. In children, fertility may be altered by oncologic conditions, by differences in sexual differentiation, by gonadotoxic drugs and other side effects of treatment for nonurologic disorders, and by urologic conditions, such as varicocele and cryptorchidism. Although fertility concerns are best addressed in a multidisciplinary setting, pediatric urologists should be aware of the underlying pathophysiology and management options to properly counsel and advocate for patients.

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Institution

(Kieran, Shnorhavorian) Division of Urology, Seattle Children's Hospital, 4800 Sand Point Way Northeast, OA.9.220, Seattle, WA 98105, United States

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W.B. Saunders

Year of Publication

2018

269.

Spigelian hernia in children: low versus classical.

Sengar M., Mohta A., Neogi S., Gupta A., Viswanathan V.

Embase

Journal of Pediatric Surgery. 53 (11) (pp 2346-2348), 2018. Date of Publication: November 2018.

[Article]

AN: 2000945597

Purpose: Pediatric spigelian hernias are very rare. They are often missed or misdiagnosed. A series of cases with spigelian hernia, presented to a tertiary care center are presented here with emphasis on different anatomy of spigelian hernias with cryptorchidism and those without associated cryptorchidism.

Material(s) and Method(s): Over a period of seven years, nine cases of spigelian hernia presented to our tertiary care center. Male:female ratio was 3:1. There was a preponderance of right sided hernias. Three patients had associated cryptorchidism. One patient had associated lumbar hernia. All three patients with cryptorchidism had low spigelian hernia while others had classical spigelian hernia.

Conclusion(s): There is a likelihood of anatomical variation in SH associated with UDT and those without UDT. Understanding this anatomy may help in correct scrotal placement of testis.

Type of Study: Prospective Observational.

Level of Evidence: 4.

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Institution

(Sengar, Mohta, Viswanathan) Department of Pediatric Surgery, Chacha Nehru BalChikitsalaya, Geeta Colony, Delhi 110031, India (Neogi) Department of Pediatric Surgery, Maulana Azad Medical College, Delhi 110031, India

(Gupta) Department of Pediatric Surgery, Chacha Nehru Bal Chikitsalaya, Delhi 110031, India

Publisher

W.B. Saunders

Year of Publication

2018

270.

Overexpression of ubiquitin carboxyl-terminal hydrolase 1 (UCHL1) in boys with cryptorchidism. Toliczenko-Bernatowicz D., Matuszczak E., Tylicka M., Szymanska B., Komarowska M., Gorodkiewicz E., Debek W., Hermanowicz A.

Embase

PLoS ONE. 13 (2) (no pagination), 2018. Article Number: e0191806. Date of Publication:

February 2018.

[Article]

AN: 620507798

Background The ubiquitin-proteasome system regulate p53, caspase and Bcl-2 family proteins, and is crucial for the degradation of the defective germ cells in testes.

Purpose(s): to evaluate the concentration of ubiquitin carboxyl-terminal hydrolase 1 (UCHL1) in the blood plasma of boys with cryptorchidism and if there is any correlation with patient age.

Methods Patients-50 boys aged 1-4 years (median = 2,4y.) with unilateral cryptorchidism.

Exclusion criteria were: previous human chorionic gonadotropin treatment, an abnormal karyotype, endocrine or immunological disorders or any long-term medication. The control group- 50 healthy, age matched boys (aged 1-4 years, median = 2,1y.), admitted to the Pediatric Surgery Department for planned herniotomy. To investigate UCHL1 in blood plasma of boys with cryptorchidism, we used a novel technique Surface PLASMON RESONANCE Imaging (SPRI).

Results The median concentration of UCHL1 in the blood plasma of boys with cryptorchidism, was 5-folds higher than in boys with inguinal hernia, whose testicles were located in the scrotum.

We also noticed statistically significant difference between UCHL1 levels in boys with cryptorchidism up to 2 years old, and above 2 years old. Older boys, whose testicles since birth were located in the inguinal pouch or in the abdominal cavity, had higher concentration of UCHL1 in their blood plasma, than boys from younger group. In the group of cryptorchid boys, we also found slightly lower concentrations of INSL3, without statistical significance and no correlation with UCHL1 levels. **Conclusions** Uchl1 concentrations in the blood plasma of boys with cryptorchidism, may reflect the heat-induced apoptosis of germ cells. Higher UCHL1 concentrations in older boys with undescended testicles, probably express intensity of germ cell apoptosis, more extensive when testicles are subjected to heat-stress for longer period. Further analyses of UCHL1 may help to elucidate its role in mechanisms influencing spermatogenesis.

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Embase

Institution

(Toliczenko-Bernatowicz, Matuszczak, Komarowska, Debek, Hermanowicz) Paediatric Surgery Department, Medical University of Bialystok, Bialystok, Poland (Tylicka) Biophysics Department, Medical University of Bialystok, Bialystok, Poland

(Szymanska, Gorodkiewicz) Electrochemistry Department, University of Bialystok, Bialystok, Poland

Publisher

Public Library of Science (E-mail: plos@plos.org)

Year of Publication

2018

271.

Age at orchiopexy for undescended testis in the United States.

Williams K., Baumann L., Shah A., Abdullah F., Johnson E.K., Oyetunji T.A.

Embase

Journal of Pediatric Surgery. 53 (1) (pp 86-89), 2018. Date of Publication: January 2018.

[Article]

AN: 619202067

Purpose: Undescended testis (UDT) is the most common congenital anomaly of the male genitalia. The American Urological Association guidelines recommend orchiopexy by age 18 months to ameliorate the risk of subfertility. The study aim was to assess adherence to these guidelines on a national level.

Method(s): We retrospectively reviewed both the State Ambulatory Surgery Database (SASD) in 2012 and the Pediatric Health Information System (PHIS) for 2015. All patients aged 18 years or less with a diagnosis of UDT who underwent orchiopexy were included. Demographic data including age at repair as well as surgical subspecialty and payer status were extracted.

Result(s): Analysis of the 2012 SASD for New Jersey, Florida, and Maryland yielded 1654 patients. The majority were white, 791 (48.3%), with a median age at repair of 4 years (IQR 1-8). Most patients, 1048 (64%), had orchiopexy later than age 2. A total of 844 males were identified from the PHIS database. Of these, 63% were white. The median age at repair was 5 years (IQR 1-9). There were 577 (68%) patients older than 2 years at orchiopexy.

Conclusion(s): Almost 70% of boys with undescended testes in the United States are undergoing orchiopexy at least 6 months later than the recommended age.

Type of Study: Retrospective.

Level of Evidence: III.

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Embase

Institution

(Williams, Baumann, Abdullah, Johnson) Ann & Robert H. Lurie Children's Hospital of Chicago, 225 E. Chicago Ave, Box 63, Chicago, IL 60611, United States (Williams, Baumann, Abdullah, Johnson) Northwestern University Feinberg School of Medicine, 420 E. Superior St, Chicago, IL 60611, United States

(Shah) Howard University Hospital, 2041 Georgia Avenue NW, Washington, DC 20060, United States

(Oyetunji) Children's Mercy Kansas City, 2401 Gillham Rd., Kansas City, MO 64108, United States

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272.

The utility of chemical shift imaging and related fat suppression as standalone technique in cryptorchidism using low field MRI.

Allam M.F.A.B., Elian M.M.M., Rahman A.M.A., Allam F.A.F.A.B.

Embase

Egyptian Journal of Radiology and Nuclear Medicine. 49 (4) (pp 1140-1144), 2018. Date of Publication: December 2018.

[Article]

AN: 2001159809

Cryptorchidism is failure of one or both testes to descend completely into the scrotum. The testis can be located anywhere along its journey of descent, between the lower pole of the kidney and the inguinal canal.

Objective(s): The aim of this study was to assess the effectiveness of chemical shift imaging and Dixon based fat suppression using low field MRI in non-palpable undescended testicle.

Patients and Methods: From July 2017 through February 2018, Twenty eight boys, presented by either unilateral or bilateral cryptorchidism, with total number of forty testicles, underwent MRI study using low field machine in T1-weighted dual gradient-echo in-phase and opposed-phase sequence with Dixon based fat suppression.

Result(s): Based on the laparoscopic/operative data, twenty one testes were located at the inguinal region, whereas fifteen testes were pelvi-abdominal and four were absent. The whole image sets of CSI and Dixon fat suppression had the highest specificity and positive predictive value (100%) and the highest overall accuracy (95%) for detection of undescended testes.

Conclusion(s): chemical shift imaging combined with Dixon based fat suppression is reliable imaging tool as a standalone technique for evaluating cryptorchidism, providing high specificity and diagnostic accuracy.

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Institution

(Allam, Elian, Rahman) Department of Radiology, Faculty of Medicine, Minia University, Egypt

(Allam) Department of Anatomy, Faculty of Medicine, Minia University, Egypt

Publisher

Elsevier B.V.

Year of Publication

2018

273.

Utilization of scrotal orchidopexy for palpable undescended testes among surgeons.

Alyami F.A., Yahya A.F.B., Albraid H.F., Almarek N.A., Alkhalifa M.A., Alhazmi H., Trbay M.S., Neel K.F.

Embase

Urology Annals. 10 (4) (pp 380-385), 2018. Date of Publication: October-December 2018.

[Article]

AN: 624423671

Introduction: Scrotal orchidopexy for palpable undescended testicle (UDT) has received attention in the last decade due to its lower morbidity. This study was conducted to determine the frequency and factors related to the use of the scrotal approach in the surgical treatment of palpable UDT among surgeons.

Method(s): An observational cross-sectional study was carried out using an online survey, which was sent to different pediatric urologists, pediatric surgeons, and urologists groups. The survey

consisted of questions on demographics as well as surgeons opinions and experience toward scrotal orchidopexy.

Result(s): Of 163 respondents, 57 (35.0%) were pediatric surgeons, 98 (60.1%) were pediatric urologists, and 8 (4.9%) were urologists. There were 86 respondents (52.8%) who used the scrotal orchidopexy approach for UDT at any time in their practice. Pediatric urologists tended to use the scrotal orchidopexy approach for UDT more significantly than others ($P < 0.001$). There were significantly more scrotal orchidopexies for UDT performed by the pediatric urologists throughout their practice and per year compared to others, respectively ($P < 0.001$). Fifty-Two respondents (31.9%) claimed that scrotal orchidopexy is not a good option for their patients, while seven respondents (4.3%) claimed that the procedure was hard to perform.

Discussion(s): Based on the results of this study, we believe that there is a discrepancy in the reported advantages and success rate of scrotal orchidopexy in the published literature and the utilization of such an approach among surgeons managing palpable UDT in children.

Conclusion(s): Scrotal orchidopexy is an underutilized approach in the management of palpable UDT in children. Only 52.8% of our respondents used it for UDT. One of the main reasons why scrotal orchidopexy is underutilized is due to the surgeons' perception that scrotal orchidopexy is not the procedure of choice for their patients and their unfamiliarity with the procedure.

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Institution

(Alyami) Department of Surgery, Division of Urology, King Saud University Medical City, Riyadh, Saudi Arabia (Yahya, Albraidi, Almarek, Alkhalifa, Alhazmi, Trbay, Neel) Department of Surgery, College of Medicine, King Saud University, Riyadh, Saudi Arabia

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274.

Undescended Testes: Contemporary Factors Accounting for Late Presentation.

Ekwunife O.H., Ugwu J.O., Onwurah C., Okoli C.C., Epundu L.K.

Embase

African Journal of Urology. 24 (3) (pp 206-211), 2018. Date of Publication: September 2018.

[Article]

AN: 2001000388

Introduction: Early detection and treatment of undescended testes by orchiopexy by 6-12 months of age is important to minimize germ cell loss and improve the individuals' fertility index. Decades since after the adverse relationship between the delayed treatment and infertility was established, we are still having young boys presenting very late with undescended testes.

Objective(s): To understand the current reasons behind the increasing rate of late presentation of boys with undescended testes.

Patients and Methods: Clinical records of patients managed for undescended testis from January 2011 to December 2016 were reviewed. In addition, where the needed information was not recorded in the case files, telephone interviews and/or invitation for a clinical examination were employed when necessary to obtain complete data.

Result(s): There were 39 patients with 50 undescended testes (16 left, 12 right and 11 bilateral). The age range at presentation to the hospital was from 1 day to 11 years. Undescended testes were noticed in 27 (69%) children at the age one year or less. Of this number, 19 (70%) sought medical attention at any hospital within one year of age of the child, 11(41%) were presented to our unit for treatment within the child's first birthday, and 7 (26%) had surgical treatment within 2 years of age. Age at surgery ranges from 16 days to 11 years. The delay in presentation progressively declined as the parental academic level increases. Only 7 (18.0%) and 2 (5.1%) children were examined by a doctor after birth and at 6 weeks post natal visits, respectively. A majority of the empty scrotum was first noticed by the parents/grand mother in 25(64%) children as against 12 (31%) by health workers. Causes of late presentation include: parental ignorance of the abnormal position of the testes, 14 (35.9%); wrong information by clinicians at the hospital of first visit, 9 (23.1%) and erroneous belief that the testes will still descend, 8 (20.5%). At surgery, more testes in those aged 2 years and above were of low volume when compared with those aged less than 2 years.

Conclusion(s): Majority of children with undescended testes are still being presented late for surgical correction, with its attendant adverse consequences on fertility. Non examination of the newborn by clinicians, inadequate parental health education and delayed or neglected referral are the major responsible factors.

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Author NameID

Ekwunife O.H.; ORCID: <http://orcid.org/0000-0002-5500-6042>

Institution

(Ekwunife, Ugwu, Onwurah, Okoli, Epundu) Department of Surgery, Nnamdi Azikiwe University Teaching Hospital Nnewi, Nigeria

Publisher

Pan African Urological Surgeons Association(PAUSA) (E-mail: sunnydoodu@yahoo.com)

Year of Publication

2018

275.

PRDM histone methyltransferase mRNA levels increase in response to curative hormone treatment for cryptorchidism-dependent male infertility.

Hadziselimovic F., Cathomas G., Verkauskas G., Dasevicius D., Stadler M.B.

Embase

Genes. 9 (8) (no pagination), 2018. Article Number: 391. Date of Publication: 01 Aug 2018.

[Article]

AN: 623371440

There is a correlation between cryptorchidism and an increased risk of testicular cancer and infertility. During orchidopexy, testicular biopsies are performed to confirm the presence of type A dark (Ad) spermatogonia, which are a marker for low infertility risk (LIR). The Ad spermatogonia are absent in high infertility risk (HIR) patients, who are treated with a gonadotropin-releasing hormone agonist (GnRHa) to significantly lower the risk of infertility. Despite its prevalence, little is known about the molecular events involved in cryptorchidism. Previously, we compared the transcriptomes of LIR versus HIR patients treated with and without hormones. Here, we interpreted data regarding members of the positive regulatory domain-containing (PRDM) family; some of which encoded histone methyltransferases that are important for reproduction. We found there were lower levels of PRDM1, PRDM6, PRDM9, PRDM13, and PRDM14 mRNA in the testes of HIR patients compared with LIR patients, and that PRDM7, PRDM9, PRDM12, and PRDM16 were significantly induced after GnRHa treatment. Furthermore, we observed PRDM9 protein staining in the cytoplasm of germ cells in the testes from LIR and HIR patients, indicating that the mRNA and protein levels corresponded. This result indicated that the curative hormonal therapy for cryptorchidism involved conserved chromatin modification enzymes.

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Institution

(Hadziselimovic) Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, Bahnhofplatz 11, Liestal CH-4410, Switzerland (Cathomas) Department of Pathology Kantonspital Liestal, Liestal 4410, Switzerland

(Verkauskas) Children's Surgery Centre, Faculty of Medicine, University of Vilnius, Vilnius 01513, Lithuania

(Dasevicius) Institute for Pathology, National Centre of Pathology, Affiliate of Vilnius University Hospital Santariskiu Klinikos, Vilnius 08406, Lithuania

(Stadler) Swiss Institute of Bioinformatics, Basel 4058, Switzerland

(Stadler) Friedrich Miescher Institute for Biomedical Research, Basel 4058, Switzerland

Publisher

MDPI AG (Postfach, Basel CH-4005, Switzerland)

Year of Publication

2018

276.

Molecular Mechanisms of Syndromic Cryptorchidism: Data Synthesis of 50 Studies and Visualization of Gene-Disease Network.

Urh K., Kolenc Z., Hrovat M., Svet L., Dovc P., Kunej T.

Embase

Frontiers in Endocrinology. 9 (JUL) (no pagination), 2018. Article Number: 425. Date of Publication: 26 Jul 2018.

[Article]

AN: 623193352

Background: Cryptorchidism is one of the most frequent congenital birth defects in male children and is present in 2-4% of full-term male births. It has several possible health effects including reduced fertility, increased risk for testicular neoplasia, testicular torsion, and psychological consequences. Cryptorchidism is often diagnosed as comorbid; copresent with other diseases. It is also present in clinical picture of several syndromes. However, this field has not been

systematically studied. The aim of the present study was to catalog published cases of syndromes which include cryptorchidism in the clinical picture and associated genomic information.

Method(s): The literature was extracted from Public/Publisher MEDLINE and Web of Science databases, using the keywords including: syndrome, cryptorchidism, undescended testes, loci, and gene. The obtained data was organized in a table according to the previously proposed standardized data format. The results of the study were visually represented using Gephi and karyotype view.

Result(s): Fifty publications had sufficient data for analysis. Literature analysis resulted in 60 genomic loci, associated with 44 syndromes that have cryptorchidism in clinical picture. Genomic loci included 38 protein-coding genes and 22 structural variations containing microdeletions and microduplications. Loci, associated with syndromic cryptorchidism are located on 16 chromosomes. Visualization of retrieved data is presented in a gene-disease network.

Conclusion(s): The study is ongoing and further studies will be needed to develop a complete catalog with the data from upcoming publications. Additional studies will also be needed for revealing of molecular mechanisms associated with syndromic cryptorchidism and revealing complete disease network.

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Institution

(Urh, Kolenc, Hrovat, Svet, Dovc, Kunej) Department of Animal Science, Biotechnical Faculty, University of Ljubljana, Ljubljana, Slovenia

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2018

277.

High (trans) scrotal orchidopexy for palpable undescended testes in children: Influence of age and testicular position.

Talabi A.O., Sownde O.A., Adejuyigbe O.

Embase

African Journal of Urology. 24 (2) (pp 114-119), 2018. Date of Publication: June 2018.

[Article]

AN: 2000885950

Objectives: To assess the influence of increasing age of subjects and testicular position on the outcome of single incision orchidopexy. Subjects and methods: A prospective randomized study of children aged ≤ 15 years with palpable undescended testes. The study was conducted between July 2015 and December 2016 in a Nigerian tertiary hospital. Patients were randomized into two groups: single incision orchidopexy and conventional orchidopexy group by simple balloting. The parameters studied were the patients' bio-data, most caudal position of the testes, duration of surgery, wound complications such as wound infection, scrotal edema and haematoma. Others included testicular position at 6 months post-operatively, testicular hypotrophy and cosmetic appearance of scar. A p-value <0.05 was deemed significant.

Result(s): There were 52 patients with 59 testes. Their ages ranged from 1 year to 13 years with a mean of 6.5 ± 3.5 years. The age groups and testicular positions were well matched, $p > 0.05$. There was no conversion from high scrotal to conventional orchidopexy irrespective of the age and testicular position during surgery. The operative time was shorter in the high scrotal group compared to conventional group, $p < 0.05$. The rate of testicular reascent between both groups did not attain statistical significance, $p > 0.05$. Within the high scrotal group, increasing age and testicular location had no influence on the operative time, $p > 0.05$. There was no statistical significant difference in the rate of testicular retraction between the young and relatively older children among the high scrotal group, $p > 0.05$. All wounds healed without wound infection, scrotal edema and haematoma.

Conclusion(s): Increasing age of patients and location of testes had no effect on the outcome of high scrotal orchidopexy in terms of successful placement of testes in the scrotum and rate of testicular retraction when compared to the conventional orchidopexy. However, the operative time was shorter and the cosmetic appearance of scar was better in the high scrotal group compared to the conventional orchidopexy.

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Institution

(Talabi, Sownde, Adejuyigbe) Department of Surgery, Obafemi Awolowo University Teaching Hospitals Complex, Ile-Ife, Osun State, Nigeria

Publisher

Pan African Urological Surgeons Association(PAUSA) (E-mail: sunnydoodu@yahoo.com)

Year of Publication

278.

Abdominoscrotal hydrocele: A systematic review and proposed clinical grading.

Gadelkareem R.A.

Embase

African Journal of Urology. 24 (2) (pp 83-92), 2018. Date of Publication: June 2018.

[Review]

AN: 2000827318

Introduction: Abdominoscrotal hydrocele is a rare hydrocele variant in pediatrics and adults. Besides the historical concerns, controversies in etiology and management of abdominoscrotal hydrocele warrant studying. Subjects and methods: A systematic review was conducted based on a multilingual search of the world literature of abdominoscrotal hydrocele through electronic engines (Google Scholar and MEDLINE/PubMed). The demographic and clinical characteristics are critically addressed and a clinical grading system is proposed.

Result(s): From the 487 delivered articles, 320 articles were eligible to this review including only 21 case series. They delivered 579 abdominoscrotal hydrocele cases. Abdominoscrotal hydrocele affects pediatrics more than adults with significantly increased rate of reporting in the last decades. Full or incomplete case descriptions were found in 558 cases versus 21 cases with deficient description. Abdominoscrotal hydrocele has been reported from 45 countries and India has the highest rate. Eight proposed hypotheses were differentiated for etiology and grouped according to the direction of fluid formation and hydrocele growth. Associated congenital anomalies include contralateral hydroceles and cryptorchidism. Complications result from compression, hemorrhage, infection, torsion, and coincident malignancy. A clinical grading system considering the increased anatomical, pathological or clinical complexities has been proposed and provided two categories; simple and complex abdominoscrotal hydroceles with further sub-classes.

Conclusion(s): Abdominoscrotal hydrocele is rare, but the number of the reported cases is far larger than the previously reported numbers. Etiology follows multiple hypotheses and management is speculative. Proposed clinical grading may support differentiation of severity of the associated cumulative risks.

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Institution

(Gadelkareem) Assiut Urology and Nephrology Hospital, Faculty of Medicine, Assiut University,

Assiut, Egypt

Publisher

Pan African Urological Surgeons Association(PAUSA) (E-mail: sunnydoodu@yahoo.com)

Year of Publication

2018

279.

The possible impact of antenatal exposure to ubiquitous phthalates upon male reproductive function at 20 years of age.

Hart R.J., Frederiksen H., Doherty D.A., Keelan J.A., Skakkebaek N.E., Minaee N.S., McLachlan R., Newnham J.P., Dickinson J.E., Pennell C.E., Norman R.J., Main K.M.

Embase

Frontiers in Endocrinology. 9 (JUN) (no pagination), 2018. Article Number: 288. Date of Publication: 04 Jun 2018.

[Article]

AN: 622581985

Phthalates are ubiquitous environmental endocrine-disrupting chemicals suspected to interfere with developmental androgen action leading to adverse effects on male reproductive function. Prenatal exposure studies in rodents show cryptorchidism, hypospadias and reduced testicular volume (TV), testosterone and anogenital distance in males. It is postulated that there is a developmental window in utero when phthalate exposure has the most potent adverse effects. Some human studies show associations between prenatal phthalate exposure and reduced calculated "free" serum testosterone in infant boys and shorter anogenital distance. However, there are no data available yet which link antenatal exposure to long-term effects in men. We aimed to correlate antenatal phthalate exposure with adult TV, semen parameters and serum reproductive hormone concentrations. 913 men from the Western Australian (Raine) Pregnancy Cohort were contacted aged 20-22 years. 423 (56%) agreed to participate; 404 underwent testicular ultrasound examination; 365 provided semen samples, and reproductive hormones

were measured in 384. Maternal antenatal serum phthalate metabolite measurements were available for 185 and 111 men, who provided serum and semen, respectively. Maternal serum collected at 18 and 34 weeks gestation, stored at -80°C, was pooled and analyzed for 32 phthalate metabolites by liquid chromatography-tandem mass spectrometry. TV was calculated, semen analysis performed by WHO approved methods, and serum concentrations of gonadotrophins, inhibin B, and testosterone measured. Eleven phthalate metabolites were detected. Primary and secondary metabolites of di-(2-ethyl-hexyl) phthalate (DEHP) and di-isoo-nonyl phthalate (DiNP) were positively correlated. After correction for adult height, BMI, presence of a varicocele and exposure to maternal smoking mono-iso-nonyl phthalate (MiNP) ($r = -0.22$) and sums of DEHP and DiNP metabolites ($r = -0.24$) and the sum of the metabolites of the high molecular weight phthalates ($r = -0.21$) were negatively correlated with TV (all $p < 0.05$). After adjustment for BMI adult serum total testosterone was positively associated with exposure to the following antenatal serum phthalate metabolites: mono-(2-ethylhexyl) phthalate ($r = 0.26$), MiNP ($r = 0.18$), the sum of metabolites for DEHP ($r = 0.21$) and DiNP ($r = 0.18$), and the sum of high molecular weight phthalates ($r = 0.20$) ($p = 0.0005$ to $p = 0.02$). Given sample size, storage duration and confounding through postnatal exposures, further studies are required.

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Status

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Institution

(Hart, Doherty, Keelan, Newnham, Dickinson, Pennell) Division of Obstetrics and Gynaecology, University of Western Australia, Perth, WA, Australia (Hart) Fertility Specialists of Western Australia, Bethesda Hospital, Claremont, WA, Australia

(Frederiksen, Skakkebaek, Main) Department of Growth and Reproduction and EDMaRC, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark

(Doherty, Keelan, Minaee, Newnham, Pennell) Women and Infants Research Foundation, King Edward Memorial Hospital, Perth, WA, Australia

(McLachlan) Hudson Institute of Medical Research, Monash Medical Centre, Melbourne, VIC, Australia

(Norman) Robinson Research Institute, University of Adelaide, Adelaide, SA, Australia

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2018

280.

Selecting infants with cryptorchidism and high risk of infertility for optional adjuvant hormonal therapy and cryopreservation of germ cells: Experience from a pilot study.

Thorup J., Clasen-Linde E., Dong L., Hildorf S., Kristensen S.G., Andersen C.Y., Cortes D.

Embase

Frontiers in Endocrinology. 9 (JUN) (no pagination), 2018. Article Number: 299. Date of Publication: 05 Jun 2018.

[Article]

AN: 622581973

Introduction: Orchiopexy for congenital cryptorchid testes is recommended between 1/2 and 1 year of age to preserve testicular germ cell maturation. Early operation is not enough to preserve fertility in 22 and 36% of cases. Aim of this study was to set up a protocol for optional adjuvant hormonal therapy after orchiopexy and thereafter cryopreservation of testicular biopsies from infants with bilateral cryptorchidism and high infertility risk.

Material(s) and Method(s): We included 17 boys with bilateral cryptorchidism, normal FSH, and impaired germ cell number per tubular transverse section (G/T) in testicular biopsies at orchiopexy, 7 months to 31/2 years old. Postoperatively, optional adjuvant LHRH (kryptocur) 0.2 mg/0.1 mL 2x every second day in 16 weeks were offered. Ten boys were applicable for age matching according to parent's choice of treatment regime and G/T. Five of them had kryptocur, and five were controls. Repeat bilateral testicular biopsy evaluation and cryopreservation were offered to all boys 12 months after primary orchiopexy. For cryopreservation, tissue pieces were incubated with a cryoprotectant with a slow program freezing.

Result(s): Two out of five kryptocur-treated boys normalized both the average G/T and the number of adult dark spermatogonia (Ad-S). Another kryptocur-treated boy with initial low G/T and no Ad-S increased the G/T and achieved normal number of Ad-S at time of cryopreservation. In the control group, two patients reached only normal lower range regarding the G/T and the number of Ad-S. None of boys with less than average 0.2 G/T improved significantly, whether they were kryptocur-treated or not.

Conclusion(s): Based on literature and the present results, we recommend adjuvant LHRH treatment to boys with cryptorchidism and insufficient genuine gonadotropin stimulation at time of surgery, as these patients have high infertility risk. Cryopreservation should be an option in case of treatment failure of adjuvant LHRH. However, to avoid repeat surgery with biopsy, some

parents may choose biopsy for cryopreservation at time of the initial bilateral orchiopexy, well informed that the procedure may only be truly indicated in 22 and 36% of the cases.

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Status

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Institution

(Thorup, Hildorf) The Department of Pediatric Surgery, Copenhagen University Hospital, Rigshospitalet, Copenhagen, Denmark (Thorup, Andersen, Cortes) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Clasen-Linde) The Department of Pathology, Copenhagen University Hospital, Rigshospitalet, Copenhagen, Denmark

(Dong, Kristensen, Andersen) Laboratory of Reproductive Biology, Section 5712, Juliane Marie Centre for Women, Children and Reproduction, Rigshospitalet, Copenhagen, Denmark

(Cortes) Section of Endocrinology, Department of Pediatrics, Copenhagen University Hospital Hvidovre, Hvidovre, Denmark

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2018

281.

Lifestyle in pregnancy and cryptorchidism in sons: A study within two large danish birth cohorts. Kjersgaard C., Arendt L.H., Ernst A., Lindhard M.S., Olsen J., Henriksen T.B., Strandberg-Larsen K., Ramlau-Hansen C.H.

Embase

Clinical Epidemiology. 10 (pp 311-322), 2018. Date of Publication: 19 Mar 2018.

[Article]

AN: 622335495

Purpose: Cryptorchidism is the most frequent congenital malformation in boys and is associated with low sperm count, infertility and testicular cancer. Unhealthy maternal lifestyle during pregnancy such as smoking, high prepregnancy body mass index (BMI) as well as alcohol and caffeine intake may constitute possible risk factors for cryptorchidism, but results from the few

previous studies are conflicting. We aimed to explore the association between maternal lifestyle factors and occurrence of cryptorchidism in sons.

Patients and Methods: The Danish National Birth Cohort and the Aarhus Birth Cohort provided information on maternal lifestyle from early pregnancy. Data were linked to several Danish health registers, multiple imputation was used to handle missing data and Cox proportional hazards models were used to adjust for potential confounders.

Result(s): In total, 85,923 boys were included, and of them, 2.2% were diagnosed with cryptorchidism. We observed the strongest associations between maternal tobacco smoking and prepregnancy BMI and cryptorchidism. Sons of women who smoked 10-14 cigarettes/day had the highest hazard ratio (HR) for cryptorchidism (1.37; 95% CI: 1.06-1.76), and for maternal BMI ≥ 30 kg/m², the HR was 1.32 (95% CI: 1.06-1.65). Binge drinking was associated with an HR < 1 , if the women had one or two episodes in pregnancy (HR: 0.81; 95% CI: 0.67-0.98). Average maternal alcohol intake and caffeine intake during pregnancy were not significantly associated with a higher occurrence of cryptorchidism detected at birth or later in life.

Conclusion(s): Maternal tobacco smoking, overweight and obesity in pregnancy were associated with higher occurrence of cryptorchidism in boys in this study.

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Institution

(Kjersgaard, Arendt, Ernst, Olsen, Ramlau-Hansen) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus, Denmark (Arendt, Lindhard, Henriksen) Perinatal Epidemiology Research Unit, Department of Pediatrics, Aarhus University Hospital, Aarhus, Denmark

(Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

(Strandberg-Larsen) Department of Public Health, Section of Social Medicine, Copenhagen University, Copenhagen, Denmark

Publisher

Dove Medical Press Ltd (PO Box 300-008, Albany, 44 Corinthian Drive, Albany, Auckland 0752, New Zealand. E-mail: angela@dovepress.com)

Year of Publication

2018

282.

Disorders of sex development: Timing of diagnosis and management in a single large tertiary center.

Kohva E., Miettinen P.J., Taskinen S., Hero M., Tarkkanen A., Raivio T.

Embase

Endocrine Connections. 7 (4) (pp 595-603), 2018. Date of Publication: 01 Apr 2018.

[Article]

AN: 621967948

Background: We describe the phenotypic spectrum and timing of diagnosis and management in a large series of patients with disorders of sexual development (DSD) treated in a single pediatric tertiary center.

Method(s): DSD patients who had visited our tertiary center during the survey period (between 2004 and 2014) were identified based on an ICD-10 inquiry, and their phenotypic and molecular genetic findings were recorded from patient charts.

Result(s): Among the 550 DSD patients, 53.3% had 46,XY DSD; 37.1% had sex chromosome DSD and 9.6% had 46,XX DSD. The most common diagnoses were Turner syndrome (19.8%, diagnosed at the mean age of 4.7 +/- 5.5 years), Klinefelter syndrome (14.5%, 6.8 +/- 6.2 years) and bilateral cryptorchidism (23.1%). Very few patients with 46,XY DSD (7%) or 46,XX DSD (21%) had molecular genetic diagnosis. The yearly rate of DSD diagnoses remained stable over the survey period. After the release of the Nordic consensus on the management of undescended testes, the age at surgery for bilateral cryptorchidism declined significantly ($P < 0.001$).

Conclusion(s): Our results show that (i) Turner syndrome and Klinefelter syndrome, the most frequent single DSD diagnoses, are still diagnosed relatively late; (ii) a temporal shift was observed in the management of bilateral cryptorchidism, which may favorably influence patients' adulthood semen quality and (iii) next-generation sequencing methods are not fully employed in the diagnostics of DSD patients.

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Institution

(Kohva, Miettinen, Taskinen, Hero, Tarkkanen, Raivio) Children's Hospital, Pediatric Research Center, University of Helsinki and Helsinki University Hospital, Helsinki, Finland (Kohva, Tarkkanen, Raivio) Faculty of Medicine, Department of Physiology, University of Helsinki, Helsinki, Finland

(Taskinen) Department of Pediatric Surgery, Children's Hospital, University of Helsinki and Helsinki University Hospital, Helsinki, Finland

Publisher

BioScientifica Ltd. (Euro House, 22 Apex Court, Woodlands, Bradley Stoke, Bristol BS32 4JT, United Kingdom)

Year of Publication

2018

283.

Anti-Mullerian hormone and testicular function in prepubertal boys with cryptorchidism.

Grinson R.P., Gottlieb S., Bedecarras P., Rey R.A.

Embase

Frontiers in Endocrinology. 9 (APR) (no pagination), 2018. Article Number: 182. Date of Publication: 25 Apr 2018.

[Article]

AN: 621869354

Introduction: The functional capacity of the testes in prepubertal boys with cryptorchidism before treatment has received very little attention. The assessment of testicular function at diagnosis could be helpful in the understanding of the pathophysiology of cryptorchidism and in the evaluation of the effect of treatment. Anti-Mullerian hormone is a well-accepted Sertoli cell biomarker to evaluate testicular function during childhood without the need for stimulation tests.

Objective(s): The aim of the study was to assess testicular function in prepubertal children with cryptorchidism before orchiopexy, by determining serum anti-Mullerian hormone (AMH). We also evaluated serum gonadotropins and testosterone and looked for associations between testicular function and the clinical characteristics of cryptorchidism.

Material(s) and Method(s): We performed a retrospective, cross-sectional, analytical study at a tertiary pediatric public hospital. All clinical charts of patients admitted at the outpatient clinic, and recorded in our database with the diagnosis of cryptorchidism, were eligible. The main outcome measure of the study was the serum concentration of AMH. Secondary outcome measures were serum LH, FSH, and testosterone. For comparison, serum hormone levels from a normal population of 179 apparently normal prepubertal boys were used.

Result(s): Out of 1,557 patients eligible in our database, 186 with bilateral and 124 with unilateral cryptorchidism were selected using a randomization software. Median AMH standard deviation score was below 0 in both the bilaterally and the unilaterally cryptorchid groups, indicating that

testicular function was overall decreased in patients with cryptorchidism. Serum AMH was significantly lower in boys with bilateral cryptorchidism as compared with controls and unilaterally cryptorchid patients between 6 months and 1.9 years and between 2 and 8.9 years of age. Serum AMH below the normal range reflected testicular dysfunction in 9.5-36.5% of patients according to the age group in bilaterally cryptorchid boys and 6.3-16.7% in unilaterally cryptorchid boys. FSH was elevated in 8.1% and LH in 9.1% of boys with bilateral cryptorchidism, most of whom were anorchid. In patients with present testes, gonadotropins were only mildly elevated in less than 5% of the cases. Basal testosterone was mildly decreased in patients younger than 6 months old, and uninformative during childhood.

Conclusion(s): Prepubertal boys with cryptorchidism, especially those with bilaterally undescended gonads, have decreased AMH production. Although serum AMH may fall within the normal range, there is a considerable prevalence of testicular dysfunction during childhood in this frequent condition.

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Institution

(Grinspon, Gottlieb, Bedecarras, Rey) Centro de Investigaciones Endocrinologicas 'Dr. Cesar Bergada' (CEDIE), CONICET - FEI - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina (Rey) Departamento de Histologia, Biologia Celular, Embriologia y Genetica, Facultad de Medicina, Universidad de Buenos Aires, Buenos Aires, Argentina

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2018

284.

Cryptorchidism in boys with cerebral palsy is associated with the severity of disease and with co-occurrence of other congenital anomalies.

Barthold J.S., Wintner A., Hagerty J.A., Rogers K.J., Hossain M.J.

Embase

Frontiers in Endocrinology. 9 (APR) (no pagination), 2018. Article Number: 151. Date of Publication: 16 Apr 2018.

[Article]

AN: 621709823

Background: Cryptorchidism is reported in 40-50% of small case series of cerebral palsy (CP) and attributed to hypothalamic-pituitary-gonadal axis abnormalities, intellectual disability (ID), or cremaster spasticity. We collected demographic and clinical data to define the frequency of cryptorchidism and clinical comorbidities in a large CP population.

Method(s): Electronic health record data were collected for all male patients ≥ 7 years of age seen in a large, multidisciplinary CP clinic between 2000 and 2016. Variables including age, testicular position, surgical findings, CP severity, birth history, and comorbidities were tested for association using univariable and stepwise backward logistic regression analyses.

Result(s): Of 839 established patients, testis position was scrotal in 553, undescended in 185 (24%), retractile in 38 (5%), and undocumented in 63 cases. Cryptorchidism were diagnosed at a mean age of 5.8 years, with 20% documented as acquired, and testes were most commonly in the superficial inguinal pouch (41%) and associated with an inguinal hernia (56%). Severity was bilateral in 114/166 (69%) undescended and 24/36 (66%) retractile cases, respectively. Mean birth weight and the frequency of prematurity (55, 58, and 54%) and multiple birth (14, 13, and 9%) were not significantly different among the three groups. We observed a strong ordinal trend in the frequency of comorbidities, including quadriplegia, syndromic features/known genetic disease, intrauterine growth restriction (IUGR), death, brain malformations, seizures, gastrostomy, absent continence, ID and hearing, speech or visual impairment, with the retractile group holding the intermediate position for the majority. The stepwise multivariable analysis showed independent positive associations of cryptorchidism with quadriplegia, syndromic features/known genetic disease, hearing loss, and absent continence, and inverse associations with gestational age and multiple birth.

Conclusion(s): These data suggest that cryptorchidism is less common than previously reported in CP cases, but most strongly associated with quadriplegia. Delayed diagnosis may be related to an acquired condition or to the multiple additional functional deficits that occur in this population. Our data suggest that UDT and CP may both be components of malformation syndromes occurring in singleton births whose clinical features are more likely to include earlier delivery, IUGR, hearing loss, and/or global spasticity.

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Embase

Institution

(Barthold, Wintner, Hagerty, Rogers, Hossain) Nemours Biomedical Research/Division of Urology, Department of Orthopedics and Biostatistics Core, Nemours/Alfred I. duPont Hospital for Children, Wilmington, DE, United States

Publisher

Frontiers Media S.A. (E-mail: info@frontiersin.org)

Year of Publication

2018

285.

Laparoscopic management of Mullerian duct remnants in the paediatric age: Evidence and outcome analysis.

Raicevic M., Saxena A.K.

Embase

Journal of Minimal Access Surgery. 14 (2) (pp 95-98), 2018. Date of Publication: April-June 2018.

[Article]

AN: 621284846

Background: This study performed a literature analysis to determine outcomes of laparoscopic management in Mullerian duct remnants (MDRs).

Patients and Methods: Literature was searched for terms 'Mullerian' 'duct' 'remnants' and 'laparoscopy'. Primary end points were age at surgery, laparoscopic technique, intraoperative complications and postoperative morbidity.

Result(s): The search revealed 10 articles (2003-2014) and included 23 patients with mean age of 1.5 years (0.5-18) at surgery. All patients were 46XY, n = 1 normal male karyotype with two cell lines. Explorative laparoscopy was performed in n = 2 and surgical management in n = 21. The 5-port technique was used in n = 10, 3-port in n = 9 and robot-assisted laparoscopic approach in n = 1 (n = 1 technique not described). Complete MDRs removal in n = 9, complete dissection and MDRs neck ligation with endoscopic loops in n = 11 and n = 1 uterus and cervix were split in the midline. After MDRs removal, there were n = 2 bilateral orchidopexy, n = 3 unilateral orchidopexy, n = 1 Fowler-Stephens stage-I and n = 1 orchiectomy. Mean operative time was 193 min (120-334), and there were no intraoperative complications. Mean follow-up was 20.5 months (3-54) and morbidity included 1 prostatic diverticula. There were 13 associations with hypospadias, of which 3 had mixed gonads and 3 bilateral cryptorchidism. Other

associations were unilateral cryptorchidism and incarcerated inguinal hernia n = 1, right renal agenesis and left hydronephrosis n = 1 and n = 2 with transverse testicular ectopy.

Conclusion(s): This MDRs analysis suggests that the laparoscopic approach is an effective and safe method of treatment as no intraoperative complication has reported, and there is low morbidity in the long-term follow-up.

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Institution

(Raicevic) Department of Pediatric Surgery and Orthopedics, Clinical Center Nis, Serbia

(Saxena) Department of Pediatric Surgery, Chelsea Children's Hospital, Chelsea and Westminster NHS Foundation Trust, Imperial College London, 369 Fulham Road, London SW109NH, United Kingdom

Publisher

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Year of Publication

2018

286.

Single nucleotide polymorphisms associated with nonsyndromic cryptorchidism in Mexican patients.

Chavez-Saldana M., Viguera-Villasenor R.M., Yokoyama-Rebollar E., Landero-Huerta D.A., Rojas-Castaneda J.C., Taja-Chayeb L., Cuevas-Alpuche J.O., Zambrano E.

Embase

Andrologia. 50 (1) (no pagination), 2018. Article Number: e12788. Date of Publication: February 2018.

[Article]

AN: 620297030

Cryptorchidism is a frequent genitourinary malformation considered as an important risk factor for infertility and testicular malignancy. The aetiology of cryptorchidism is multifactorial in which

certain SNPs, capable of inhibiting the development of the gubernaculum, are implicated. We analysed 16 SNPs by allelic discrimination and automated sequencing in 85 patients and 99 healthy people, with the objective to identify the association between these variants and isolated cryptorchidism. In two different patients with unilateral cryptorchidism, we found the variants rs121912556 and p.R105R of INSL3 gene in a heterozygous form associated with cryptorchidism, so we could consider them as risk factors for cryptorchidism. On the other hand, SNPs rs10421916 of INSL3 gene, as well as the variants rs1555633 and rs7325513 in the RXFP2 gene, and rs3779456 variant of the HOXA10 gene were statistically significant, when the patients and controls were compared and could be considered as protective factors since are predominantly present in controls. The genotype-phenotype correlation did not show statistical significance. With these results, we could conclude that these polymorphisms can be considered as important variants in our population and would contribute in the future knowledge of the aetiology and physiopathology of cryptorchidism.

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Status

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Institution

(Chavez-Saldana, Viguera-Villasenor, Landero-Huerta, Rojas-Castaneda) Laboratorio de Biología de la Reproducción, Instituto Nacional de Pediatría, Ciudad de México, México

(Yokoyama-Rebollar) Departamento de Genética Humana, Instituto Nacional de Pediatría, Ciudad de México, México

(Taja-Chayeb) Laboratorio de Genética Molecular y Farmacogenética, Instituto Nacional de Cancerología, Ciudad de México, México

(Cuevas-Alpuche) Servicio de Urología, Instituto Nacional de Pediatría, Ciudad de México, México

(Zambrano) Departamento de Biología de la Reproducción, Instituto Nacional de Ciencias Médicas y Nutrición Salvador Zubirán, Ciudad de México, México

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287.

Genetic analysis of HOXA11 gene in Chinese patients with cryptorchidism.

Lu P., Wang Y., Wang F., Huang J., Zeng Y., He D., Huang H., Cheng Z.

Embase

Andrologia. 50 (1) (no pagination), 2018. Article Number: e12790. Date of Publication: Februaryy 2018.

[Article]

AN: 614698085

Cryptorchidism is the most common congenital anomaly in male children. Its aetiology remains unknown in the majority of cases. Because HOXA11 plays a vital role in regulating testicular descent, genetic variants in HOXA11 genes may contribute to the risk of cryptorchidism. In this study, mutation analysis was performed on the HOXA11 gene in a cohort of 89 patients with cryptorchidism. Furthermore, an association analysis of the HOXA11 tag single nucleotide polymorphism rs6461992 was performed in 168 patients with unilateral cryptorchidism and 193 controls. No pathogenic mutations were found. A significant difference in genotype and allele distribution was detected between cases and controls ($p = .029$ and $.022$ respectively). These results suggest that mutations in the coding sequence of HOXA11 might not be a common cause of cryptorchidism, while common polymorphisms in the HOXA11 gene might contribute to the risk of developing unilateral cryptorchidism.

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Institution

(Lu, He) Department of Urology, Children's Hospital of Chongqing Medical University, Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing, China (Wang)

Department of Reproduction and Infertility, Suizhou Central Hospital, Affiliated Hospital of Hubei University of Medicine, Suizhou, Hubei, China

(Wang, Huang, Zeng, Huang, Cheng) School of Basic Medical Sciences, Chongqing Medical University, Chongqing, China

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2018

288.

Testicular Cancer: Diagnosis and Treatment.

Baird D.C., Meyers G.J., Hu J.S.

Embase

American family physician. 97 (4) (pp 261-268), 2018. Date of Publication: 15 Feb 2018.

[Review]

AN: 627678924

Testicular cancer is the most common solid tumor among males 15 to 34 years of age, with an estimated 8,850 new cases and 410 deaths during 2017 in the United States. With effective treatment, the overall five-year survival rate is 97%. Risk factors for testicular cancer include undescended testis (cryptorchidism), personal or family history of testicular cancer, age, ethnicity, and infertility. The U.S. Preventive Services Task Force recommends against routine screening in asymptomatic men. Men with symptoms should receive a complete history and physical examination. Scrotal ultrasonography is the preferred initial imaging study. If a solid intratesticular mass is discovered, orchiectomy is both diagnostic and therapeutic. Staging through chest radiography, chemistry panel, liver function tests, and tumor markers guides treatment. Active surveillance, chemotherapy, retroperitoneal lymph node dissection, and radiation therapy are treatment options following orchiectomy. For patients desiring future fertility, sperm banking should be discussed early in the course of treatment. Family physicians often play a role in the care of cancer survivors and should be familiar with monitoring for recurrence and future complications, including secondary malignant neoplasms, cardiovascular risk, and infertility and subfertility.

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Institution

(Baird, Meyers) Carl R. Darnall Army Medical Center, Fort Hood, TX, United States (Hu) Fort PolkLAUnited States

Publisher

NLM (Medline)

Year of Publication

2018

289.

"Off-Label" Usage of Testicular Self-Examination (TSE): Benefits Beyond Cancer Detection.

Rovito M.J., Leone J.E., Cavayero C.T.

Embase

American journal of men's health. 12 (3) (pp 505-513), 2018. Date of Publication: 01 May 2018.

[Article]

AN: 626406903

Testicular cancer (TCa) is the most common cancer among 15- to 34-year-old males. Treatments are highly effective, which help foster approximately 98% 5-year survival rate. There are very few known causal factors of the disease (e.g., cryptorchidism and family history), thus possibly limiting primary prevention methods. Secondary preventative measures, on the other hand, most notably testicular self-examination (TSE), are well-known and are promoted to help prevent late-stage diagnosis of TCa. However, debate ensues as to whether or not TSE provides any benefit. In light of a recent systematic review conducted by these authors assessing the effectiveness of TSE promotion interventions, we propose that the behavior can serve as a tool not just for detection of TCa, but other male-specific urogenital health concerns, including varicoceles, hydroceles, among others. Furthermore, we suggest that TSE can also help foster informed decision-making skills among males with regard to health concerns and treatment options. However, our advocacy is in direct conflict with U.S. Preventive Services Task Force's influential "D" rating of TSE and others who recommend against performing TSE. This article offers an overview of the dispute over TSE's purpose and net benefit. We conclude that TSE is a behavior that is beneficial beyond detecting cancer. These proposed "off-label" uses of the procedure make for an effectual means to promote testicular health, self-awareness, and wellness among males. Recommendations for future research and advocacy are presented to the academy.

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Institution

(Rovito) University of Central Florida, Orlando, FL, United States (Leone) Bridgewater State University, Bridgewater, MA, United States

(Cavayero) Lake Erie College of Osteopathic Medicine, Bradenton, FL, United States

Publisher

NLM (Medline)
Year of Publication
2018

290.

Cryptorchidism in Sweden: A Nationwide Study of Prevalence, Operative Management, and Complications.

Bergbrant S., Omling E., Bjork J., Hagander L.

Embase

The Journal of pediatrics. 194 (pp 197-203), 2018. Date of Publication: 01 Mar 2018.

[Article]

AN: 626324339

OBJECTIVES: To review the cumulative prevalence, operative management, and complications of treatment for cryptorchidism in Sweden. STUDY DESIGN: A nationwide observational study from longitudinal register data of all Swedish-born boys 0-18 years of age, diagnosed with cryptorchidism from 2001 to 2014. Primary outcomes were occurrence and age at primary surgery. Secondary outcomes included type of procedure and surgical site infection.

RESULT(S): Of 20375 boys diagnosed with cryptorchidism in 2001-2014, 12766 were surgically treated. The cumulative childhood prevalence was 1.8% (95% CI, 1.5-2.0), with a higher prevalence in boys born prematurely, small for gestational age, or with low birth weight. The median age at treatment decreased from 6.2 years in 2001 to 3.4 years in 2014 ($P < .001$). Still, 94.1% (95% CI, 92.7-95.6) had surgery after the recommended 1 year of age in 2014. Variations in age at surgery between Swedish counties were great (range, 2.9-5.9 years of age). There were no deaths within 30 days after surgery and the frequency of surgical site infection was low (1.4%; 95% CI, 1.1-1.6).

CONCLUSION(S): The cumulative childhood prevalence of cryptorchidism was high, and complications were rare. Few boys underwent surgery in a timely manner according to clinical guidelines, and standards of care varied considerably across the country. Further research and collective actions are needed to improve the detection and management of congenital cryptorchidism.

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Institution

(Bergbrant, Omling, Hagander) Lund University, World Health Organization Collaborating Center for Surgery and Public Health, Department of Clinical Sciences Lund, Pediatric Surgery, Skane University Hospital, Lund, Sweden (Bjork) Lund University, Skane University Hospital, Department of Laboratory Medicine, Lund, Sweden; Clinical Studies Sweden, Forum South, Skane University Hospital, Lund, Sweden

Publisher

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Year of Publication

2018

291.

Impact of Preoperative Ultrasonographic Evaluation for Detection of a Viable Testis in Patients With a Unilateral Nonpalpable Testis.

Moriya K., Nakamura M., Nishimura Y., Nishida M., Kudo Y., Kanno Y., Kitta T., Kon M., Shinohara N.

Embase

Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine. 37 (7) (pp 1665-1670), 2018. Date of Publication: 01 Jul 2018.

[Article]

AN: 625908248

OBJECTIVES: To investigate the impact of preoperative ultrasonography (US) for detecting a viable testis in patients with a unilateral nonpalpable testis.

METHOD(S): Patients with a unilateral nonpalpable testis or unilateral palpable undescended testis who underwent preoperative US were enrolled. Patients were divided into 3 groups as follows: nonpalpable testis/no testis (n=27), which included patients who had a unilateral nonpalpable testis with no viable testis detected at surgery; nonpalpable testis/viable testis (n=10), which included patients who had a unilateral nonpalpable testis with a viable testis identified at surgery; and palpable undescended testis (n=63), which included patients who had a unilateral palpable undescended testis. Preoperative US findings were compared among each group.

RESULT(S): The testicular volume on the contralateral descended side in the nonpalpable testis/no testis group was significantly greater than that in the nonpalpable testis/viable testis and palpable undescended testis groups. When a testicular volume of 0.54mL was used as the cutoff value, the sensitivity, specificity, positive predictive value, and negative predictive value for the presence of the affected testis were 75.3%, 100%, 100%, and 60.0%, respectively. The testis on the affected side was detected in none of the nonpalpable testis/no testis group, 7 of the nonpalpable testis/viable testis group, and all of the palpable undescended testis group. When a visible testis on the affected side and a testicular volume of 0.54mL or less were defined as positive, all patients in the nonpalpable testis/viable testis and palpable undescended testis groups had positive findings versus none in the nonpalpable testis/no testis group.

CONCLUSION(S): Preoperative US provides valuable information for predicting the presence of a viable testis in patients with a unilateral nonpalpable testis by estimating both the unaffected testis and the affected side.

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Author NameID

Moriya, Kimihiko; ORCID: <http://orcid.org/0000-0001-6161-9403>

Institution

(Moriya, Nakamura, Nishimura, Kanno, Kitta, Kon, Shinohara) Department of Renal and Genitourinary Surgery, Hokkaido University Graduate School of Medicine, Sapporo, Japan
(Nishida, Kudo) Diagnostic Center for Sonography and Division of Laboratory and Transfusion Medicine, Hokkaido University Hospital, Sapporo, Japan

Year of Publication

2018

292.

Clinical and Cytogenetic Study of Egyptian Patients with Sex Chromosome Disorders of Sex Development.

Mazen IM; Mekkawy MK; Ibrahim HM; Kamel AK; Hamza RT; Elaidy AA.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Sexual Development. 2018 Jul 14.

[Journal Article]

UI: 30007989

Disorders of sex development (DSD) are conditions with an abnormal development of chromosomal, gonadal, or anatomical sex. Sex chromosome DSD involve conditions associated with either numerical or structural abnormalities of the sex chromosomes. This study included patients comprising a wide spectrum of presenting features suggestive of DSD and aimed at studying the frequency of sex chromosome abnormalities among 108 Egyptian DSD patients who presented to the Clinical Genetics and Endocrinology Clinics, National Research Centre (NRC) over the 2-year period of 2013 and 2014. The age of the studied patients ranged from 2 months to 39 years. The patients exhibited various presentations, including ambiguous genitalia, undescended testis, hypogonadism, short stature with Turner manifestations, primary or secondary amenorrhea, primary infertility, edema of the dorsum of the hands and feet, and dysmorphic features. The patients were subjected to detailed clinical examination, pubertal staging, and cytogenetic analysis. Our study reported a wide karyotypic diversity and a high frequency of sex chromosome DSD, reaching 44.44% (48/108). In conclusion, we showed a high incidence of sex chromosome DSD among Egyptian DSD patients with wide karyotype/phenotype diversity. The most frequent sex chromosome DSD detected among patients of the present study was Turner syndrome and variants (52.08%; 25/48) followed by Klinefelter syndrome and variants (43.75%; 21/48). Further long term studies are necessary for accurate detection of frequencies of different types of sex chromosomal anomalies and associated phenotypes.

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Mazen, Inas M; Mekkawy, Mona K; Ibrahim, Hanan M; Kamel, Alaa K; Hamza, Rasha T; Elaidy, Aya A.

Year of Publication

2018

293.

Evaluation of Fowler-Stephens orchiopexy for high-level intra-abdominal cryptorchidism: A systematic review and meta-analysis.

Yu C; Long C; Wei Y; Tang X; Liu B; Shen L; Dong X; Lin T; He D; Wu S; Wei G.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

International Journal Of Surgery. 60:74-87, 2018 Dec.

[Journal Article. Meta-Analysis. Systematic Review]

UI: 30408514

BACKGROUND AND OBJECTIVE: There has been no consensus regarding the best surgical strategy for patients with cryptorchidism involving high-level intra-abdominal testes. This systematic review and meta-analysis compared the outcomes of Fowler-Stephens orchiopexy (FSO) conducted as 1-stage or 2-stage, open or laparoscopic.

METHODS: The databases PubMed, Cochrane Library, Web of Science Database, Russian Science Citation Index, SciELO Citation Index, China National Knowledge Infrastructure, WanFang Data, and China Biology Medical disc were systematically searched for relevant articles.

RESULTS: Sixty studies involving 1991 operated testes were included in the final analysis. The overall success rates for 1- and 2-stage FSOs were 85% and 87%, respectively; overall atrophy rates for both were 10%. The success rates of 1-stage FSOs, open and laparoscopic, were 83% and 87%; with atrophy rates of 12% and 8%. The corresponding success rates of 2-stage FSOs were 81% and 89%; with atrophy rates of 17% and 8%. The odds ratios indicated that 2-stage FSO was significantly superior to 1-stage, and laparoscopic superior to open. Evaluating laparoscopic FSO over time, the success rates prior to year 2000, 2000 through 2010, and after 2010 were 85%, 89%, and 88%, and atrophy rates were 15%, 9%, and 6%, with no heterogeneity in the reports, and the funnel plot showed no publication bias.

CONCLUSION: Each surgical technique for correcting high-level intra-abdominal testes (IATs) had an acceptable success rate, from 81% to 89%. However, in terms of highest success rate and lowest atrophy rate, 2-stage laparoscopic FSO is the first choice for treating high intra-abdominal cryptorchidism.

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1

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Authors Full Name

Yu, Chengjun; Long, Chunlan; Wei, Yi; Tang, Xiangliang; Liu, Bin; Shen, Lianju; Dong, Xueyi; Lin, Tao; He, Dawei; Wu, Shengde; Wei, Guanghui.

Institution

Yu, Chengjun. Department of Urology, Children's Hospital of Chongqing Medical University, China; Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, China. Electronic address: 398429065@qq.com. Long, Chunlan. Ministry of Education Key Laboratory of Child Development and Disorders, China; China International Science and Technology Cooperation Base of Child Development and Critical Disorders, China; Chongqing Key Laboratory of Pediatrics Chongqing, China. Electronic address: 332005363@qq.com.

Wei, Yi. Department of Urology, Children's Hospital of Chongqing Medical University, China; Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, China. Electronic address: 351187861@qq.com.

Tang, Xiangliang. Department of Urology, Children's Hospital of Chongqing Medical University, China; Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, China. Electronic address: 313830881@qq.com.

Liu, Bin. Department of Urology, Children's Hospital of Chongqing Medical University, China; Ministry of Education Key Laboratory of Child Development and Disorders, China. Electronic address: 458018708@qq.com.

Shen, Lianju. Department of Urology, Children's Hospital of Chongqing Medical University, China; China International Science and Technology Cooperation Base of Child Development and Critical Disorders, China. Electronic address: 384851872@qq.com.

Dong, Xueyi. College of Life Science, Zhejiang University, China. Electronic address: dong.x@wehi.edu.au.

Lin, Tao. Department of Urology, Children's Hospital of Chongqing Medical University, China; Ministry of Education Key Laboratory of Child Development and Disorders, China. Electronic address: 2169575045@qq.com.

He, Dawei. Department of Urology, Children's Hospital of Chongqing Medical University, China; Ministry of Education Key Laboratory of Child Development and Disorders, China. Electronic address: 3192482030@qq.com.

Wu, Shengde. Department of Urology, Children's Hospital of Chongqing Medical University, China; Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, China; Ministry of Education Key Laboratory of Child Development and Disorders, China; Chongqing Key Laboratory of Pediatrics Chongqing, China. Electronic address: shengdewu@yeah.net.

Wei, Guanghui. Department of Urology, Children's Hospital of Chongqing Medical University, China; Chongqing Key Laboratory of Children Urogenital Development and Tissue Engineering, China; Ministry of Education Key Laboratory of Child Development and Disorders, China;

Chongqing Key Laboratory of Pediatrics Chongqing, China. Electronic address:
u806806@cqmu.com.

Year of Publication

2018

294.

miR-34c disrupts spermatogonial stem cell homeostasis in cryptorchid testes by targeting Nanos2.

Huang Z; Tang D; Gao J; Dou X; Cheng P; Peng D; Zhang Y; Mao J; Zhang L; Zhang X.

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Reproductive Biology & Endocrinology. 16(1):97, 2018 Oct 15.

[Journal Article]

UI: 30322389

BACKGROUND: Cryptorchidism as a common genitourinary malformation with the serious complication of male infertility draws widespread attention. With several reported miRNAs playing critical roles in spermatogonial stem cells (SSCs), we aimed to explore the fundamental function of the highly conserved miR-34c in cryptorchidism.

METHODS: To explore whether miR-34c participates in spermatogenesis by regulating Nanos2, we examined the effect of overexpression and inhibition for miR-34c on Nanos2 expression in GC-1 cells. Moreover, the expression levels of miR-34c and Nanos2 with cryptorchidism in humans and mice were examined. Furthermore, the homeostasis of SSCs was evaluated through counting the number of promyelocytic leukemia zinc finger (PLZF) positive spermatogonia in murine cryptorchid testes.

RESULTS: In the present study, we show that miR-34c could inhibit the expression of Nanos2 in GC-1 cells. Meanwhile, miR-34c significantly decreased in both the testicular tissues of patients with cryptorchidism and surgery-induced murine model of cryptorchidism. Western blot revealed that the protein level of Nanos2 was up-regulated and showed to be negatively correlated to the expression of miR-34c in our model. The abnormal expression of miR-34c/Nanos2 disrupted the balance between SSC self-renewal and differentiation, eventually damaging the spermatogenesis of cryptorchid testes.

CONCLUSIONS: The miR-34c/Nanos2 pathway provides new insight into the mechanism of male infertility caused by cryptorchidism. Our results indicate that miR-34c may serve as a biological marker for treatment of infertility caused by cryptorchidism.

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1

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Authors Full Name

Huang, Zhenyu; Tang, Dongdong; Gao, Jingjing; Dou, Xianming; Cheng, Peng; Peng, Dangwei; Zhang, Yao; Mao, Jun; Zhang, Li; Zhang, Xiansheng.

Institution

Huang, Zhenyu. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China. Tang, Dongdong. Reproductive Medicine Center, Department of Obstetrics and Gynecology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Tang, Dongdong. Anhui Province Key Laboratory of Reproductive Health and Genetics, Anhui Medical University, Hefei, 230032, China.

Tang, Dongdong. Anhui Provincial Engineering Technology Research Center for Biopreservation and Artificial Organs, Hefei, 230022, China.

Gao, Jingjing. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Dou, Xianming. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Cheng, Peng. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Peng, Dangwei. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Zhang, Yao. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Mao, Jun. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Zhang, Li. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China.

Zhang, Xiansheng. Department of Urology, The First Affiliated Hospital of Anhui Medical University, Hefei, 230022, China. xiansheng-zhang@163.com.

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2018

295.

Testicular cancer. [Review]

Cheng L; Albers P; Berney DM; Feldman DR; Daugaard G; Gilligan T; Looijenga LHJ.

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Nature Reviews. Disease Primers. 4(1):29, 2018 10 05.

[Journal Article. Research Support, Non-U.S. Gov't. Review]

UI: 30291251

Testicular cancer is the most common malignancy among men between 14 and 44 years of age, and its incidence has risen over the past two decades in Western countries. Both genetic and environmental factors contribute to the development of testicular cancer, for which cryptorchidism is the most common risk factor. Progress has been made in our understanding of the disease since the initial description of carcinoma in situ of the testis in 1972 (now referred to as germ cell neoplasia in situ), which has led to improved treatment options. The combination of surgery and cisplatin-based chemotherapy has resulted in a cure rate of >90% in patients with testicular cancer, although some patients become refractory to chemotherapy or have a late relapse; an improved understanding of the molecular determinants underlying tumour sensitivity and resistance may lead to the development of novel therapies for these patients. This Primer provides an overview of the biology, epidemiology, diagnosis and current treatment guidelines for testicular cancer, with a focus on germ cell tumours. We also outline areas for future research and what to expect in the next decade for testicular cancer.

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1

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Authors Full Name

Cheng, Liang; Albers, Peter; Berney, Daniel M; Feldman, Darren R; Daugaard, Gedske; Gilligan, Timothy; Looijenga, Leendert H J.

Institution

Cheng, Liang. Departments of Pathology and Laboratory Medicine and Urology, Indiana University School of Medicine, Indianapolis, IN, USA. lcheng@iupui.edu. Albers, Peter. Department of Urology, Heinrich-Heine University, Medical Faculty, Dusseldorf, Germany.

Berney, Daniel M. Department of Molecular Oncology, Barts Cancer Institute, St Bartholomew's Hospital, Queen Mary University of London, London, UK.

Feldman, Darren R. Department of Medicine, Memorial Sloan Kettering Cancer Center, New York, NY, USA.

Feldman, Darren R. Department of Medicine, Weill Cornell Medical College, New York, NY, USA.

Daugaard, Gedskke. Department of Oncology, Copenhagen University Hospital, Rigshospitalet, Copenhagen, Denmark.

Gilligan, Timothy. Department of Solid Tumor Oncology, Cleveland Clinic Taussig Cancer Institute, Cleveland, OH, USA.

Looijenga, Leendert H J. Department of Pathology, Josephine Nefkens Building, Erasmus MC-University Medical Center, Cancer Institute, Rotterdam, Netherlands.

Year of Publication
2018

296.

Clinical features, treatment, and outcomes of bilateral Wilms' tumor: A systematic review and meta-analysis.

Han Q; Li K; Dong K; Xiao X; Yao W; Liu G.

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Journal of Pediatric Surgery. 53(12):2465-2469, 2018 Dec.

[Journal Article. Meta-Analysis. Systematic Review]

UI: 30274708

BACKGROUND: Wilms' tumor(WT) is the most common malignant renal tumor of childhood. Despite the good prognosis of WT, bilateral Wilms' tumor (BWT) still has a poor outcome. We systematically reviewed the literature on BWT, aiming to define its clinical features, treatment, and outcomes.

METHODS: PubMed, OVID EMBase, Web of Science, and Cochrane Library were systematically searched for studies published from 1980 to 2017. Case series and comparative studies reported clinical data of BWT patients were included.

RESULTS: A total of 32 studies comprising 1457 patients were retained for primary outcome. Hemihypertrophy, cryptorchidism, and Beckwith-Wiedemann syndrome(BWS) are the most common congenital anomalies and syndrome. 86% of patients had favorable histology (FH). Patients with local stage I or II accounted for 64%, and 12.6% had metastasis at diagnosis. Bilateral nephron-sparing surgery (NSS) was achieved in 33.8%. Recurrence and renal failure occurred in 20% and 8%. The overall survival (OS) was 73%. In comparative studies, OS of patients undergoing bilateral NSS was similar to that of other operation types.

CONCLUSION: Prognosis of BWT has been improved but is significantly poorer than WT. Bilateral NSS was recommended by most centers to preserve more renal volume. However, finding a balance between retaining renal function and avoiding recurrence remains a question.

TYPE OF STUDY: Systematic review.

LEVEL OF EVIDENCE: Level IV.

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1

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MEDLINE

Authors Full Name

Han, Qilei; Li, Kai; Dong, Kuiran; Xiao, Xianmin; Yao, Wei; Liu, Gongbao.

Institution

Han, Qilei. Department of Pediatric Surgery, Children's Hospital of Fudan University, Shanghai, China. Li, Kai. Department of Pediatric Surgery, Children's Hospital of Fudan University, Shanghai, China. Electronic address: likai2727@163.com.

Dong, Kuiran. Department of Pediatric Surgery, Children's Hospital of Fudan University, Shanghai, China.

Xiao, Xianmin. Department of Pediatric Surgery, Children's Hospital of Fudan University, Shanghai, China.

Yao, Wei. Department of Pediatric Surgery, Children's Hospital of Fudan University, Shanghai, China.

Liu, Gongbao. Department of Pediatric Surgery, Children's Hospital of Fudan University, Shanghai, China.

Year of Publication

2018

297.

Impalpable Testis: Evaluation of Diagnostic and Treatment Procedures and Our Treatment Protocol.

Fratric I; Sarac D; Antic J; Dermanov M; Jokic R.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
BioMed Research International. 2018:3143412, 2018.

[Journal Article]

UI: 30112376

Introduction: The aim of this study is to present our treatment protocol for impalpable testis.

Material and Methods: In a retrospective study we analyzed clinical data including diagnostic procedures, intraoperative findings, final diagnosis, treatment modality, and outcome of patients with impalpable testis who underwent surgery from January 2010 until December 2015.

Results: Ninety-one patients were admitted under the diagnosis of impalpable testis. In 39 patients ultrasound detected testis in the inguinal canal and orchidopexy was done. In 25 patients (48.08%) laparoscopy showed the entrance of the spermatic cord into the inguinal canal. Open exploration of the inguinal canal was done, testicular remnant removed, and appropriate testicular prosthesis implanted. Twenty patients (20/52) underwent orchidopexy of the abdominal testis (46.51%), 4 of which underwent Fowler-Stevens procedure in two stages, and in 16 patients deliberation of the testis and spermatic cord was sufficient to place the testis into the scrotum.

Conclusions: Excision of the testicular nubbin is highly recommendable, as well as implantation of the testicular prosthesis at the time of orchiectomy.

Version ID

1

Status

MEDLINE

Author NameID

Fratric, Ivana; ORCID: <https://orcid.org/0000-0001-7124-8087>

Authors Full Name

Fratric, Ivana; Sarac, Dragan; Antic, Jelena; Dermanov, Marina; Jokic, Radoica.

Institution

Fratric, Ivana. Institute for Children and Youth Healthcare of Vojvodina, Clinic for Pediatric Surgery, Hajduk Veljkova 10, 21000 Novi Sad, Serbia. Fratric, Ivana. University of Novi Sad, Faculty of Medicine, Department of Surgery, Hajduk Veljkova 3, 21137 Novi Sad, Serbia.

Sarac, Dragan. University of Novi Sad, Faculty of Medicine, Department of Surgery, Hajduk Veljkova 3, 21137 Novi Sad, Serbia.

Antic, Jelena. Institute for Children and Youth Healthcare of Vojvodina, Clinic for Pediatric Surgery, Hajduk Veljkova 10, 21000 Novi Sad, Serbia.

Antic, Jelena. University of Novi Sad, Faculty of Medicine, Department of Surgery, Hajduk Veljkova 3, 21137 Novi Sad, Serbia.

Dermanov, Marina. Institute for Children and Youth Healthcare of Vojvodina, Clinic for Pediatric Surgery, Hajduk Veljkova 10, 21000 Novi Sad, Serbia.

Dermanov, Marina. University of Novi Sad, Faculty of Medicine, Department of Surgery, Hajduk Veljkova 3, 21137 Novi Sad, Serbia.

Jokic, Radoica. Institute for Children and Youth Healthcare of Vojvodina, Clinic for Pediatric Surgery, Hajduk Veljkova 10, 21000 Novi Sad, Serbia.

Jokic, Radoica. University of Novi Sad, Faculty of Medicine, Department of Surgery, Hajduk Veljkova 3, 21137 Novi Sad, Serbia.

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Year of Publication

2018

298.

Association between androgen receptor polymorphic CAG and GGC repeat lengths and cryptorchidism: A meta-analysis of case-control studies.

Wang Y; Wei Y; Tang X; Liu B; Shen L; Long C; Lin T; He D; Wu S; Wei G.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of pediatric urology. 14(5):432.e1-432.e9, 2018 10.

[Journal Article. Research Support, Non-U.S. Gov't]

UI: 29914823

INTRODUCTION: Previous studies have revealed the relationship between androgen receptor (AR) CAG and/or GGC polymorphisms and risk of cryptorchidism, yet the results have been elusive and controversial.

AIM: To determine whether AR polymorphic CAG and/or GGC repeats are related to cryptorchidism.

STUDY DESIGN: The relevant studies were obtained from PubMed, Embase, China National Knowledge Infrastructure, and Wanfang. The pooled odds ratios with 95% confidence intervals (CIs) were used to assess the strength of associations. Subgroup analyses were performed based on ethnicity and source of controls. Moreover, Begg's funnel plots and Egger's linear regression test were conducted to determine publication bias.

RESULTS: Eight case-control studies containing 321 patients and 784 normal controls were included. There was a significant association between longer CAG repeats and cryptorchidism risk (weighted mean difference (WMD) = 0.62; 95% CIs 0.06, 1.18; P = 0.031). Moreover, there was a significant association between the longer GGC repeats and cryptorchidism risk (WMD = 0.87; 95% CIs 0.04, 1.74; P = 0.040). There was significant association between the longer CAG repeats and bilateral cryptorchidism (WMD = 0.88; 95% CIs -0.18, 1.94; P = 0.011), while there was no significant association between the longer CAG repeats and unilateral cryptorchidism (WMD = -0.09; 95% CIs -0.50, 0.31; P = 0.554). There were significant associations between the longer GGC repeats and unilateral cryptorchidism (WMD = 0.88; 95% CIs -0.30, 2.05; P = 0.005) and bilateral cryptorchidism (WMD = 1.35; 95% CIs -0.52, 3.21; P = 0.000). Stratifying analysis revealed an association between longer CAG/GGC repeats and cryptorchidism in Caucasian populations from Europe (WMD = 0.73; 95% CIs 0.00, 1.46; P = 0.017), while there was no association with Asian populations.

DISCUSSION: This meta-analysis found that CAG/GGC repeats in the AR gene were longer in cryptorchidism patients compared to controls. Both the longer CAG repeats and GGC repeats in the AR gene were associated with cryptorchidism risk. The longer CAG repeats were associated with bilateral cryptorchidism, whereas the longer GGC repeats were associated with unilateral and bilateral cryptorchidism. Stratifying analysis revealed an association between longer CAG/GGC repeats and cryptorchidism in Caucasian populations from Europe, while there was no association between longer CAG/GGC repeats and cryptorchidism in Asian populations.

CONCLUSION: The CAG/GGC repeats in the AR gene were longer in cryptorchidism than in controls. Longer CAG repeats may play a role in determining bilateral cryptorchidism, and longer GGC repeats may play a role in determining unilateral and bilateral cryptorchidism. These observations were more applicable to Caucasian populations.

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Version ID

1

Status

MEDLINE

Authors Full Name

Wang, Y; Wei, Y; Tang, X; Liu, B; Shen, L; Long, C; Lin, T; He, D; Wu, S; Wei, G.

Institution

Wang, Y. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Chongqing Key Laboratory of Children's Urogenital Development and Tissue Engineering, China. Wei, Y. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Chongqing Key Laboratory of Children's Urogenital Development and Tissue Engineering, China.

Tang, X. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Chongqing Key Laboratory of Children's Urogenital Development and Tissue Engineering, China.

Liu, B. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Ministry of Education Key Laboratory of Child Development and Disorders, China.

Shen, L. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; China International Science and Technology Cooperation Base of Child Development and Critical Disorders, China.

Long, C. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Chongqing Key Laboratory of Pediatrics Chongqing, China.

Lin, T. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Ministry of Education Key Laboratory of Child Development and Disorders, China.

He, D. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Ministry of Education Key Laboratory of Child Development and Disorders, China.

Wu, S. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Chongqing Key Laboratory of Children's Urogenital Development and Tissue Engineering, China; Ministry of Education Key Laboratory of Child Development and Disorders, China.

Electronic address: shengdewu@yeah.net.

Wei, G. Department of Urology, Children's Hospital of Chongqing Medical University, Chongqing, China; Chongqing Key Laboratory of Children's Urogenital Development and Tissue Engineering, China; Ministry of Education Key Laboratory of Child Development and Disorders, China.

Year of Publication

2018

299.

Deficiency in GnRH receptor trafficking due to a novel homozygous mutation causes idiopathic hypogonadotropic hypogonadism in three prepubertal siblings.

Zhang R; Linpeng S; Li Z; Cao Y; Tan H; Liang D; Wu L.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Gene. 669:42-46, 2018 Aug 30.

[Journal Article]

UI: 29777911

Idiopathic hypogonadotropic hypogonadism (IHH) is characterized by low levels of gonadotropins and delayed or absent sexual development. Most of the patients are diagnosed in late adolescence or early adulthood. Determining the diagnosis of IHH in prepubertal patients can be challenging. Making a timely, correct diagnosis has important clinical implications. Here we aimed to identify the genetic cause of IHH in three prepubertal siblings from a Chinese Han family and give appropriate treatment advice. Using whole exome sequencing (WES), we identified a novel homozygous GNRHR mutation (NM_000406; c.364C>T, p.L122F) in two prepubertal boys with cryptorchidism and micropenis. Sanger sequencing showed that their younger asymptomatic sister also had the homozygous GNRHR mutation. This mutation was inherited from the father and the mother. Immunofluorescence analysis showed that in permeabilized cells, expression of the mutant receptor on the cell membrane was significantly lower than that of wild-type. Calcium mobilization assays demonstrated that c.364C>T in the GNRHR gene is a complete loss-of-function mutation that caused IHH. These results may contribute to the genetic diagnosis of the three prepubertal siblings with IHH. According to this diagnosis, timely hormonal treatment can be given for the three prepubertal patients to induce pubertal development, especially for the asymptomatic female.

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Version ID

1

Status

MEDLINE

Authors Full Name

Zhang, Rui; Linpeng, Siyuan; Li, Zhuo; Cao, Yingxi; Tan, Hu; Liang, Desheng; Wu, Lingqian.

Institution

Zhang, Rui. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China; Department of Prenatal Diagnosis, Bao'an Maternity and Child Health Hospital, Shenzhen, China. Linpeng, Siyuan. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China.

Li, Zhuo. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China.

Cao, Yingxi. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China.

Tan, Hu. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China.

Liang, Desheng. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China. Electronic address: liangdesheng@sklmg.edu.cn.

Wu, Lingqian. Center for Medical Genetics, School of Life Sciences, Central South University, Changsha, China. Electronic address: wulingqian@sklmg.edu.cn.

Year of Publication

2018

300.

Trends in the Prevalences of Selected Birth Defects in Korea (2008-2014).

Ko JK; Lamichhane DK; Kim HC; Leem JH.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

International Journal of Environmental Research & Public Health [Electronic Resource]. 15(5), 2018 05 05.

[Journal Article. Research Support, Non-U.S. Gov't]

UI: 29734759

Little information is available on the prevalences of birth defects in Korea. The aims of this study were to estimate recent prevalences of selected birth defects and to analyze the prevalence trends of these defects during the period from 2008 to 2014. Prevalences were calculated for 69 major birth defects using health insurance claim data obtained from the Korea National Health Insurance Service (NHIS). Prevalence rate ratios were calculated using Poisson regression to analyze trends over the 7-year study period. The overall prevalence of a major birth defect was

446.3 per 10,000 births (95% CI: 444.0-448.6); 470.9 per 10,000 births (95% CI: 467.6-474.2) for males and 420.2 per 10,000 births (95% CI: 417-423.4) for females. The prevalence rates of the most common birth defects over the study period were; septal defect (138.2 per 10,000; 95% CI: 136.9-139.5), congenital hip dislocation (652 per 10,000; 95% CI: 64.1-65.9), and ventricular septal defect (62.62 per 10,000; 95% CI: 61.7-63.5). During the study period, a significant increase in the prevalence of a major birth defect was observed with a prevalence rate ratio (PRR) of 1.091. The strongest trend was observed for renal dysplasia, which had a PRR of 1.275 (95% CI: 1.211-1.343), and upward trends were observed for urogenital anomalies, such as, renal agenesis (PRR 1.102, 95% CI: 1.067-1.138), undescended testis (PRR 1.082, 95% CI: 1.072-1.093) and hypospadias (PRR 1.067, 95% CI: 1.044-1.090). This study shows an overall increase in the prevalences of birth defects, including hypospadias and undescended testis, which are known to be associated with endocrine factors. In the future, standardized birth defect registries should be established to enable these trends to be monitored.

Version ID

1

Status

MEDLINE

Authors Full Name

Ko, Jung-Keun; Lamichhane, Dirga Kumar; Kim, Hwan-Cheol; Leem, Jong-Han.

Institution

Ko, Jung-Keun. Department of Social and Preventive Medicine, School of Medicine, Inha University, Incheon 22212, Korea. ecokjk@naver.com. Lamichhane, Dirga Kumar. Department of Social and Preventive Medicine, School of Medicine, Inha University, Incheon 22212, Korea. dirgalamichhane@gmail.com.

Kim, Hwan-Cheol. Department of Social and Preventive Medicine, School of Medicine, Inha University, Incheon 22212, Korea. cheol17@hanmail.net.

Kim, Hwan-Cheol. Department of Occupational and Environmental Medicine, School of Medicine, Inha University, Incheon 22212, Korea. cheol17@hanmail.net.

Leem, Jong-Han. Department of Social and Preventive Medicine, School of Medicine, Inha University, Incheon 22212, Korea. ekeeper21@naver.com.

Leem, Jong-Han. Department of Occupational and Environmental Medicine, School of Medicine, Inha University, Incheon 22212, Korea. ekeeper21@naver.com.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5981962>

Year of Publication

2018

301.

Staged laparoscopic traction orchiopexy for intra-abdominal testis: Is it always feasible?.

Elsherbeny M; Abdallah A; Abouzeid A; Ghanem W; Zaki A.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of pediatric urology. 14(3):267.e1-267.e4, 2018 06.

[Journal Article. Multicenter Study]

UI: 29567012

BACKGROUND AND OBJECTIVES: Laparoscopic orchiopexy for intra-abdominal testis is a well-known and commonly practiced technique. The traction technique is based on elongation of the testicular vessels without cutting them, in contrast with the two-stage Fowler-Stephens technique in which the testicular vessels are divided. The current study evaluated the feasibility of the traction technique according to the type of intra-abdominal testis.

PATIENTS AND METHODS: The study included 20 boys with 22 intra-abdominal testes and who underwent staged laparoscopic traction orchiopexy between October 2013 and October 2015. Eighteen testes were proximal to the internal ring by <2 cm (type III), while the remaining four were high and away from the internal ring by >2 cm (type IVa). The patients' ages ranged between 6 months and 5 years (mean 2.2 years; median 2 years). Patients were followed up for 1 year for testicular atrophy and satisfactory scrotal site relocation.

RESULTS: This study did not detect any cases of testicular atrophy among the participants (0/22), while failure to achieve satisfactory scrotal site was found in two out of the four cases with type IVa intra-abdominal testes.

DISCUSSION: Traction techniques for elongating the testicular vessels were employed years ago, but this was revisited by Shehata in 2008. It has the advantage of preserving the main testicular blood supply, which can be reflected by the decreasing rate of testicular atrophy when compared with the Fowler-Stephens orchiopexy. The results of the traction technique may be less satisfactory regarding the testicular location for higher types of intra-abdominal testes (type IVa).

CONCLUSION: Staged laparoscopic traction orchiopexy was a feasible technique for intra-abdominal testes (within 2 cm of the internal ring), but the results were less satisfactory when applied for higher intra-abdominal testes (type IVa).

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Version ID

1

Status

MEDLINE

Authors Full Name

Elsherbeny, Mohammed; Abdallah, Ahmed; Abouzeid, Amr; Ghanem, Wael; Zaki, Ahmed.

Institution

Elsherbeny, Mohammed. Faculty of Medicine, Ain Shams University, Cairo, Egypt. Electronic address: mohamedsaid@med.asu.edu.eg. Abdallah, Ahmed. Benha Children's Hospital, Qalyoubyah, Egypt.

Abouzeid, Amr. Faculty of Medicine, Ain Shams University, Cairo, Egypt.

Ghanem, Wael. Faculty of Medicine, Ain Shams University, Cairo, Egypt.

Zaki, Ahmed. Faculty of Medicine, Ain Shams University, Cairo, Egypt.

Year of Publication

2018

302.

Semen quality associated with subsequent hospitalizations - Can the effect be explained by socio-economic status and lifestyle factors?.

Latif T; Lindahl-Jacobsen R; Mehlsen J; Eisenberg ML; Holmboe SA; Pors K; Brinth L; Skouby SO; Jorgensen N; Jensen TK.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Andrology. 6(3):428-435, 2018 05.

[Journal Article. Research Support, Non-U.S. Gov't]

UI: 29481730

Semen quality is suggested to be a universal biomarker for future health. Previous studies have mostly been registry based excluding the possibility to address the importance of lifestyle, fertility status, health and socio-economic status. We aimed to investigate whether the association between semen quality and subsequent risk of hospitalization could be explained by differences in occupation, education, fertility, cryptorchidism, BMI or smoking; 1423 men with first semen sample at Fertility Clinic, Frederiksberg Hospital, Denmark, from 1977 to 2010 responded to a

questionnaire in 2012 about current health, lifestyle, educational level and occupation. They were followed in the Danish National Patient Registry to first-time hospitalizations using ICD-8 and ICD-10 classification. Data were analysed by Cox proportional hazard regression models to adjust for the possible confounding factors. We found a significant higher risk of being hospitalized with decreasing sperm concentrations (0-15 mill/mL: HR1.78, 95% CI:1.51-2.09; 16-50 mill/mL: HR 1.37 95% CI: 1.17-1.60; 51-100 mill/mL: HR1.25 95% CI: 1.07-1.45). Same significant association of being hospitalized with decreasing total sperm counts was seen. The dose-response increase in risk in hospitalization with decreasing sperm concentration and total sperm count remained constant after further individual adjustment for occupation, marital status, fertility, cryptorchidism, BMI or smoking. The association between semen quality and subsequent morbidity was not explained by differences in lifestyle, behavioural or fertility status. We were unable to adjust for all possible confounders simultaneously due to limited sample size, and reverse causation is a possible explanation as information about education and lifestyle was obtained after semen analysis and hospitalizations occurred and may have changed as consequence of both. Semen quality may be a universal biomarker for future health not explained by lifestyle and socio-economic status, but this needs to be addressed further in future studies. Copyright © 2018 American Society of Andrology and European Academy of Andrology.

Version ID

1

Status

MEDLINE

Author NameID

Latif, T; ORCID: <https://orcid.org/0000-0002-6250-7208> Jorgensen, N; ORCID: <https://orcid.org/0000-0003-4827-0838>

Authors Full Name

Latif, T; Lindahl-Jacobsen, R; Mehlsen, J; Eisenberg, M L; Holmboe, S A; Pors, K; Brinth, L; Skouby, S O; Jorgensen, N; Jensen, T K.

Institution

Latif, T. Coordinating Research Centre, Bispebjerg & Frederiksberg Hospitals, Frederiksberg, Denmark. Latif, T. Department of Environmental Medicine, Institute of Public Health, University of Southern Denmark, Odense, Denmark.

Lindahl-Jacobsen, R. Danish Aging Research Centre, Unit of Epidemiology, Biostatistics and Biodemography, University of Southern Denmark, Odense, Denmark.

Lindahl-Jacobsen, R. Max-Planck Odense Centre on the Biodemography of Aging, University of Southern Denmark, Odense, Denmark.

Mehlsen, J. Coordinating Research Centre, Bispebjerg & Frederiksberg Hospitals, Frederiksberg, Denmark.

Eisenberg, M L. Departments of Urology and Obstetrics/Gynaecology, Stanford University School of Medicine, Stanford, CA, USA.

Holmboe, S A. University Department of Growth and Reproduction and International Centre for Research and Research Training in Endocrine Disruption of Male Reproduction and Child Health (EDMaRC), Rigshospitalet, Copenhagen, Denmark.

Pors, K. Coordinating Research Centre, Bispebjerg & Frederiksberg Hospitals, Frederiksberg, Denmark.

Brinth, L. Coordinating Research Centre, Bispebjerg & Frederiksberg Hospitals, Frederiksberg, Denmark.

Skouby, S O. Department of Gynaecology and Obstetrics, Faculty of Health and Medical Sciences, Herlev & Gentofte Hospital, University of Copenhagen, Copenhagen, Denmark.

Jorgensen, N. University Department of Growth and Reproduction and International Centre for Research and Research Training in Endocrine Disruption of Male Reproduction and Child Health (EDMaRC), Rigshospitalet, Copenhagen, Denmark.

Jensen, T K. Department of Environmental Medicine, Institute of Public Health, University of Southern Denmark, Odense, Denmark.

Year of Publication

2018

303.

A collection of XY female cell lines.

Kasai F; Ferguson-Smith MA.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Human Cell. 31(2):175-178, 2018 Apr.

[Journal Article]

UI: 29330774

Discordance between sexual phenotype and the 46,XY sex chromosome complement may be found in certain disorders of sexual development (DSD). Many of these DSD patients with female external genitalia and secondary sex characteristics have undescended testes and male internal genitalia. Causative mutations involving genes of the sex determining pathway, including the androgen receptor, SRY and the 5-alpha-reductase genes, are well-known, but the origin of other

cases remain unresolved. In this report, we introduce our collection of lymphoblastoid lines derived from female patients with a 46,XY karyotype. These cell lines have been deposited and registered with the JCRB Cell Bank. They are available for comparison with other DSD cases and for further characterization of genetic loci involved in the mammalian sex determining pathway.

Version ID

1

Status

MEDLINE

Authors Full Name

Kasai, Fumio; Ferguson-Smith, Malcolm A.

Institution

Kasai, Fumio. Japanese Collection of Research Bioresources (JCRB) Cell Bank, Laboratory of Cell Cultures, National Institutes of Biomedical Innovation, Health and Nutrition, Saito-Asagi, Ibaraki, Osaka, 567-0085, Japan. k-230@umin.ac.jp. Kasai, Fumio. Department of Veterinary Medicine, University of Cambridge, Madingley Road, Cambridge, CB3 0ES, UK. k-230@umin.ac.jp.

Ferguson-Smith, Malcolm A. Department of Veterinary Medicine, University of Cambridge, Madingley Road, Cambridge, CB3 0ES, UK. maf12@cam.ac.uk.

Year of Publication

2018

304.

Surgical exploration for impalpable testis: Which should be first, inguinal exploration or laparoscopic abdominal exploration?. [Review]

Igarashi A; Kikuchi K; Ogushi K; Hasegawa M; Hatanaka M; Fujino J; Kishi Y; Ikeda H.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Pediatric Surgery. 53(9):1766-1769, 2018 Sep.

[Journal Article. Review]

UI: 29132799

PURPOSE: To discuss an optimal surgical approach for impalpable testis in children, our own treatment results and those reported in the literature were reviewed.

MATERIALS AND METHODS: Seventy-two impalpable testes were diagnosed in 68 patients: unilateral in 64 patients and bilateral in 4 patients. All patients underwent surgical exploration at the ages of 6 to 140 months (median, 15 months). The inguinal canal was initially explored, and abdominal exploration was performed with laparoscopy when an extra-abdominal testis was not identified. In addition, articles regarding surgical exploration for impalpable testis, published over the last 20 years, were retrieved and the results were examined.

RESULTS: Testes were detected by inguinal exploration in 28 of 72 (39%) impalpable testes: intracanalicular in 22 testes and at the internal inguinal ring (peeping or low abdominal testis) in 6 testes. All these testes were treated by conventional inguinal orchidopexy. Laparoscopic exploration was performed in 44 (61%) impalpable testes, and 4 (5.6%) high abdominal testes were detected and treated by two-stage Fowler-Stephens orchidopexy. Vanishing or absent testis was the final diagnosis in the remaining 40 testes (55.6%). The literature review showed that the ratios of intra- and extra-abdominal testes were lower in the articles that reported the results of inguinal or scrotal exploration than in those of laparoscopic exploration, although the difference was not significant.

CONCLUSIONS: Considering the relatively low incidence of high abdominal testis, we recommend to start with inguinal exploration for impalpable testis. When an extra-abdominal testis is not detected, transinguinal laparoscopic exploration should be indicated.

LEVEL OF EVIDENCE: Treatment study, Level IV.

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Version ID

1

Status

MEDLINE

Authors Full Name

Igarashi, Akihiro; Kikuchi, Kenta; Ogushi, Kenjiro; Hasegawa, Mariko; Hatanaka, Masahiro; Fujino, Junko; Kishi, Yoko; Ikeda, Hitoshi.

Institution

Igarashi, Akihiro. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan. Kikuchi, Kenta. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan.

Ogushi, Kenjiro. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan.

Hasegawa, Mariko. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan.

Hatanaka, Masahiro. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan.

Fujino, Junko. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan.

Kishi, Yoko. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan.

Ikeda, Hitoshi. Department of Pediatric Surgery, Dokkyo Medical University Koshigaya Hospital, 2-1-50, Minami-Koshigaya, Koshigaya, Saitama, 343-8555, Japan. Electronic address:

hike@dokkyomed.ac.jp.

Year of Publication

2018

305.

Prenatal exposure to glycol ethers and cryptorchidism and hypospadias: a nested case-control study.

Warembourg C; Botton J; Lelong N; Rouget F; Khoshnood B; Le Gleau F; Monfort C; Labat L; Pierre F; Heude B; Slama R; Multigner L; Charles MA; Cordier S; Garlantezec R.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Occupational & Environmental Medicine. 75(1):59-65, 2018 01.

[Journal Article. Research Support, Non-U.S. Gov't]

UI: 29055888

OBJECTIVES: Glycol ethers (GE) are oxygenated solvents frequently found in occupational and consumer products. Some of them are well-known testicular and developmental animal toxicants. This study aims to evaluate the risk of male genital anomalies in association with prenatal exposure to GE using urinary biomarkers of exposure.

METHODS: We conducted a case-control study nested in two joint mother-child cohorts (5303 pregnant women). Cases of cryptorchidism and hypospadias were identified at birth and confirmed during a 2-year follow-up period (n=14 cryptorchidism and n=15 hypospadias). Each case was matched to three randomly selected controls within the cohorts for region of inclusion and gestational age at urine sampling. Concentrations of five GE acidic metabolites were

measured in spot maternal urine samples collected during pregnancy. ORs were estimated with multivariate conditional logistic regressions including a Firth's penalisation.

RESULTS: Detection rates of urinary GE metabolites ranged from 8% to 93% and only two were sufficiently detected (>33%) in each cohort to be studied: methoxyacetic acid (MAA) and phenoxyacetic acid (PhAA). A significantly higher risk of hypospadias was associated with the highest tertile of exposure to MAA: OR (95% CI) 4.5(1.4 to 23.4). No association were observed with urinary concentration of PhAA, nor with the risk of cryptorchidism.

CONCLUSIONS: In view of the toxicological plausibility of our results, this study, despite its small sample size, raises concern about the potential developmental toxicity of MAA on the male genital system and calls for thorough identification of current sources of exposure to MAA.

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1

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Authors Full Name

Warembourg, Charline; Botton, Jeremie; Lelong, Nathalie; Rouget, Florence; Khoshnood, Babak; Le Gleau, Florent; Monfort, Christine; Labat, Laurence; Pierre, Fabrice; Heude, Barbara; Slama, Remy; Multigner, Luc; Charles, Marie-Aline; Cordier, Sylvaine; Garlantezec, Ronan.

Institution

Warembourg, Charline. Exposure Assessment and Epidemiological Research on Environment, Reproduction and Development, INSERM UMR 1085 IRSET, Rennes, France. Warembourg, Charline. Universite de Rennes I, Rennes, France.

Botton, Jeremie. Center for Epidemiology and Statistics Sorbonne Paris Cite, INSERM UMR 1153, Team: Early Origin of the Child's Health and Development, Paris Descartes University, Villejuif, France.

Botton, Jeremie. Faculty of Pharmacy, University Paris Saclay, Malabry, France.

Lelong, Nathalie. Center for Epidemiology and Statistics Sorbonne Paris Cite, DHU Risks in Pregnancy, INSERM UMR 1153, Team: Obstetrical, Perinatal and Pediatric Epidemiology Research, Paris Descartes University, Paris, France.

Rouget, Florence. Exposure Assessment and Epidemiological Research on Environment, Reproduction and Development, INSERM UMR 1085 IRSET, Rennes, France.

Rouget, Florence. Universite de Rennes I, Rennes, France.

Rouget, Florence. Department of Pediatrics, University Hospital of Rennes, Brittany Registry of Congenital Malformations, Rennes, France.

Khoshnood, Babak. Center for Epidemiology and Statistics Sorbonne Paris Cite, DHU Risks in Pregnancy, INSERM UMR 1153, Team: Obstetrical, Perinatal and Pediatric Epidemiology Research, Paris Descartes University, Paris, France.

Le Gleau, Florent. LABOCEA Laboratory, Plouzane, France.

Monfort, Christine. Exposure Assessment and Epidemiological Research on Environment, Reproduction and Development, INSERM UMR 1085 IRSET, Rennes, France.

Monfort, Christine. Universite de Rennes I, Rennes, France.

Labat, Laurence. Laboratoire de Pharmacologie Toxicologie, Assistance Publique-Hopitaux de Paris, Groupe Cochin, Paris, France.

Pierre, Fabrice. Service de Gynecologie Obstetrique et Medecine de la Reproduction, Centre Hospitalier Universitaire de Poitiers, Poitiers, France.

Heude, Barbara. Center for Epidemiology and Statistics Sorbonne Paris Cite, INSERM UMR 1153, Team: Early Origin of the Child's Health and Development, Paris Descartes University, Villejuif, France.

Slama, Remy. Department of Environmental Epidemiology, INSERM, CNRS, University Grenoble-Alpes, IAB Institute of Advanced Biosciences Joint Research Center (U1209), Grenoble, France.

Multigner, Luc. Exposure Assessment and Epidemiological Research on Environment, Reproduction and Development, INSERM UMR 1085 IRSET, Rennes, France.

Multigner, Luc. Universite de Rennes I, Rennes, France.

Charles, Marie-Aline. Center for Epidemiology and Statistics Sorbonne Paris Cite, INSERM UMR 1153, Team: Early Origin of the Child's Health and Development, Paris Descartes University, Villejuif, France.

Cordier, Sylvaine. Exposure Assessment and Epidemiological Research on Environment, Reproduction and Development, INSERM UMR 1085 IRSET, Rennes, France.

Garlantezec, Ronan. Exposure Assessment and Epidemiological Research on Environment, Reproduction and Development, INSERM UMR 1085 IRSET, Rennes, France.

Garlantezec, Ronan. Universite de Rennes I, Rennes, France.

Garlantezec, Ronan. Service de Sante Publique et d'Epidemiologie, Centre Hospitalo-Universitaire de Rennes, Rennes, France.

Comments

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Year of Publication

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306.

Evaluation of two-staged Fowler-Stephens laparoscopic orchidopexy (FSLO) for intra-abdominal testes (IAT).

Bagga D; Prasad A; Grover SB; Sugandhi N; Tekchandani N; Acharya SK; Samie A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Pediatric Surgery International. 34(1):97-103, 2018 Jan.

[Journal Article]

UI: 28980063

INTRODUCTION: The best operative intervention for intrabdominal testis (IAT) has not been standardized as yet. The question of whether to bring down an IAT with a single-staged vessel-intact laparoscopic orchidopexy (VILO) or a two-staged laparoscopic Fowler-Stephens orchidopexy (FSLO) is still undergoing debate, with both the procedures being popular. The present study has been designed to evaluate the factors predicting the success or failure of two-staged FSLO for (IAT).

METHODS: 43 boys with 49 non-palpable testes underwent diagnostic laparoscopy out of which 35 underwent two-staged FSLO. Size of the testis was measured with a graduated probe in both stages. Independent variables such as age, height, testis-to-internal ring distance (T-IR), neo internal ring-to-midscrotal distance (NIR-MS), and mobility-to-contralateral ring (MCIR) were analysed. Postoperatively 34 IATs were followed up clinically as well as ultrasonologically after 6 months, to see for the size, position, and vascularity. Based on this, the patients were divided into two groups, Group A (successful) and Group B (Failed).

RESULTS: 24 IATs had a successful outcome (Group A) and 11 were failure (Group B). The overall success rate of the study was 68.6%. The difference in mean age of patients in both groups was insignificant ($p = 0.89$) (Fig. 1), and similarly, the difference in mean height was insignificant ($p = 0.61$). The difference in mean T-IR in both the groups was insignificant (1.85 versus 2.77 cm; $p = 0.09$) and mean NIR-MS was 5.41 cm in Group A and 5.10 cm in Group B, and the difference again was insignificant ($p = 0.23$).

CONCLUSION: The success rate of FSLO was 68.6%. None of the above-described independent variables have any effect on the outcome of two-staged FSLO. While VILO remains the treatment of choice for IAT located at or near the ring, but IAT higher than this, two-staged FSLO gives a better chance for achieving intra-scrotal orchidopexy.

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Authors Full Name

Bagga, Deepak; Prasad, Ashish; Grover, Shabnam Bhandari; Sugandhi, Nidhi; Tekchandani, Narender; Acharya, Samir Kant; Samie, Amat.

Institution

Bagga, Deepak. Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India. Prasad, Ashish. Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India. drashishprasad82@gmail.com.

Grover, Shabnam Bhandari. Department of Radiology, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India.

Sugandhi, Nidhi. Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India.

Tekchandani, Narender. Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India.

Acharya, Samir Kant. Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India.

Samie, Amat. Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, 110029, India.

Year of Publication

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307.

Prune belly syndrome: Approaches to its diagnosis and management.

Achour R; Bennour W; Ksibi I; Cheour M; Hamila T; Hmid RB; Kacem S.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Irr. 7(4):271-274, 2018 Nov.

[Journal Article]

UI: 30560020

Prune Belly syndrome (PBS) or Eagle-Barrett syndrome is an anatomo-radiological syndrome consisting of a complex and rare malformation characterized by the following triad of symptoms: deficiency of the abdominal muscles, malformations of the urinary tract, and bilateral cryptorchidism. The exact etiology is unknown, though PBS predominantly occurs in males. The clinical manifestations can vary widely, from stillbirth to renal and major respiratory dysplasia to almost normal children. The current study included a total of 3 patients. The findings included clinical characteristics, diagnostics, therapy, and clinical outcomes. All patients were diagnosed with congenital aplasia of the abdominal wall and a variety of urogenital malformations. Cryptorchidism and a mega-bladder were observed in 2 patients and distinctive renal malformations, such as renal dysplasia, were observed in 1 patient. Treatment varies but usually includes surgical management of symptoms. One patient required urgent urinary surgery; a vesicotomy was urgently performed due to anuria. These aspects explain the great diversity of opinions on the approach to this syndrome, but the severity of renal dysplasia is the main prognostic factor. Two newborns died a few days later due to severe renal failure. Despite these concerns, many patients with PBS report being in physical and mental health and having a good quality of life.

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1

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Authors Full Name

Achour, Radhouane; Bennour, Wafa; Ksibi, Imen; Cheour, Meriem; Hamila, Tarek; Hmid, Rim Ben; Kacem, Samia.

Institution

Achour, Radhouane. Emergency Department of Gynecology and Obstetrics, Maternity and Neonatology Center, Faculty of Medicine, Tunis-EI Manar University, Tunis, Tunisia. Bennour, Wafa. Neonatology Department, Maternity and Neonatology Center, Faculty of Medicine, Tunis-EI Manar University, Tunis, Tunisia.

Ksibi, Imen. Neonatology Department, Maternity and Neonatology Center, Faculty of Medicine, Tunis-EI Manar University, Tunis, Tunisia.

Cheour, Meriem. Neonatology Department, Maternity and Neonatology Center, Faculty of Medicine, Tunis-EI Manar University, Tunis, Tunisia.

Hamila, Tarek. Emergency Department of Gynecology and Obstetrics, Maternity and Neonatology Center, Faculty of Medicine, Tunis-EI Manar University, Tunis, Tunisia.

Hmid, Rim Ben. Emergency Department of Gynecology and Obstetrics, Maternity and Neonatology Center, Faculty of Medicine, Tunis-EI Manar University, Tunis, Tunisia.

Kacem, Samia. Neonatology Department, Maternity and Neonatology Center, Faculty of Medicine, Tunis-El Manar University, Tunis, Tunisia.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6290839>

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308.

Undescended Testes and Laparoscopy: Experience from the Developing World.

Dar SA; Bali RS; Zahoor Y; Rashid Kema A; Bhardwaj R.

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Advances in Urology. 2018:1620470, 2018.

[Journal Article]

UI: 30473708

Background: Cryptorchidism or undescended testes is the most common disorder of the male endocrine glands in children. With the advancements in laparoscopic techniques and instruments, laparoscopic orchidopexy has become the standard procedure in the management of nonpalpable undescended testes.

Aim: To evaluate and determine the therapeutic role, sensitivity, and specificity of laparoscopy in localizing nonpalpable testes and the mean operative time, the conversion rate (and reasons thereof), postoperative wound infection, postoperative stay, and time taken for return to daily activities following laparoscopic orchidopexy or orchidectomy.

Materials and Methods: This was a prospective study carried out in the Postgraduate Department of Surgery, Government Medical College, Srinagar, J&K, India, from May 2008 to August 2011.

All patients who presented to the outpatient department with complaints of absent testes were examined, and the ones with nonpalpable testes were included in the study.

Results: The mean operative time for bilateral and unilateral nonpalpable testis was 102.76 and 53.67 minutes, respectively. Minor postoperative wound infections were noted in 4 of our patients.

Mean duration of hospital stay was 14.23 hrs for unilateral cases and 16.27 hrs for bilateral cases. Patients who underwent laparoscopic orchidopexy resumed their normal activities within 4 +/- 1 days.

Conclusion: Laparoscopy clearly demonstrates the anatomy and provides visual information upon which a definitive decision can be made for further management of the undescended nonpalpable testis.

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Author NameID

Bali, Rajandeep Singh; ORCID: <https://orcid.org/0000-0001-6972-9551> Rashid Kema, Arshad;

ORCID: <https://orcid.org/0000-0002-7109-7773>

Authors Full Name

Dar, Shabir Ahmad; Bali, Rajandeep Singh; Zahoor, Yawar; Rashid Kema, Arshad; Bhardwaj, Rajni.

Institution

Dar, Shabir Ahmad. MS General Surgery, Resident Surgeon, SKIMS, Jammu and Kashmir, India.

Bali, Rajandeep Singh. MS General Surgery, Consultant Surgeon, GMC-Jammu, Jammu and Kashmir, India.

Zahoor, Yawar. MS General Surgery, Resident Surgeon, MAMC, Delhi, India.

Rashid Kema, Arshad. MS General Surgery, Consultant Surgeon, DHSK, Jammu and Kashmir, India.

Bhardwaj, Rajni. MS General Surgery, Consultant Surgeon, GMC-Jammu, Jammu and Kashmir, India.

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Year of Publication

2018

309.

Single-Port Laparoscopic Assisted Transcrotal Orchidopexy for Palpable Inguinal Canalicular Cryptorchidism Accompany With Indirect Inguinal Hernia.

Ma Y; Cai J; Li S; Wang W; Liu L.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Frontiers in Pediatrics. 6:293, 2018.

[Journal Article]

UI: 30356669

Purpose: To assess the outcomes of a novel laparoscopic assisted transrotal orchidopexy (LATO) combined with percutaneous extraperitoneal closure (PEC) for palpable inguinal canalicular cryptorchidism accompany with indirect inguinal hernia, and evaluate its safety and efficiency. **Materials and Methods:** A retrospective cohort study for single-port LATO-PEC and traditional inguinal orchidopexy (TIO) was performed between 2011 and 2014. Totally 53 children with both palpable inguinal canalicular testes and indirect inguinal hernia were included. Median patient age was 15month (range, 6 months to 4 years). Of them, 35 patients underwent LATO-PEC procedure, utilizing an umbilical trocar for laparoscope, transrotal dissection for orchidopexy, and an inner two-hooked cannula for ligation of the patent processus at the level of the internal ring. Three of them were bilateral, 12 on the left side and 20 on the right. Eighteen patients received TIO, seven of them on the left side and 11 on the right. Patient demographics, surgical technique, complications, and clinical outcomes were reviewed. Follow-up visits were performed to reassess position and size of the testes.

Results: All 56 undescended testes were delivered into the scrotum successfully. In the LATO-PEC group, nine contralateral herniorrhaphy were accomplished simultaneously. Fifteen contralateral patent processus vaginalis (PPVs) in 32 unilateral undescended testis (UDT) were newly confirmed during the laparoscopy, while 6 of them received percutaneous extra-peritoneal herniorrhaphy for visible inguinal bubble in pneumoperitoneum condition. No additional port placement or conversion to open procedure was needed. Mean operative time for unilateral and bilateral LATO-PEC in this study was (37.81 +/- 5.23) min and (53.33 +/- 2.98) min, respectively. In TIO group, mean operative time was (41.11 +/- 8.67) min. There was no statistical difference in operative time between the two approaches for unilateral UDTs ($p = 0.098$). Median follow-up interval was 24 months (range, 12-84 months). No operative complications were found in either group to date.

Conclusions: Single-port LATO-PEC is a safe, effective, and cosmetic choice for inguinal canalicular cryptorchidism accompany with indirect inguinal hernia, minimizing injuries to the vas deferens and testicular vessels. Laparoscopy can provide a diagnostic and therapeutic solution of contralateral PPV.

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1

Status

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Authors Full Name

Ma, Yazhen; Cai, Jianhui; Li, Suolin; Wang, Wenbo; Liu, Lin.

Institution

Ma, Yazhen. Department of Surgery, Graduate School of Hebei Medical University, Shijiazhuang, China. Ma, Yazhen. Department of Pediatric Surgery, The Second Hospital of Hebei Medical University, Shijiazhuang, China.

Cai, Jianhui. Department of Surgery, Graduate School of Hebei Medical University, Shijiazhuang, China.

Cai, Jianhui. Department of General Surgery, Hebei General Hospital, Shijiazhuang, China.

Li, Suolin. Department of Surgery, Graduate School of Hebei Medical University, Shijiazhuang, China.

Li, Suolin. Department of Pediatric Surgery, The Second Hospital of Hebei Medical University, Shijiazhuang, China.

Wang, Wenbo. Department of Pediatric Surgery, The Second Hospital of Hebei Medical University, Shijiazhuang, China.

Liu, Lin. Department of Pediatric Surgery, The Second Hospital of Hebei Medical University, Shijiazhuang, China.

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2018

310.

Male Central Hypogonadism in Paediatrics - the Relevance of Follicle-stimulating Hormone and Sertoli Cell Markers. [Review]

Grinson RP; Urrutia M; Rey RA.

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European Endocrinology. 14(2):67-71, 2018 Sep.

[Journal Article. Review]

UI: 30349597

The definition of male hypogonadism, used in adult endocrinology, is not fully applicable to paediatrics. A clear understanding of the developmental physiology of the hypothalamic-pituitary-testicular axis is essential for the comprehension of the pathogenesis of hypogonadal states in boys and for the establishment of adequate definitions and classifications in paediatric ages. This is particularly true for central hypogonadism, usually called hypogonadotropic in adults. Because childhood is a period characterised by a physiological state of low gonadotropin and testosterone production, these markers of hypogonadism, typically used in adult endocrinology, are uninformative in the child. This review is focused on the physiological importance of prepubertal Sertoli cell markers - anti-Mullerian hormone (AMH) and inhibin B - and of the intratesticular actions of follicle-stimulating hormone (FSH) and testosterone during early infancy and the first stages of pubertal development. We discuss the role of FSH in regulating the proliferation of Sertoli cells - the main determinant of prepubertal testicular volume - and the secretion of AMH and inhibin B. We also address how intratesticular testosterone concentrations have different effects on the seminiferous tubule function in early infancy and during pubertal development.

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1

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Authors Full Name

Grinspon, Romina P; Urrutia, Mariela; Rey, Rodolfo A.

Institution

Grinspon, Romina P. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), National Scientific and Technical Research Council (CONICET) - Fundacion de Endocrinologia Infantil (FEI) - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina. Urrutia, Mariela. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), National Scientific and Technical Research Council (CONICET) - Fundacion de Endocrinologia Infantil (FEI) - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina.

Rey, Rodolfo A. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), National Scientific and Technical Research Council (CONICET) - Fundacion de Endocrinologia Infantil (FEI) - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina.

Rey, Rodolfo A. Departamento de Histologia, Biologia Celular, Embriologia y Genetica, Facultad de Medicina, Universidad de Buenos Aires, Buenos Aires, Argentina.

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Year of Publication

311.

A rare variety of congenital adrenal hyperplasia with mosaic Klinefelter syndrome: a unique combination presenting with ambiguous genitalia and sexual precocity.

Shehab MA; Mahmood T; Hasanat MA; Fariduddin M; Ahsan N; Hossain MS; Hossain MS; Jahan S.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Endocrinology, Diabetes & Metabolism Case Reports. 2018(1):18-0108, 2018 Oct 13.

[Journal Article]

UI: 30328339

Congenital adrenal hyperplasia (CAH) due to the three-beta-hydroxysteroid-dehydrogenase (3beta-HSD) enzyme deficiency is a rare autosomal recessive disorder presenting with sexual precocity in a phenotypic male. Klinefelter syndrome (KS) is the most common sex chromosome aneuploidy presenting with hypergonadotropic hypogonadism in a male. However, only a handful of cases of mosaic KS have been described in the literature. The co-existence of mosaic KS with CAH due to 3beta-HSD enzyme deficiency portrays a unique diagnostic paradox where features of gonadal androgen deficiency are masked by simultaneous adrenal androgen excess. Here, we report a 7-year-old phenotypic male boy who, at birth presented with ambiguous genitalia, probably a microphallus with penoscrotal hypospadias. Later on, he developed accelerated growth with advanced bone age, premature pubarche, phallic enlargement and hyperpigmentation. Biochemically, the patient was proven to have CAH due to 3beta-HSD deficiency. However, the co-existence of bilateral cryptorchidism made us to consider the possibility of hypogonadism as well, and it was further explained by concurrent existence of mosaic KS (47,XXY/46,XX). He was started on glucocorticoid and mineralocorticoid replacement and underwent right-sided orchidopexy on a later date. He showed significant clinical and biochemical improvement on subsequent follow-up. However, the declining value of serum testosterone was accompanied by rising level of FSH thereby unmasking hypergonadotropic hypogonadism due to mosaic KS. In future, we are planning to place him on androgen replacement as well. Learning points: ** Ambiguous genitalia with subsequent development of sexual precocity in a phenotypic male points towards some unusual varieties of CAH. ** High

level of serum testosterone, adrenal androgen, plasma ACTH and low basal cortisol are proof of CAH, whereas elevated level of 17-OH pregnenolone is biochemical marker of 3beta-HSD enzyme deficiency. ** Final diagnosis can be obtained with sequencing of HSD3B2 gene showing various mutations. ** Presence of bilateral cryptorchidism in such a patient may be due to underlying hypogonadism. ** Karyotyping in such patient may rarely show mosaic KS (47,XXY/46,XX) and there might be unmasking of hypergonadotropic hypogonadism resulting from adrenal androgen suppression from glucocorticoid treatment.

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Authors Full Name

Shehab, M A; Mahmood, Tahseen; Hasanat, M A; Fariduddin, Md; Ahsan, Nazmul; Hossain, Mohammad Shahnoor; Hossain, Md Shahdat; Jahan, Sharmin.

Institution

Shehab, M A. Department of Endocrinology, BSMMU, Dhaka, Bangladesh. Mahmood, Tahseen.

Department of Endocrinology, BSMMU, Dhaka, Bangladesh.

Hasanat, M A. Department of Endocrinology, BSMMU, Dhaka, Bangladesh.

Fariduddin, Md. Department of Endocrinology, BSMMU, Dhaka, Bangladesh.

Ahsan, Nazmul. Department of Genetic Engineering & Biotechnology, University of Dhaka, Dhaka, Bangladesh.

Hossain, Mohammad Shahnoor. Department of Genetic Engineering & Biotechnology, University of Dhaka, Dhaka, Bangladesh.

Hossain, Md Shahdat. Department of Genetic Engineering & Biotechnology, University of Dhaka, Dhaka, Bangladesh.

Jahan, Sharmin. Department of Endocrinology, BSMMU, Dhaka, Bangladesh.

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312.

Testicular microlithiasis in a boy with X-linked adrenal hypoplasia congenita.

Serbis A; Tsinopoulou VR; Mouzaki K; Kotanidou EP; Giza S; Galli-Tsinopoulou A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Annals of Pediatric Endocrinology & Metabolism. 23(3):162-165, 2018 Sep.

[Journal Article]

UI: 30286574

X-linked adrenal hypoplasia congenita (AHC) is a rare disorder that usually presents clinically as adrenal insufficiency in early infancy. It is caused by mutations in the NR0B1 gene which is located on the short arm of chromosome X (Xp21). The NR0B1 gene plays an important role in normal development and function of both the adrenal and gonadal axes and some patients with the disease can present in adolescence with hypogonadotropic hypogonadism. Testicular microlithiasis is an ultrasonographic finding of unknown etiology that has been associated with several benign conditions such as cryptorchidism, congenital adrenal hyperplasia, varicoceles, and testicular malignancy. We report the case of an 11-year-old boy who was diagnosed at the age of 8 months with X-linked AHC due to adrenal failure and presented testicular microlithiasis during follow-up. To the best of our knowledge, this is the first case of an X-linked AHC patient diagnosed with testicular microlithiasis in follow-up.

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1

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Authors Full Name

Serbis, Anastasios; Tsinopoulou, Vassiliki Regina; Mouzaki, Konstantina; Kotanidou, Eleni P; Giza, Styliani; Galli-Tsinopoulou, Assimina.

Institution

Serbis, Anastasios. 4th Department of Pediatrics, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Papageorgiou General Hospital, Thessaloniki, Greece.

Tsinopoulou, Vassiliki Regina. 4th Department of Pediatrics, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Papageorgiou General Hospital, Thessaloniki, Greece.

Mouzaki, Konstantina. 4th Department of Pediatrics, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Papageorgiou General Hospital, Thessaloniki, Greece.

Kotanidou, Eleni P. 4th Department of Pediatrics, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Papageorgiou General Hospital, Thessaloniki, Greece.

Giza, Styliani. 4th Department of Pediatrics, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Papageorgiou General Hospital, Thessaloniki, Greece.

Galli-Tsinopoulou, Assimina. 4th Department of Pediatrics, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Papageorgiou General Hospital, Thessaloniki, Greece.

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313.

Systematic review and meta-analysis comparing outcomes following orchidopexy for cryptorchidism before or after 1 year of age. [Review]

Allin BSR; Dumann E; Fawkner-Corbett D; Kwok C; Skerritt C; Paediatric Surgery Trainees Research Network.

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Bjs Open. 2(1):1-12, 2018 Feb.

[Journal Article. Review]

UI: 29951624

Background: Current guidelines recommend orchidopexy for cryptorchidism by 12 months of age, yet this is not universally adhered to. The aim of this systematic review and meta-analysis was to compare outcomes between orchidopexies performed before and after 1 year of age.

Methods: MEDLINE and Embase were searched (September 2015) using terms relating to cryptorchidism, orchidopexy and the outcomes of interest. Studies were eligible for inclusion if they compared orchidopexy at less than 1 year of age (early) with orchidopexy at 1 year or more of age (delayed) and reported the primary outcome (testicular atrophy) or one of the secondary outcomes (fertility potential, postoperative complication, malignancy). Studies were excluded when more than 50 per cent of infants had intra-abdominal testes, or the population included infants with disorders of sexual differentiation. Additional studies were identified through

reference list searching. Unpublished data were sought from the ORCHESTRA study investigators.

Results: Fifteen eligible studies were identified from 1387 titles. There was no difference in atrophy rate between early orchidopexy and delayed orchidopexy (risk ratio 0.64, 95 per cent c.i. 0.25 to 1.66; 912 testes). Testicular volume was greater (mean difference 0.06 (95 per cent c.i. 0.01 to 0.10) ml; 346 testes) and there were more spermatogonia per tubule (mean difference 0.47 (0.31 to 0.64); 382 testes) in infants undergoing early orchidopexy, with no difference in complication rate (risk ratio 0.68, 0.27 to 1.68; 426 testes). No study reported malignancy rate. Conclusion: Atrophy and complication rates do not appear different between early and delayed orchidopexy, and fertility potential may be better with early orchidopexy. Imprecision of the available data limits the robustness of these conclusions.

Version ID

1

Status

PubMed-not-MEDLINE

Author NameID

Dumann, E; ORCID: <https://orcid.org/0000-0001-7378-2072>

Authors Full Name

Allin, B S R; Dumann, E; Fawkner-Corbett, D; Kwok, C; Skerritt, C; Paediatric Surgery Trainees Research Network.

Institution

Allin, B S R. National Perinatal Epidemiology Unit University of Oxford Oxford UK. Allin, B S R. Oxford Children's Hospital Oxford UK.

Dumann, E. Medical Sciences Division University of Oxford Oxford UK.

Fawkner-Corbett, D. Oxford Children's Hospital Oxford UK.

Kwok, C. Oxford Children's Hospital Oxford UK.

Skerritt, C. Evelina Children's Hospital London UK.

PMC Identifier

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Year of Publication

2018

Management of the patients with persistent Mullerian duct syndrome: Is the ultimate goal testicular descent?.

Sancar S; Ozcakil E; Kaya M.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Turkish Journal of Urology. 44(2):166-171, 2018 Mar.

[Journal Article]

UI: 29511588

Objective: Persistent Mullerian duct syndrome (PMDS) is a rare congenital disease characterized by the presence of rudimentary Mullerian structures within an intra-abdominal or hernial sac in a virilized male, often presenting as undescended testes. In this study, we aim to present a series of the PMDS patients who were managed by orchiopexy without removal of Mullerian remnants (MR).

Material and methods: Between May 2010 and June 2017, we treated six cases diagnosed as PMDS in our department. Laparoscopy and gonadal biopsy were performed in all patients, and vessel ligation was done in four patients for the first session of Stephen-Fowler orchiopexy. After initial diagnosis, genetic analyses and endocrine investigations were performed. Demographic and clinical features of the patients, operative methods and follow-up data were analyzed retrospectively.

Results: Mean age of the patients was 5.5 years. Three boys had undergone inguinal surgery due to hernia or undescended testis, while others were diagnosed during laparoscopic investigation of nonpalpable testis. As a definitive operation, testes and MR were completely removed in an adult patient, and the remaining patients were treated with laparoscopic or open orchiopexy with or without utero-cervical splitting and the MRs were left in situ. Two testes atrophied during follow-up period.

Conclusion: The goal of the approach in PMDS patients is to preserve testes, as well as carry them to their natural location. Leaving the MR in place is a suitable option for blood circulation of the testes but the long-term results are still unknown.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Sancar, Serpil; Ozcakil, Esra; Kaya, Mete.

Institution

Sancar, Serpil. Department of Pediatric Surgery, University of Health Sciences, Yuksek Ihtisas Training and Research Hospital, Bursa, Turkey. Ozcakir, Esra. Department of Pediatric Surgery, University of Health Sciences, Yuksek Ihtisas Training and Research Hospital, Bursa, Turkey. Kaya, Mete. Department of Pediatric Surgery, University of Health Sciences, Yuksek Ihtisas Training and Research Hospital, Bursa, Turkey.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5832380>

Year of Publication

2018

315.

Curative GnRHa treatment has an unexpected repressive effect on Sertoli cell specific genes.

Gegenschatz-Schmid K; Verkauskas G; Demougin P; Bilius V; Dasevicius D; Stadler MB; Hadziselimovic F.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Basic & Clinical Andrology. 28:2, 2018.

[Journal Article]

UI: 29456864

Background: Follicle stimulating hormone and testosterone stimulate Sertoli cells to support germ cell function and differentiation. During mini-puberty, when gonadotropin (GnRH) stimulates increases in plasma luteinizing hormone (LH) and testosterone levels, gonocytes are transformed into Ad spermatogonia. In cryptorchidism, impaired gonadotropin secretion during mini-puberty results in insufficient LH and testosterone secretion, impaired gonocyte transition to Ad spermatogonia, and perturbed Sertoli cell proliferation. Treatment with a gonadotropin-releasing hormone agonist (GnRHa/Buserelin) induced gonocytes to differentiate into Ad spermatogonia and rescued fertility. The present study evaluated the impact of low LH secretion on Sertoli cell function by comparing differential gene expression data between testes with low LH that lacked Ad spermatogonia (Ad-) and testes that completed mini-puberty (Ad+). Furthermore, we analyzed changes in the transcription of selected Sertoli cell specific genes in response to GnRHa treatment.

Results: Ad- testes showed reduced expression of nine out of 40 selected Sertoli cell specific genes compared to Ad+ testes. GnRHa treatment repressed most of the Sertoli cell specific genes, including the inhibins, but it increased the expression of genes that regulate apoptosis (FASLG) and proliferation (GDNF).

Conclusions: Impaired-minipuberty with decreased LH and testosterone levels affected Ad and Sertoli cell development through positive and negative regulation of morphoregulatory and apoptotic genes. GnRHa treatment had a repressive effect on most Sertoli cell specific genes, which suggested that Sertoli cells underwent a cellular rearrangement. We propose that gonadotropin-dependent increases in FASLG and GDNF expression drove Sertoli cell proliferation and germ cell self-renewal and supported the transition of gonocytes to Ad spermatogonia, independent of inhibins.

Version ID

1

Status

PubMed-not-MEDLINE

Author NameID

Verkauskas, Gilvydas; ISNI: 0000 0001 2243 2806 Verkauskas, Gilvydas; GRID: 0000 0001 2243 2806

Demougin, Philippe; ISNI: 0000 0004 1937 0642

Demougin, Philippe; GRID: 0000 0004 1937 0642

Bilius, Vytautas; ISNI: 0000 0001 2243 2806

Bilius, Vytautas; GRID: 0000 0001 2243 2806

Dasevicius, Darius; ISNI: 0000 0004 0567 3159

Dasevicius, Darius; GRID: 0000 0004 0567 3159

Stadler, Michael B; ISNI: 0000 0001 2110 3787

Stadler, Michael B; GRID: 0000 0001 2110 3787

Stadler, Michael B; ISNI: 0000 0001 2223 3006

Stadler, Michael B; GRID: 0000 0001 2223 3006

Authors Full Name

Gegenschatz-Schmid, Katharina; Verkauskas, Gilvydas; Demougin, Philippe; Bilius, Vytautas; Dasevicius, Darius; Stadler, Michael B; Hadziselimovic, Faruk.

Institution

Gegenschatz-Schmid, Katharina. Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, 4410 Liestal, Switzerland. Verkauskas, Gilvydas. 2Children's Surgery Centre, Faculty of Medicine, Vilnius University, 01513 Vilnius, Lithuania.

Demougin, Philippe. 3Biozentrum, Life Sciences Training Facility, University of Basel, 4001 Basel, Switzerland.

Bilius, Vytautas. 2Children's Surgery Centre, Faculty of Medicine, Vilnius University, 01513 Vilnius, Lithuania.

Dasevicius, Darius. 4Institute for Pathology, National Centre of Pathology, Affiliate of Vilnius University Hospital Santariskiu Klinikos, 08406 Vilnius, Lithuania.

Stadler, Michael B. 5Friedrich Miescher Institute for Biomedical Research, 4058 Basel, Switzerland.

Stadler, Michael B. 6Swiss Institute of Bioinformatics, Basel, Switzerland.

Hadziselimovic, Faruk. Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, 4410 Liestal, Switzerland.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5806254>

Other Abstract

Publisher: L'hormone folliculostimulante et la testostérone stimulent les cellules de Sertoli pour soutenir la fonction et la différenciation des cellules germinales. Pendant la minipuberté, lorsque la gonadotrophine (GnRH) stimule les augmentations des taux plasmatiques d'hormone lutéinisante (LH) et de testostérone, les gonocytes sont transformés en spermatogonies Ad. Dans la cryptorchidie, une sécrétion altérée de gonadotrophine lors de la minipuberté entraîne une sécrétion insuffisante de LH et de testostérone, une altération de la transition des gonocytes en spermatogonies Ad, et une perturbation de la prolifération des cellules de Sertoli. Un traitement par agoniste de la gonadolibérine (GnRHa/Buserelin) induit une différenciation des gonocytes en spermatogonies Ad et sauvegarde la fertilité. Cette étude a évalué l'impact d'un taux de LH bas sur la fonction des cellules de Sertoli en comparant les données d'expression différentielle de gènes entre des testicules avec LH basse qui sont dépourvus de spermatogonies Ad (Ad-) et des testicules qui ont fini la minipuberté (Ad+). En outre, la réponse au traitement par GnRHa a été analysée par les modifications de la transcription de gènes sélectionnés pour être spécifiques de la cellule de Sertoli.; Language: French

Year of Publication

2018

316.

Pediatric Surgery remains the only true General Surgery.

Tovar J.A.

Embase

Porto Biomedical Journal. (no pagination), 2017. Date of Publication: 2017.

[Article In Press]

AN: 618003335

This article states that Pediatric Surgery remains probably the only remaining General Surgery because it is not about organs and systems but rather the whole Surgery from fetal life until completion of growth and maturation. Pediatric surgeons are currently involved in prenatal treatments for fetal diseases, they take in charge the surgery of congenital malformations, acquired neonatal diseases, common conditions like hernias, undescended testes and appendicitis, but also of the more complex gastrointestinal, broncho-pulmonary or genitourinary conditions, tumors, trauma and solid organ transplantation. For this, like other surgical specialists, they use open, endoscopic and minimally invasive techniques. The broad spectrum of diseases, many of them scarcely prevalent, makes training long and hard, but this challenge accounts for the greatness of this specialty. Pediatric surgeons also carry out research work in their field because they are aware that understanding of why the conditions treated by them occur is mandatory. In summary, Pediatric Surgery is a lively, exciting, difficult specialty that offers an attractive alternative to young doctors interested in surgery.

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Status

Article-In-Press

Institution

(Tovar) Emeritus Professor of Pediatrics, Universidad Autonoma de Madrid, Hospital Universitario La Paz, Madrid, Spain

Publisher

Elsevier Espana S.L.

Year of Publication

2017

317.

Male hypogonadism resulting from mutations in the genes for the gonadotropin subunits and their receptors.

Huhtaniemi I.T.

Embase

Contemporary Endocrinology. (pp 127-152), 2017. Date of Publication: 2017.

[Chapter]

AN: 627405078

Mutations in the genes of gonadotropin subunits (CGA, LHB, FSHB, and CGB) and receptors (LHCGR and FSHR) are extremely rare causes of male hypogonadism. No germ line mutations of CGA have been reported, apparently because of the incompatibility of pregnancy maintenance in the absence of hGG. Five inactivating LHB mutations have been described in men with normal prenatal masculinization but arrested pubertal development. The three men so far described with FSHB mutation were azoospermic. Constitutively activating mutations of the LHCGR gene give rise to very early onset familial male-limited precocious puberty (FMPP) also termed testotoxicosis. Inactivating LHCGR mutation results in an array of male phenotypes ranging from micropenis and hypospadias to complete sex reversal (XY, disorder of sexual development), depending on the completeness of receptor inactivation. Inactivating FSHR mutations in men cause a decrease in testicular size and suppressed quality and quantity of spermatogenesis but not azoospermia, and some affected men may be fertile. Only two cases of activating FSHR mutations have been detected, and they suggest that the mutation does not have phenotype in men with otherwise normal endocrine function. The discrepancy between the phenotypes of men with inactivating FSHB (azoospermia) and FSHR (no azoospermia) mutations must be clarified with additional subjects. Information about the genotypic effects of common polymorphisms in gonadotropin and gonadotropin receptor genes is gradually mounting. A common polymorphism in LHB affects bioactivity of the hormone and has multiple mild phenotypic effects, including slow tempo of puberty in boys and is enriched in post-term boys with cryptorchidism. Some FSHB and FSHR polymorphisms have been shown to affect spermatogenesis and the response of oligozoospermic men to FSH therapy. Such polymorphisms may represent important targets for the pharmacogenetic evaluation of gonadotropin treatment in infertility.

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Status

Embase

Institution

(Huhtaniemi) Institute of Reproductive and Developmental Biology, Department of Surgery and Cancer, Imperial College, London W12 0NN, United Kingdom

Publisher

Humana Press Inc. (E-mail: humana@humanapr.com)

Year of Publication

2017

318.

The undescended testis.

Lee P.A., Houk C.P., Toppari J.

Embase

Contemporary Endocrinology. (pp 197-217), 2017. Date of Publication: 2017.

[Chapter]

AN: 627402698

Cryptorchidism, the condition in which one or both testes are not descended fully into the scrotum, is one of the most frequent developmental anomalies of the human male. Although there is spontaneous descent in the first few months of life in many males born with this condition, the prevalence thereafter is about 1%. The genetic direction of testicular descent is only partially understood. There are multiple etiologies of cryptorchidism, both anatomic and hormonal, with many testes having the potential for normal function. Germ cells in the undescended testis fail to undergo normal differentiation from early infancy, hence the recommendation for treatment between 6 and 12A months of age. Men with undescended testes frequently have impaired sperm production and decreased inhibin B and elevated FSH levels. Only a small portion of men who had unilateral cryptorchidism are infertile based upon paternity; however, in contrast nearly half of men with previous bilateral cryptorchidism are infertile. The risk of developing a testicular tumor, primarily of germ cell origin, is increased substantially, particularly among the bilateral group and those not corrected before puberty.

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Status

Embase

Institution

(Lee) The Milton S. Hershey Medical Center, 500 University Drive, P.O. Box 850, Hershey, PA

17033-0850, United States (Houk) Department of Pediatrics, Medical College of Georgia,

Georgia Rege University, Augusta, GA, United States

(Toppari) Department of Physiology, Institute of Biomedicine, University of Turku, Department of

Pediatrics, Turku University Hospital, Turku, Finland

Publisher

Humana Press Inc. (E-mail: humana@humanapr.com)

Year of Publication

2017

319.

The Nonpalpable Testis: A Narrative Review.

Shepard C.L., Kraft K.H.

Embase

Journal of Urology. (pp 1410-1417), 2017. Date of Publication: 2017.

[Article In Press]

AN: 618847816

Purpose: While the nonpalpable testis represents a small portion of all cryptorchid testes, it remains a clinical challenge for pediatric urologists. Controversy exists surrounding the best evaluation and management of this entity. In this review we update what is known about the nonpalpable testis, including the etiology, preoperative evaluation and best surgical management as well as novel techniques and ongoing controversies.

Material(s) and Method(s): We searched PubMed and MEDLINE from January 2000 to January 2017 using relevant key terms. Of 367 articles 115 were considered for inclusion based on a priori design. Using a narrative review format, an update on the evaluation and management of the nonpalpable testis including novel concepts and techniques was synthesized.

Result(s): The nonpalpable testis should be evaluated by physical examination only. Imaging is not indicated for routine cases. The optimal surgical approach and technique remain debatable but several novel techniques have been described. Due to the rarity of the nonpalpable testis, randomized controlled trials and other quality comparisons are difficult. Therefore, management remains controversial.

Conclusion(s): Evaluation and management of the nonpalpable testis remain difficult, and some aspects are still debated. Future research should focus on multi-institutional collaborative trials to determine the optimal operative management.

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PMC Identifier

28434984 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28434984>]

Status

Article-in-Press

Institution

(Shepard, Kraft) Department of Urology, University of Michigan, Ann Arbor, Michigan, United States

Publisher

Elsevier Inc. (E-mail: usjcs@elsevier.com)

Year of Publication

2017

320.

Abnormalities of the external genitalia and groins among primary school boys in Bida, Nigeria. Adekanye A.O., Adefemi S.A., Onawola K.A., James J.A., Adeleke I.T., Francis M., Sheshi E.U., Atakere M.E., Jibril A.D.

Embase

African Health Sciences. 17 (4) (pp 1120-1125), 2017. Date of Publication: 2017.

[Article]

AN: 620208280

Background: Abnormalities of the male external genitalia and groin, a set of lesions which may be congenital or acquired, are rather obscured to many kids and their parents and Nigerian health care system has no formal program to detect them.

Objective(s): To identify and determine the prevalence of abnormalities of external genitalia and groin among primary school boys in Bida, Nigeria.

Method(s): This was a cross-sectional study of primary school male pupils in Bida. A detailed clinical examination of the external genitalia and groin was performed on them.

Result(s): Abnormalities were detected in 240 (36.20%) of the 663 boys, with 35 (5.28%) having more than one abnormality. The three most prevalent abnormalities were penile chordee (37, 5.58%), excessive removal of penile skin (37, 5.58%) and retractile testis (34, 5.13%). The prevalence of complications of circumcision was 15.40% and included excessive residual foreskin, excessive removal of skin, skin bridges and meatal stenosis. Undescended testes were seen in 6 (0.90%) boys, with median age of 9 years and 2 were bilateral. Also, micropenis was detected in 27 (4.07%) of the pupils.

Conclusion(s): Inguino-penoscrotal abnormalities are common in our community (36.20%).

Screening of pre-school and school children to detect them should be introduced into the school health programs in Nigeria.

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PMC Identifier

29937883 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=29937883>]

Status

Embase

Institution

(Adekanye, Adefemi, Onawola, James, Adeleke, Francis, Sheshi, Atakere, Jibril) Centre for Health & Allied Researches (CHAR), Federal Medical Centre, Bida, Nigeria (Adekanye, Onawola, James, Francis) Department of Surgery, Federal Medical centre, Bida, Nigeria (Adefemi, Sheshi) Department of Family Medicine, Federal Medical centre, Bida, Nigeria (Adeleke) Department of Health Information management, Federal Medical centre, Bida, Nigeria (Atakere, Jibril) Department of Obstetrics & Gynaecology, Federal Medical centre, Bida, Nigeria

Publisher

Makerere University, Medical School (E-mail: pic@infocom.co.ug)

Year of Publication

2017

321.

Identification of a novel mutation of NR0B1 in a patient with X-linked adrenal hypoplasia and symptomatic treatment.

Yang J., Lv Y., Zhou Y., Xiao X.

Embase

Journal of Pediatric Endocrinology and Metabolism. 30 (12) (pp 1299-1304), 2017. Date of Publication: 27 Nov 2017.

[Article]

AN: 619571884

X-linked congenital adrenal hypoplasia (X-linked AHC) is characterized by acute onset of primary adrenal insufficiency in infancy or early childhood and hypogonadotropic hypogonadism (HH) at puberty. Mutations in NR0B1, the gene located on Xp21.3 and encoding an orphan nuclear receptor named DAX1, are responsible for this disease. The entire coding region of the NR0B1 gene of a 14-year-old X-linked AHC proband as well as his family members was sequenced. Clinical and endocrine evaluations with symptomatic treatment results were recorded. DNA

sequencing revealed a missense mutation (c.383-384 insA) in exon 1, which resulted in a novel frameshift mutation, thereby resulting in a truncated protein (p.Leu129 Pro fs*137). The therapeutic trial with an observation period of 20 weeks showed an effective improvement in symptoms of hypogonadism with human chorionic gonadotropin (HCG) administration, including a rapid improvement of serum testosterone level, descending of testicles as well as enlargement of testicles and growth of penis. Our study identified a novel frameshift mutation of the NR0B1 gene in a proband with X-linked AHC/HH and further expanded the number of NR0B1 mutations reported in the literature. Moreover, the symptomatic treatment observation provided referential evidence in the treatment of X-linked AHC associated hypogonadism and bilateral inguinal cryptorchidism.

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PMC Identifier

29176027 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=29176027>]

Status

Embase

Institution

(Yang, Zhou, Xiao) Department of Metabolism and Endocrinology, First Affiliated Hospital, University of South China, 69 Chuanshan Road, Hengyang 421001, China (Lv) Laboratory of Clinical Anatomy, University of South China, Hengyang, China

Publisher

Walter de Gruyter GmbH

Year of Publication

2017

322.

Testicular volume and elasticity changes in young children with undescended testes.

Shin H.J., Lee Y.S., Yoon H., Kim M.-J., Han S.W., Kim H.Y., Lee J., Lee M.-J.

Embase

Medical Ultrasonography. 19 (4) (pp 380-385), 2017. Date of Publication: 2017.

[Article]

AN: 619495361

Aims: To evaluate the differences and changes of testicular volume and elasticity in normal (NL) testes and undescended testes (UDTs) of children using shear wave elastography (SWE).

Material(s) and Method(s): Testicular ultrasound images from children younger than 60 months old were retrospectively reviewed. Testicular volumes and elasticities were compared between the UDT group and NL group. In patients with unilateral UDT (uUDT), we also compared the values between uUDT and contralateral grossly normal (CGN) testis groups.

Result(s): There were 25 UDTs including 4 bilateral in the UDT group and 54 normal testes in the NL group. While testicular volume was significantly smaller in UDT (vs. NL) and uUDT (vs. CGN) groups, the elasticity was not different. Testicular volume was positively correlated with age in both NL ($r=0.474$) and CGN ($r=0.729$) groups ($p < 0.001$), while there was no correlation in the UDT group. Testicular stiffness showed negative correlation with age in the NL group ($r=-0.390$, $p=0.004$) and positive correlation in the UDT group ($r=0.426$, $p=0.034$).

Conclusion(s): Instead of increasing volume and decreasing stiffness of normal testes during development of the early 60 months, UDTs exhibited smaller volume and increasing stiffness. The CGN testes of uUDT patients showed increasing volume without stiffness change.

PMC Identifier

29197914 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=29197914>]

Status

Embase

Institution

(Shin, Yoon, Kim, Lee) Department of Radiology and Research Institute of Radiological Science, Severance Children's Hospital, Yonsei University College of Medicine, Seoul, South Korea (Lee, Han) Department of Urology and Urological Science Institute, Severance Children's Hospital, Yonsei University College of Medicine, Seoul, South Korea

(Kim, Lee) Biostatistics Collaboration Unit, Yonsei University College of Medicine, Seoul, South Korea

Publisher

Societatea Romana de Ultrasonografie in Medicina si Biologie (E-mail: dfodor@ymail.com)

Year of Publication

2017

Involvement of the bone morphogenic protein/SMAD signaling pathway in the etiology of congenital anomalies of the kidney and urinary tract accompanied by cryptorchidism.

Mizuno K., Nakane A., Nishio H., Moritoki Y., Kamisawa H., Kurokawa S., Kato T., Ando R., Maruyama T., Yasui T., Hayashi Y.

Embase

BMC Urology. 17 (1) (no pagination), 2017. Article Number: 112. Date of Publication: 02 Dec 2017.

[Article]

AN: 619479116

Background: Congenital anomalies of the kidney and urinary tract (CAKUT), such as renal dysplasia, hydronephrosis, or vesicoureteral reflux, are the most common causes of end-stage renal disease. However, the genetic etiology of CAKUT remains unclear. In this study, we performed whole exome sequencing (WES) to elucidate the genetic etiology of symptomatic CAKUT and CAKUT accompanied by cryptorchidism.

Method(s): Three patients with unilateral renal dysplasia accompanied by ipsilateral cryptorchidism were included in this analysis. Genomic DNA was extracted from peripheral blood, and WES was performed. Disease-specific single nucleotide polymorphisms (SNPs) were determined by comparison with the human genome reference sequence (hg19). Additionally, we searched for SNPs that were common to all three patients, with a particular focus on the coding regions of the target genes.

Result(s): In total, 8710 SNPs were detected. Of the genes harboring these SNPs, 32 associated with renal or testicular development were selected for further analyses. Of these, eight genes (i.e., SMAD4, ITGA8, GRIP1, FREM1, FREM2, TNXB, BMP8B, and SALL1) carried a single amino acid substitution that was common to all three patients. In particular, SNPs in SMAD4 (His290Pro and His291Pro) have not been reported previously in patients with symptomatic CAKUT. Of the candidate genes, four genes (i.e., ITGA8, GRIP1, FREM1, and FREM2) were Fraser syndrome-related genes, encoding proteins that functionally converged on the glial cell-derived neurotrophic factor/RET/bone morphogenic protein (BMP) signaling pathways. As another candidate gene, the protein encoded by BMP8B activates the nuclear translocation of SMAD4, which regulates the expression of genes associated with the differentiation of primordial germ cells or testicular development. Additionally, BMP4, a member of the BMP family, regulates the interaction between metanephric mesenchyme and ureteric buds by suppressing GDNF.

Conclusion(s): Taken together, our findings suggested that the development of the kidney and urinary tract is intimately linked with that of male reproductive organs via BMP/SMAD signaling pathways.

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PMC Identifier

29197384 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=29197384>]

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Embase

Institution

(Mizuno, Nakane, Nishio, Moritoki, Kamisawa, Kurokawa, Kato, Ando, Maruyama, Yasui)
Department of Nephro-urology, Nagoya City University Graduate, School of Medical Sciences,
Nagoya, Japan (Hayashi) Department of Pediatric Urology, Nagoya City University Graduate,
School of Medical Sciences, 1 Kawasumi, Mizuho-cho, Mizuho-ku, Nagoya, Japan

Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2017

324.

Undescended testis: What paediatricians need to know.

Hutson J.M., Vikraman J., Li R., Thorup J.

Embase

Journal of Paediatrics and Child Health. 53 (11) (pp 1101-1104), 2017. Date of Publication:
November 2017.

[Review]

AN: 619353526

Undescended testis (UDT) occurs when something goes wrong with testicular descent from high in the abdominal cavity to the scrotum. Normal descent occurs in two steps, with the transabdominal phase controlled by a new testicular hormone, insulin-like hormone 3, and the inguinoscrotal phase controlled by androgens. The latter phase requires a complex process of migration from the inguinal abdominal wall to the scrotum and is commonly defective, leading to the high incidence (2-4%) of UDT at birth. The clinical examination of babies and infants aims to confirm the persistence of congenital UDT by 3-6 months, so surgery can be optimally timed at 6-12 months. For those boys who develop acquired UDT later in childhood, the 'ascending' testis often needs surgery between 5 years and 10 years, so all boys should be screened again for UDT at school entry.

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Status

Embase

Author NameID

Hutson, John M.; ORCID: <http://orcid.org/0000-0002-3922-8366>

Institution

(Hutson) Urology Department, Royal Children's Hospital, Melbourne, VIC, Australia (Hutson, Vikraman, Li) F Douglas Stephens Surgical Research Group, Murdoch Children's Research Group, Melbourne, VIC, Australia

(Hutson, Vikraman) Department of Paediatrics, University of Melbourne, Melbourne, VIC, Australia

(Thorup) Department of Pediatric Surgery, Rigshospitalet, Copenhagen, Denmark

(Thorup) Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

Publisher

Blackwell Publishing

Year of Publication

2017

325.

Circumcision-incision orchidopexy: A novel technique for palpable, low inguinal undescended testis.

Chua M.E., Silangcruz J.M.A., Gomez O., Dy J.S., Morales M.L.

Embase

Investigative and Clinical Urology. 58 (6) (pp 468-473), 2017. Date of Publication: November 2017.

[Article]

AN: 619122606

Given that both orchidopexy and circumcision are commonly done in a single operative setting, we adopted a technique of combined orchidopexy and circumcision using a single circumcision incision. We applied this new technique to boys with palpable, low inguinal cryptorchidism. Here we describe a case series of 7 boys who underwent concurrent orchidopexy via the circumcision site. We present this novel technique and discuss our preliminary outcomes, including the anatomic basis and feasibility. The technique appears to be an alternative for concurrent circumcision and cryptorchid cases with palpable, low inguinal testes.

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Status

Embase

Institution

(Chua, Silangcruz, Dy, Morales) Institute of Urology, St. Luke's Medical Center, Quezon City, Philippines (Gomez) Institute of Radiology, Section of Pediatric Radiology, St. Luke's Medical Center, Global City, Philippines

(Morales) Department of Urology, National Kidney and Transplant Institute, Quezon City, Philippines

Publisher

Korean Urological Association (E-mail: uro-edit@urology.or.kr)

Year of Publication

2017

326.

Paternal age and risk of testicular germ cell tumors: a cohort study of 1,000,000 men.

Levine H., Keinan-Boker L., Leiba A., Derazne E., Rais A., Kark J.D.

Embase

Andrology. 5 (6) (pp 1124-1130), 2017. Date of Publication: November 2017.

[Article]

AN: 619085843

Testicular germ cell tumors (TGCT) are the most frequent cancer among young men, with increasing incidence worldwide. Advanced paternal age has been linked to adverse health

outcomes in offspring, but reports on the association of paternal age with TGCT are few and inconsistent. We aimed to examine the relationship of paternal age (PAB) at birth with the risk of TGCT and by histologic type: seminoma and non-seminoma. A population-based cohort of 1,056,058 males, examined at ages 16-19 between the years 1980-2011, was linked to the Israel National Cancer Registry to obtain incident TGCT through 2012. We applied multivariable Cox regression. During 16.5 million person-years of follow-up, 1247 incident cases (604 seminomas and 643 non-seminomas) were detected. Increasing PAB was linearly associated with lower risk of TGCT (HRper year = 0.983, 95% CI: 0.974-0.993, p = 0.001), after adjustment for year of birth, years of education, height, cryptorchidism history and origin, and also with additional adjustment for maternal age at birth (MAB) (HRper year = 0.980: 0.965-0.995, p = 0.008). The association was stronger for seminoma (HRper year = 0.968: 0.946-0.989, p = 0.004) and persisted in a subset adjusted for sibship size (HRper year = 0.950: 0.917-0.983, p = 0.003). In the fully adjusted model, young PAB (15-24 vs. >=30) was a risk factor for seminoma (HR = 1.41: 1.07-1.85, p = 0.014). In models adjusted for PAB, MAB was not associated with risk of TGCT. In conclusion, our findings suggest that young paternal age is a risk factor of TGCT, especially seminoma. The findings warrant further investigation into the possible impact of young paternal age on their offsprings' testes.

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Author NameID

Levine H.; ORCID: <http://orcid.org/0000-0002-5597-4916>

Institution

(Levine, Kark) Braun School of Public Health and Community Medicine, Hebrew University-Hadassah, Jerusalem, Israel (Keinan-Boker) The National Cancer Registry, Israel Center for Disease Control, Jerusalem, Israel

(Keinan-Boker) School of Public Health, University of Haifa, Haifa, Israel

(Leiba, Derazne, Rais) The Israel Defense Forces Medical Corps, Tel Hashomer, Israel

(Leiba, Derazne) Sackler Faculty of Medicine, Tel-Aviv University, Tel Aviv, Israel

(Leiba) Institute of Nephrology and Hypertension, Sheba Medical Center, Tel Hashomer, Israel

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2017

327.

Frequency of revision orchidopexy in Australia 1995-2014.

Vikraman J., Vidmar S., Donath S., Hutson J.M.

Embase

Journal of Pediatric Surgery. 52 (12) (pp 1940-1943), 2017. Date of Publication: December 2017.

[Article]

AN: 618525322

Background/aim International criteria currently suggest orchidopexy at 6-12 months for congenital undescended testis (UDT). Some children require repeat orchidopexy for recurrent UDT. This study aimed to assess practice in Australia over a 20-year period. Methods We examined 20 years of Australian orchidopexy data (1995-2014) from the Department of Human Services to explore the national revision orchidopexy rates over time. Results The total number of orchidopexy revisions was 890 over 20 years compared with 25,984 primary operations. More than 50% of all primary and revision orchidopexies in 0-14 year-old boys were performed in major population centers of NSW and Victoria (which hold 52% male population of same age), with a small number of revisions on 15-24 year-old males. The incidence of revision orchidopexy significantly decreased over the 20-year period in boys ages 0-14 years old, from 276 operations between 1995 and 1999 decreasing to 165 operations between 2010 and 2014 (- 53%), compared to a population increase of + 15% ($p < 0.05$). Conclusion These data demonstrate a decrease in revision orchidopexy since 1995, which may be related to change in referral practice with more children undergoing orchidopexy (primary and revision) by pediatric surgeons over the 20-year period. Level of evidence Level IV. Type of Study Therapeutic Case Series with no Comparison Group.

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Embase

Institution

(Vikraman, Hutson) Douglas Stephens Surgical Research Laboratory, Murdoch Childrens Research Institute, Melbourne, Australia (Vikraman, Hutson) Royal Children's Hospital, Melbourne, Australia

(Vikraman, Hutson) Department of Paediatrics, University of Melbourne, Australia
(Vidmar, Donath) Clinical Epidemiology and Biostatistics, Murdoch Childrens Research Institute,
Melbourne, Australia

Publisher

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Year of Publication

2017

328.

Clinical and genetic features of 64 young male paediatric patients with congenital hypogonadotropic hypogonadism.

Wang Y., Gong C., Qin M., Liu Y., Tian Y.

Embase

Clinical Endocrinology. 87 (6) (pp 757-766), 2017. Date of Publication: December 2017.

[Article]

AN: 618284675

Context: The diagnosis of congenital hypogonadotropic hypogonadism (CHH) in prepuberty has always been challenging. Here, we aimed at studying the clinical and genetic features of paediatric CHH, especially the phenotype of hypospadias and dual defects (patients showing hypothalamic and/or pituitary defects and testicular hypoplasia), so as to have a better understanding of CHH.

Design(s): The clinical and genetic features of patients with CHH were analysed, and the relationships between hypospadias, dual defects and genetics were investigated.

Patient(s): Patients who visited Beijing Children's Hospital and were positively diagnosed with CHH. Measurements: The collected data included sex hormones, MRI of the olfactory bulb, human chorionic gonadotrophin (hCG) test and genetic testing. We analysed clinical features and genetic results, especially hypospadias and dual defects, and compared the stimulated testosterone (T) levels in patients with and without cryptorchidism.

Result(s): Sixty-four patients were positively diagnosed, and forty-seven (73.4%) had Kallmann syndrome (KS). Four patients (6.3%) had hypospadias, including 2 KS. Micropenis combined with cryptorchidism was the most common phenotype (39%). Approximately two-third of patients showed a poor response to hCG; 15 cases were diagnosed with dual defects, and there were no

significant differences between those with and without cryptorchidism. Twenty-six cases (51%) of 51 patients were identified as having classical HH mutations, affecting 10 different genes, with oligogenic mutations in 5 cases (9.8%). The most common mutations were in PROKR2 (17.6%), FGFR1 (13.7%) and CHD7 (7.8%). The frequency of PROKR2 mutations was higher in dual HH when compared to other HH cases (6/15 vs 3/36, P =.021).

Conclusion(s): Micropenis and/or cryptorchidism can serve as important signs for the diagnosis of HH in paediatrics, and the coexistence of hypospadias does not exclude the diagnosis of CHH, including KS or normosmic isolated HH (nHH). Testicular function may be impaired earlier than expected, and PROKR2 mutations need to be evaluated to identify presumed dual defects.

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Author NameID

Gong, Chunxiu; ORCID: <http://orcid.org/0000-0002-1262-7383>

Institution

(Wang, Gong, Qin, Liu, Tian) National Center for Children's Health, Capital Medical University, Beijing, China (Wang, Gong, Qin, Tian) Department of Endocrinology, Genetics, Metabolism

and Adolescent Medicine, Beijing Children's Hospital, Capital Medical University, Beijing, China

(Wang, Gong, Qin, Tian) Beijing Key Laboratory for Genetics of Birth Defects, Beijing Children's Hospital, Capital Medical University, Beijing, China

(Liu) Department of Pharmacy, Beijing Children's Hospital, Capital Medical University, Beijing, China

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2017

329.

Prevalence and Chronological Changes of Testicular Microlithiasis in Isolated Congenital Undescended Testes Operated On at Less Than 3 Years of Age.

Nishimura Y., Moriya K., Nakamura M., Nishida M., Sato M., Kudo Y., Omotehara S., Iwai T., Wakabayashi Y., Kanno Y., Kitta T., Kon M., Shinohara N.

Embase

Urology. 109 (pp 159-164), 2017. Date of Publication: November 2017.

[Article]

AN: 618160272

Objective To clarify the prevalence and chronological changes of testicular microlithiasis in isolated congenital undescended testes, retrospective chart review was performed. **Materials and Methods** Among children with palpable isolated undescended testes who underwent orchiopexy at less than 3 years of age between January 2009 and May 2016, those who had preoperative testicular ultrasonography were enrolled. Testicular microlithiasis was classified as limited or classic. **Results** Sixty-five patients (54 unilateral undescended testes and 11 bilateral undescended testes) were enrolled. Preoperative evaluation demonstrated limited testicular microlithiasis in only 2 undescended testes in 2 patients (1 with unilateral undescended testes and 1 with bilateral undescended testes). Of these patients, 1 with unilateral undescended testes had limited testicular microlithiasis and the other with bilateral undescended testes had classic testicular microlithiasis after surgery. Among 53 unilateral undescended testes without microlithiasis preoperatively, limited and classic testicular microlithiasis was found in 1 and 6 testes, respectively, during follow-up. Testicular microlithiasis was identified in 2 on the contralateral descended testis of unilateral undescended testes postoperatively. Among 10 patients with bilateral undescended testes without microlithiasis preoperatively, limited testicular microlithiasis was detected in 4 during follow-up. Testicular microlithiasis was not diminished or resolved during follow-up. The overall prevalence of testicular microlithiasis in undescended testes (21.1%) was significantly higher than that in the contralateral descended testis in patients with unilateral undescended testes (3.7%) ($P < .01$). **Conclusion** Most testicular microlithiasis was identified postoperatively and never improved. The prevalence of testicular microlithiasis in isolated congenital undescended testes increased with time even if operated on early in life.

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Author NameID

Moriya, Kimihiko; ORCID: <http://orcid.org/0000-0001-6161-9403>

Institution

(Nishimura, Moriya, Nakamura, Kanno, Kitta, Kon, Shinohara) Department of Renal and Genitourinary Surgery, Hokkaido University Graduate School of Medicine, Japan (Nishida, Sato,

Kudo, Omotehara, Iwai, Wakabayashi) Diagnostic Center for Sonography, Hokkaido University Hospital, Japan

(Nishida, Kudo, Omotehara, Iwai) Division of Laboratory and Transfusion Medicine, Hokkaido University Hospital, Japan

(Sato, Wakabayashi) Department of Radiological Technology, Hokkaido University Hospital, Japan

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Year of Publication

2017

330.

Telehealth in paediatric surgery: Accuracy of clinical decisions made by videoconference.

Brownlee G.L., Caffery L.J., McBride C.A., Patel B., Smith A.C.

Embase

Journal of Paediatrics and Child Health. 53 (12) (pp 1220-1225), 2017. Date of Publication: December 2017.

[Article]

AN: 616652723

Aim: Telehealth is a useful method of providing specialist consultation to a geographically diverse population. Canadian studies of telehealth for paediatric surgery demonstrate good accuracy, but have low numbers of cryptorchid patients in their cohorts. Our aim was to confirm Canadian studies for our cohort and to assess accuracy regarding cryptorchidism.

Method(s): We conducted a cohort study of patients seen via paediatric surgical telehealth over a 12-month period, to determine accuracy of telediagnosis with respect to face-to-face diagnosis and plan.

Result(s): A total of 183 children had 224 videoconferences, resulting in 74 surgical bookings. There was high diagnostic concordance, except for undescended testes. One discharged patient, and two patients booked for review, have subsequently required an orchidopexy (false negatives). Of 15 patients booked for surgery, three did not require an operation (false positives). Other patients had their procedures upgraded (from open to laparoscopic) or downgraded (from laparoscopic to open) due to inaccuracies in far-end assessment.

Conclusion(s): Telehealth for paediatric surgery is accurate for most conditions seen, but for cryptorchidism there are significant concerns.

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Institution

(Brownlee, Caffery, Smith) Centre for Online Health, The University of Queensland, Brisbane, QLD, Australia (McBride, Patel) Department of Paediatric Surgery, Lady Cilento Children's Hospital, Children's Health Queensland, Brisbane, QLD, Australia

Publisher

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Year of Publication

2017

331.

A comparison between totally laparoscopic hydrocelectomy and scrotal incision hydrocelectomy with laparoscopic high ligation for pediatric cord hydrocele.

Choi B.S., Byun G.Y., Hwang S.B., Koo B.H., Lee S.R.

Embase

Surgical Endoscopy. 31 (12) (pp 5159-5165), 2017. Date of Publication: 01 Dec 2017.

[Article]

AN: 616100223

Background: The purpose of this study is to report clinical characteristics and to investigate the feasibility and safety of totally laparoscopic hydrocelectomy (TLH) compared to scrotal incision hydrocelectomy with laparoscopic high ligation (SIH) for pediatric cord hydrocele (CH).

Method(s): From September 2011 to February 2016, 148 patients underwent SIH, and 342 patients underwent TLH for CH. In the TLH group, a large hydrocele that could not pass through the internal ring was removed after percutaneous syringe aspiration. Age, laterality of hydrocele, inguinal comorbidities, operation time, surgical complications, and recurrences were evaluated.

Result(s): All the patients had spermatic cord cysts and patent processus vaginalis in proximity to hydrocele (mixed type). The mean age of CH patients was 34.1 +/- 22.1 months. CHs are more common on the right side (61.0%) than on the left (35.7%). Bilaterality occurred in 3.3%. Comorbidities such as hernia (8.6%) and cryptorchidism (1.2%) were observed. There were no complications except for two cases of wound hematoma in SIH group. There was one (0.7%) case of recurrence appeared in communicating hydrocele in SIH group. There were no significant differences in the age, laterality of hydrocele, inguinal comorbidities, operation time, complications, and recurrences between TLH and SIH groups. However, TLH for unilateral cord hydrocele had significantly shorter operation time compared to SIH. The mean operation time in TLH group was 15.6 +/- 5.96 min and there was no conversion to open surgery.

Conclusion(s): TLH for pediatric CH is a feasible and safe procedure without additional incisions. Therefore, TLH can be one of the surgical options for pediatric CH especially in mixed type.

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Institution

(Choi, Byun, Hwang, Koo, Lee) Department of Surgery, Damsoyu Hospital, 213 Bongeunsa-ro, Gangnam-gu, Seoul, South Korea

Publisher

Springer New York LLC (E-mail: barbara.b.bertram@gsk.com)

Year of Publication

2017

332.

Update on the surgical approach for reconstruction of the male genitalia.

Romao R.L.P., Pippi Salle J.L.

Embase

Seminars in Perinatology. 41 (4) (pp 218-226), 2017. Date of Publication: June 2017.

[Article]

AN: 615923217

The majority of patients with DSD will be found to carry an XY karyotype and be assigned male gender. From a phenotypical standpoint, most will present with proximal hypospadias +/- cryptorchidism. In this review article, the authors present the current status of reconstruction of the male genitalia in this setting. The report addresses the following topics: surgical input in the evaluation of the newborn with an undervirilized external genitalia, including gender assignment considerations; controversies surrounding timing and indication for hypospadias surgery in proximal cases as well as use of testosterone; surgical techniques and decision-making process for one- vs. two-stage repairs; complications of hypospadias surgery based on technique used for repair; and long-term follow-up. The high complication rates observed in the treatment of proximal hypospadias attest to its challenging nature. Concentration of experience, tracking carefully identified patient-centered outcomes and long-term follow-up of this patient population are recommended.

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Institution

(Romao) IWK Health Centre, Dalhousie University, Halifax, Nova Scotia, Canada (Pippi Salle)
Division of Urology, Department of Surgery, Sidra Medical and Research Center, PO Box 26999,
Doha, Qatar

Publisher

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Year of Publication

2017

333.

Gonadotrophin abnormalities in an infant with lowe syndrome.

Warner B.E., Inward C.D., Burren C.P.

Embase

Endocrinology, Diabetes and Metabolism Case Reports. 2017 (no pagination), 2017. Article
Number: 17-0042. Date of Publication: April 2017.

[Article]

AN: 620382652

This case, presenting with bilateral impalpable testes, illustrates the relevance of a broad differential disorders of sex development case management. It provides new insights on hypothalamic-pituitary-gonadal (HPG) axis and testicular function abnormalities in the multisystem disorder of Lowe syndrome. Lowe syndrome, also known as oculocerebrorenal syndrome, is a rare disorder characterised by eye abnormalities, central nervous system involvement and proximal renal tubular acidosis. There are a handful of reports of pubertal delay, infertility and cryptorchidism in Lowe syndrome. Biochemistry aged 72 h: testosterone 6.4 nmol/L, LH <0.5 IU/L and FSH <0.5 IU/L. Gonadotropin-releasing hormone stimulation test identified significantly raised baseline LH = 45.4 IU/L (contrasts with earlier undetectable LH), with a 20% increase on stimulation, while baseline FSH = 4.3 IU/L with no increase on stimulation. Day 14 HCG stimulation test produced an acceptable 50% increase in testosterone. The constellation of further abnormalities suggested Lowe syndrome: hypotonia, bilateral cataracts (surgical extraction and intraocular lens implantation) and renal tubular acidosis (microscopic haematuria, hypercalciuria, proteinuria, generalised aminoaciduria, hypophosphataemia and metabolic acidosis). DNA sequencing identified de novo hemizygous frameshift mutation OCRL c.2409_2410delCT in exon 22. Interpretation of initial and repeat GnRH and HCG testing indicates the likelihood of testicular failure. Partial testicular descent occurred but left orchidopexy was required. Improving long-term gonadal function in Lowe syndrome assumes increased importance for current cohorts as advances in renal replacement therapy have greatly improved life expectancy. Noting HPG axis abnormalities in Lowe syndrome in infancy can identify cases requiring increased surveillance of pubertal progress for earlier detection and management. Copyright © 2017 The authors.

Status

Embase

Institution

(Warner, Burren) Department of Paediatric Endocrinology and Diabetes, University Hospitals Bristol NHS Foundation Trust, Bristol, United Kingdom (Inward) Department of Paediatric Nephrology, Bristol Royal Hospital for Children, University Hospitals Bristol NHS Foundation Trust, Bristol, United Kingdom

(Burren) NIHR Biomedical Research Unit in Nutrition, Diet & Lifestyle, University Hospitals Bristol NHS Foundation Trust, Education Centre, Bristol, United Kingdom

Publisher

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Year of Publication

2017

334.

A case-control study of maternal polybrominated diphenyl ether (PBDE) exposure and cryptorchidism in Canadian populations.

Goodyer C.G., Poon S., Aleksa K., Hou L., Atehortua V., Carnevale A., Jednak R., Emil S., Bagli D., Dave S., Hales B.F., Chevrier J.

Embase

Environmental Health Perspectives. 125 (5) (no pagination), 2017. Date of Publication: May 2017.

[Article]

AN: 619170302

Background: Polybrominated diphenyl ethers (PBDEs) are flame retardants found in North American household products during the past four decades. These chemicals leach out in dust as products age, exposing individuals daily through inhalation and ingestion. Animal studies suggest that PBDEs disrupt sex hormones and adversely affect development of the reproductive system. Objective(s): In the present study, we examined whether there is a link between maternal hair PBDE concentrations and the risk of cryptorchidism (undescended testes) in male infants; testis descent is known to be dependent on androgens.

Method(s): Full-term male infants were recruited through clinics in Montreal, Toronto, and London, Canada. Boys with cryptorchidism at 3-18 months of age (n=137) were identified by pediatric urologists and surgeons; similar-aged controls (n=158) had no genitourinary abnormalities as assessed by pediatricians. Eight BDE congeners (BDE-28, -47, -99, -100, -153, -154, -183, -209) were measured by GC-MS (gas chromatography-mass spectrometry) in maternal hair samples collected at the time of recruitment.

Result(s): The PBDE geometric mean for maternal hair was 45.35 pg/mg for controls and 50.27 pg/mg for cases; the concentrations of three BDEs (BDE-99, -100, and -154) were significantly higher in cases than controls in unadjusted models. In adjusted models, every 10-fold increase in the concentration of maternal hair BDE-99 [OR=2.53 (95% CI: 1.29, 4.95)] or BDE-100 [OR=2.45 (95% CI: 1.31, 4.56)] was associated with more than a doubling in the risk of cryptorchidism. BDE-154 [OR=1.88 (95% CI: 1.08, 3.28)] was also significant.

Conclusion(s): Our results suggest that maternal exposure to BDE-99, -100, and -154 may be associated with abnormal migration of testes in the male fetus. This may be due to the anti-androgenic properties of the PBDEs.

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Institution

(Goodyer, Atehortua) Research Institute of McGill University Health Centre, Montreal, QC, Canada (Goodyer) Department of Pediatrics, McGill University, Montreal, QC, Canada

(Poon, Aleksa, Carnevale) Department of Pharmacology and Toxicology, Hospital for Sick Children, University of Toronto, Toronto, ON, Canada

(Aleksa) Leslie Dan School of Pharmacy, University of Toronto, Toronto, ON, Canada

(Hou, Chevrier) Department of Epidemiology, Biostatistics and Occupational Health, McGill University, Montreal, QC, Canada

(Jednak) Department of Pediatric Urology, McGill University, Montreal, QC, Canada

(Emil) Department of Pediatric General and Thoracic Surgery, McGill University, Montreal, QC, Canada

(Bagli) Department of Pediatric Urology, Hospital for Sick Children, University of Toronto, Toronto, ON, Canada

(Dave) Division of Pediatric Urology, London Health Sciences Centre, London, ON, Canada

(Hales) Department of Pharmacology and Therapeutics, McGill University, Montreal, QC, Canada

Publisher

Public Health Services, US Dept of Health and Human Services (E-mail: ehp@jeditorial.com)

Year of Publication

2017

335.

Laparoscopic pediatric inguinal hernia repair: a controlled randomized study.

Abd-Alrazek M., Alsherbiny H., Mahfouz M., Alsamahy O., Shalaby R., Shams A., Elian A., Ashour Y.

Embase

Journal of Pediatric Surgery. 52 (10) (pp 1539-1544), 2017. Date of Publication: 2017.

[Article]

AN: 617494642

Background Inguinal hernia repair is one of the most frequently performed surgical procedure in infants and young children. Laparoscopic hernia repair in infancy and childhood is still debatable. There are many techniques available for laparoscopic hernia repair in pediatrics. The aim of the study is to compare laparoscopic intracorporeal purse-string suture ligation of the hernia defect leaving the sac intact versus disconnection of the hernia sac with intracorporeal suturing of proximal part. Patients and methods A prospective controlled randomized study of laparoscopic repair of congenital inguinal hernia (CIH) was conducted over a period of 2 years and 8 months from April 2014 to December 2016. All patients were randomized into two equal groups: Group I (n = 66) received intracorporeal purse string suture ligation of the hernia sac at internal inguinal ring [IIR] leaving the sac intact; and Group II (n = 66) received disconnection of the hernia sac with intracorporeal suture of proximal part at IIR. Inclusion criteria Male patient with bilateral CIH, questionable other side, cases of CIH associated with umbilical hernia and parental request. Exclusion criteria Recurrent cases, complicated cases, hernia of canal of Nuck in females, inguinal hernia with undescended testis, parental refusal. The main outcome measurements were operative time, postoperative hydrocele formation, recurrence rate. Results This study included 132 patients with 157 hernia defects. Their age ranged from 6 months to 3 years. Statistically significant differences regarding the demographic data of the groups. All cases were completed successfully without conversion. There was no statistically significant difference between groups regarding intraoperative complications and hospital stay. There was statistically significant difference in the operative time and post-operative complications between the studied groups. Conclusions Laparoscopic inguinal hernia repair by disconnection of the hernia sac at the IIR with peritoneal closure is safe and feasible method. It has a lower recurrence rate than the purse string suturing leaving the sac intact.

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Institution

(Abd-Alrazek, Alsherbiny, Mahfouz, Alsamahy, Shalaby, Shams, Elian, Ashour) Pediatric Surgery Department, Al-Azhar University Hospitals, Cairo, Egypt

Publisher
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2017

336.

The possible role of AMH in shortening the gubernacular cord in testicular descent: A reappraisal of the evidence.

Hutson J.M., Lopez-Marambio F.A.

Embase

Journal of Pediatric Surgery. 52 (10) (pp 1656-1660), 2017. Date of Publication: 2017.

[Article]

AN: 616717218

Background/Aim Anti-Mullerian hormone (AMH), also called Mullerian inhibiting substance (MIS), is glycoprotein hormone secreted by the fetal Sertoli cells to regulate regression of the Mullerian ducts, the anlagen of the uterus, fallopian tubes, and upper vagina. After its existence was predicted in 1946 and its isolation and purification in the 1970's, a huge amount of information has been gathered on its molecular biology and function in the last 30-40 years. Once thought to be a locally acting factor in the male fetus during sexual differentiation, it is now recognized as an endocrine hormone present in both sexes and with functions throughout life. One of the remaining controversies is the possible role of AMH during fetal testicular descent. In the human with aberrant AMH function, the boy has cryptorchidism with persistent Mullerian duct syndrome (PMDS), where the testes are often intraabdominal and on an abnormally long gubernacular cord. By contrast, in rodent models knockout of the AMH gene does not cause cryptorchidism.

Methods/Results In this review we examined the evidence in the literature for and against a role for AMH in testicular descent and considered the implications of the different anatomy of the gubernacular cord in rodents versus children. Conclusion We conclude that AMH may have a role in shortening the gubernacular cord in humans which is concealed in rodent models by differences in anatomy of the gubernacular cord in rodents. The controversy could be resolved by re-examination of the gubernacular cord in boys with PMDS and mice with AMHKO. Type of study Review. Level of evidence V

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Status

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Author NameID

Hutson, John M.; ORCID: <http://orcid.org/0000-0002-3922-8366>

Institution

(Hutson) Department of Paediatrics, University of Melbourne, Australia (Hutson) Urology

Department, The Royal Children's Hospital, Melbourne, Australia

(Hutson) F Douglas Stephens Surgical Research Group, Murdoch Childrens Research Institute, Melbourne, Australia

(Lopez-Marambio) Melbourne Medical School, University of Melbourne, Australia

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337.

Evaluation of Pediatric Undescended Testes with Elastasonography.

Cildag M.B.

Embase

Journal of Medical Ultrasound. 25 (3) (pp 157-160), 2017. Date of Publication: September 2017.

[Article]

AN: 615952164

Background Undescended testes, which are defined as the failure of testes to descend to scrotum, are the most common developmental defect in male infants. Indirect evaluation of histologic damage can be performed with the help of palpation during operation. Hard texture of testes tissue is likely related with histological damage. Real-time elastography is an emerging technology of ultrasonic imaging of soft tissue strain and elasticity, it aims at providing information regarding the mechanical properties of tissues, such as their hardness or stiffness. This study aim was to investigate the changes in strain and elasticity of testes tissue by using elastography technique. Materials and methods A total of 32 patients, who had undescended testes were

included in this study. Only two patients had bilateral undescended testes, other patients had unilateral. The age of the patients were recorded according to the time of ultrasonographic (USG) examination. The undescended testes was displayed in the elastographic box with the neighbouring subcutaneous fat tissues. The strain ratios were measured as the ratios of the elasticities of the subcutaneous fat tissue to the elasticities of the undescended testes. Results A total of 32 patients with 34 testes were included in the study. The mean age of the patients with undescended testes was 32.6 months (range 7-60 months). The mean strain ratios were 0.67 (range 0.12-1.41) for the undescended testes and there were no significant differences in undescended testes strain ratios related to patient age ($p = 0.453$). Conclusion This preliminary study showed that there were no significant fibrosis which can be demonstrated with elastosonography before the age of 5 years old. Additional studies with histopathological results are needed to identify sensitivity and specificity of elastosonography in undescended testis and in planning optimal operation time for these patients.

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(Cildag) Department of Diagnostic and Interventional Radiology, Adnan Menderes University, Aydin, Turkey

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Year of Publication

2017

338.

Anti-Mullerian hormone (AMH) determinations in the pediatric and adolescent endocrine practice. Weintraub A., Eldar-Geva T.

Embase

Pediatric Endocrinology Reviews. 14 (4) (pp 364-370), 2017. Date of Publication: June 2017.

[Article]

AN: 624293922

Anti-Mullerian hormone (AMH), secreted by immature testicular Sertoli-cells, triggers the regression of male fetal Mullerian ducts. During puberty, AMH is downregulated by intratesticular testosterone. In females, AMH is secreted from granulosa cells of immature ovarian follicles from late prenatal life until menopause; serum concentration is 5-20 times lower in females than in males through lifetime. In boys, AMH determination is useful in the clinical setting as a marker of Sertoli cell function. Serum AMH is low in infants with hypogonadotrophic hypogonadism (and increases with FSH treatment), in patients with primary hypogonadism from early postnatal life and in Klinefelter syndrome from midpuberty. In boys with nonpalpable gonads, AMH determination is useful to distinguish between cryptorchidism and anorchism, as well as differentiating the dysgenetic causes of disorders of sexual development from those due to defective androgen synthesis or action. AMH can be used as a marker of sertoli/granulosa cell tumors and primary ovarian insufficiency in girls with delayed puberty, Turner Syndrome and after treatment with gonadotoxic agents.

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PMC Identifier

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Embase

Institution

(Weintraub, Eldar-Geva) IVF Unit, Department of Obstetrics and Gynecology, Shaare Zedek Medical Center, 12 Shmuel Bait St. PO Box 3235, Jerusalem 9103102, Israel

Publisher

YS Medical Media Ltd. (E-mail: per@medmedia.co.il)

Year of Publication

2017

339.

Contralateral metachronous undescended testis: Is it predictable?.

Cancian M., Ellsworth P., Caldamone A.

Embase

Journal of Pediatric Urology. 13 (5) (pp 504.e1-504.e5), 2017. Date of Publication: October 2017.

[Article]

AN: 615611417

Introduction Metachronous undescended testis (mcUDT), an acquired UDT after contralateral orchiopexy, can occur in some boys. If one were able to predict its occurrence, one might consider a proactive approach or at least one would be able to counsel the parents accordingly. Our hypothesis was there may be characteristics evident at the time of initial orchiopexy which could predict the development of contralateral mcUDT. Objective The aim was to identify factors present at initial orchiopexy that predict development of subsequent mcUDT. Study design Subjects were identified using the Current Procedural Terminology code for inguinal orchiopexy (54640). We included patients from January 1997 to October 2015. We included patients who underwent orchiopexy for unilateral UDT (uUDT). The study population consisted of patients who had undergone metachronous orchiopexies; controls were patients who were 17 years at time of data collection with a single orchiopexy. Cox proportional hazard regression was used to model the relationship between possible predictors of subsequent UDT using PROC PHREG with SAS Software 9.4. Results From 1035 eligible patients we identified 38 with mcUDT and 207 controls (uUDT). Median age at the first orchiopexy of mcUDT patients was 2.5 years (min/max, 0.50/10.4 years) and 8.2 years (min/max 0.70/12.8 years) for uUDT, $p < 0.0001$. Subjects with a contralateral retractile testis on preoperative exam had a 4.2 times higher rate of subsequent UDT than patients with a contralateral descended testis (95% CI 2.077-8.353). The rate of mcUDT was 6.7 times higher if the testis was a retractile testis under anesthesia (95% CI 2.7-16.5) (Table). Discussion Contralateral retractile UDT was a significant predictor of mcUDT. We believe patients with a contralateral retractile testis at time of orchiopexy should be counseled on bilateral orchiopexy. The risks of complications with orchiopexy should be weighed against risks of a subsequent surgery and anesthesia event. Conclusion A discussion of risks and benefits regarding bilateral orchiopexies should be undertaken with the parents prior to surgery in the setting of an UDT with contralateral retractile testis. [Table presented]

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Embase

Author NameID

Cancian, Madeline; ORCID: <http://orcid.org/0000-0003-4503-846X>

Institution

(Cancian, Caldamone) Department of Urology, Warren Alpert Medical School, Brown University, Providence, RI, United States (Ellsworth) Division of Urology, Nemours Children's Hospital, Orlando, FL, United States

Publisher

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2017

340.

The prostatic utricle: An under-recognized condition resulting in significant morbidity in boys with both hypospadias and normal external genitalia.

Hester A.G., Kogan S.J.

Embase

Journal of Pediatric Urology. 13 (5) (pp 492.e1-492.e5), 2017. Date of Publication: October 2017.

[Article]

AN: 614885522

Introduction Pediatric presentations of a prostatic utricle have received only scant attention. Though recognized with increased frequency in boys with hypospadias, little is described about their incidence and potential for morbidity in boys with normal external genitalia. Methods We initially reviewed a cohort of 64 patients with hypospadias seen over a 3-year period to determine the frequency of investigative lower urinary tract studies and utricle identification. Children with disorders of sexual differentiation were excluded from this review. A subsequent group of 70 boys with hypospadias and 23 boys with normal external genitalia presenting with lower urinary tract symptoms (LUTS) who were found to have an unsuspected utricle were reviewed. This comparative group was investigated since symptomatology was the indication for evaluation, contrasting with those in the hypospadias group who were investigated because of hypospadias presence alone. Results In our initial review of 64 patients only 24 (37.5%) underwent an investigative study and six (9.4%) had a utricle. Three (50%) required surgical excision, allowing their hypospadias repair to proceed. Results in the subsequent group with hypospadias confirmed these findings with increased rates of investigation and identification. The boys with normal external male genitalia all required surgery since symptoms were the result of the utricle alone. Penile pain with voiding, hematuria, epididymitis, and urinary infection were the most common causes for interventions. Conclusions The prostatic utricle should be considered as a cause of morbidity in boys with both normal external genitalia and those with hypospadias. Endoscopic or radiological evaluation (see Figure) should be undertaken in all boys with proximal hypospadias, boys with hypospadias and associated cryptorchidism, and those with hypospadias with

associated urinary symptoms. Boys with normal external genitalia with lower urinary tract symptoms not explained with imaging should undergo cystoscopy, as an unidentified unsuspected utricle may be the underlying cause.[Figure presented]

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Embase

Author NameID

Hester, Austin G.; ORCID: <http://orcid.org/0000-0003-1495-6487>

Institution

(Hester, Kogan) Department of Urology, Wake Forest University, Winston-Salem, NC, United States

Publisher

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2017

341.

Prevalence of untreated surgical conditions in Rural Rwanda: A population-based cross-sectional study in burera district.

Maine R.G., Linden A.F., Riviello R., Kamanzi E., Mody G.N., Ntakiyiruta G., Kansayisa G., Ntaganda E., Niyonkuru F., Mubiligi J.M., Mpunga T., Meara J.G., Hedt-Gauthier B.L.

Embase

JAMA Surgery. 152 (12) (no pagination), 2017. Article Number: e174013. Date of Publication: December 2017.

[Article]

AN: 620071761

IMPORTANCE In low- and middle-income countries, community-level surgical epidemiology is largely undefined. Accurate community-level surgical epidemiology is necessary for surgical health systems planning. **OBJECTIVE** To determine the prevalence of surgical conditions in Burera District, Northern Province, Rwanda. **DESIGN, SETTING, AND PARTICIPANTS** A cross-

sectional study with a 2-stage cluster sample design (at village and household level) was carried out in Burera District in March and May 2012. A team of surgeons randomly sampled 30 villages with probability proportionate to village population size, then sampled 23 households within each village. All available household members were examined. MAIN OUTCOMES AND MEASURES The presence of 10 index surgical conditions (injuries/wounds, hernias/hydroceles, breast masses, neck masses, obstetric fistulas, undescended testes, hypospadias, hydrocephalus, cleft lip/palate, and clubfoot) was determined by physical examination. Prevalence was estimated overall and for each condition. Multivariable logistic regression was performed to identify factors associated with surgical conditions, accounting for the complex survey design. RESULTS Of the 2165 examined individuals, 1215 (56.2%) were female. The prevalence of any surgical condition among all examined individuals was 12%(95%CI, 9.2-14.9%). Half of conditions were hernias/hydroceles (49.6%), and 44%were injuries/wounds. In multivariable analysis, children 5 years or younger had twice the odds of having a surgical condition compared with married individuals 21 to 35 years of age (reference group) (odds ratio [OR], 2.2; 95%CI, 1.26-4.04; P = .01). The oldest group, people older than 50 years, also had twice the odds of having a surgical condition compared with the reference group (married, aged >50 years: OR, 2.3; 95%CI, 1.28-4.23; P = .01; unmarried, aged >50 years: OR, 2.38; 95%CI, 1.02-5.52; P = .06). Unmarried individuals 21 to 35 years of age and unmarried individuals aged 36 to 50 years had higher odds of a surgical condition compared with the reference group (aged 21-35 years: OR, 1.68; 95%CI, 0.74-3.82; P = .22; aged 36-50 years: OR, 3.35; 95%CI, 1.29-9.11; P = .02). There was no statistical difference in odds by sex, wealth, education, or travel time to the nearest hospital. CONCLUSIONS AND RELEVANCE The prevalence of surgically treatable conditions in northern Rwanda was considerably higher than previously estimated modeling and surveys in comparable low- and middle-income countries. This surgical backlog must be addressed in health system plans to increase surgical infrastructure and workforce in rural Africa.

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Status

Embase

Institution

(Maine) Department of Surgery, Harborview Medical Center, Seattle, WA, United States (Maine)

Now with Department of Surgery, University of North Carolina, School of Medicine, 4008 Burnett-Womack Building, Campus Box 7228, Chapel Hill, NC 27599-7228, United States

(Linden, Riviello, Mody, Meara) Program in Global Surgery and Social Change, Harvard Medical School, Boston, MA, United States

(Linden) Section of Pediatric Surgery, General Surgery, University of Chicago Comer's Children Hospital, Chicago, IL, United States

(Riviello, Mody) Center for Surgery and Public Health, Brigham AndWomen's Hospital, Boston, MA, United States

(Kamanzi) University of Global Health Equity (UGHE), Kigali, Rwanda

(Ntakiyiruta) Ejo Heza Surgical Centre, Kigali, Rwanda

(Kansayisa) Rwanda Military Hospital, Kigali, Rwanda

(Ntaganda, Niyonkuru) University Teaching Hospital-Kigali (CHUK), Kigali, Rwanda

(Mubiligi, Hedt-Gauthier) Partners in Health/Inshuti Mu Buzima, Kigali, Rwanda

(Mpunga) Rwanda Ministry of Health, Butaro District Hospital, Burera, Rwanda

(Meara) Department of Plastic and Oral Surgery, Boston Children's Hospital, Boston, MA, United States

(Hedt-Gauthier) Department of Global Health and Social Medicine, Harvard Medical School, Boston, MA, United States

Publisher

American Medical Association (E-mail: smcleod@itsa.ucsf.edu)

Year of Publication

2017

342.

The hematological parameters in testicular cancer.

Caliskan S., Kaba S., Ozsoy E., Koca O., Akyuz M., Ozturk M.I.

Embase

Journal of Oncological Science. 3 (3) (pp 117-119), 2017. Date of Publication: December 2017.

[Article]

AN: 618844244

Although testicular cancer (TCa) is rare neoplasm that occurs in young men aged between 18 and 35 years. The risk factors are cryptorchidism, family history, and infertility. The aim of this study was to investigate the diagnostic efficacy of neutrophil to lymphocyte ratio for the diagnosis of TCa. The patients who underwent orchiectomy in our unit reviewed retrospectively. Age of the patients, the laboratory results and pathological reports were recorded. The neutrophil to lymphocyte ratio (NLR) was calculated as the neutrophil counts divided to the lymphocyte counts.

The patients were divided into two groups according to the pathology record. The testicular malign neoplasms are included in group 1 and group 2 includes the patients who had cryptorchidism and atrophic testes without any malignancy and inflammation. For statistical analysis, student t test was used for comparing the data between groups and the area under curves were used for NLR, neutrophil and lymphocyte counts in the diagnosis of testicular malign neoplasms. There were 285 patients in the present study. The patients' age was between 10 and 90 with a mean age of 36.87 +/- 11.83 and 37.24 +/- 20.31 years in groups respectively. The neutrophil, white blood cell counts and NLR were significantly higher in group 1 and lymphocyte count was lower in patients with testicular cancer with statistical significance. The area under curve was 0.645, 0.626, 0.578 for NLR, neutrophil and lymphocyte counts for the diagnosis of TCa. Mixed germ cell tumor was the most common histologic subtype with an incidence of 51.58% (n:65 patients) and seminomas were reported 37.30% (n:47 patients) of the patients. Testicular cancer has low incidence when compared the other urologic malignancies. There are only three tumor markers that include alpha-fetaprotein, human chorionic gonadotropin and lactate dehydrogenase for testicular cancer diagnosis. The current study showed Neutrophil to lymphocyte ratio (NLR) may be used as a biomarker for TCa. Further studies are needed to define the association between NLR and testicular cancer.

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Institution

(Caliskan, Kaba, Ozsoy, Koca, Akyuz, Ozturk) Haydarpasa Numune Training and Research Hospital, Department of Urology, Istanbul, Turkey

Publisher

Turkish Society of Medical Oncology

Year of Publication

2017

343.

Smoking and its association with cryptorchidism in Down syndrome.

De Rezende Duarte A.M.B., De Bessa J., Mrad F.C.C., Tibirica S.H.C., De Sa Camargo M.L., Vieira A.A.P., Araujo F.C.C., De Figueiredo A.A., De Bastos-Netto J.M.

Embase

Revista da Associacao Medica Brasileira. 63 (8) (pp 693-696), 2017. Date of Publication: August 2017.

[Article]

AN: 618665118

Introduction: Cryptorchidism is a common and prevalent condition in patients with Down syndrome. Environmental factors, such as smoking, can be associated with malformations during fetal development. The study of the prevalence of cryptorchidism and its association with parental tobacco use in Down syndrome can contribute to alert health care professionals, patients and family members regarding the prevention of the harms caused by cryptorchidism and its possible predisposing factors.

Objective(s): To evaluate the prevalence of cryptorchidism in Down syndrome and its association with maternal and paternal smoking.

Method(s): Forty (40) patients of a public clinic specialized in Down syndrome were evaluated, using a semi-structured questionnaire for evaluation of antecedents and sociodemographic characteristics, as well as physical and complementary examinations.

Result(s): Cryptorchidism was observed in 27.5% of the patients (95CI 15.98-42.96). Of these, 55% (5/9) were the children of mothers who smoked during pregnancy, and 19.35% (6/31) were the children of mothers who did not smoke during pregnancy (OR = 5.26 [95CI 1.06-25.41]; p=0.032). Similarly, paternal smoking was also observed in greater frequency among the parents of cryptorchid patients compared with subjects with descended testis, 63.36% (7/11) and 31.03% (9/29), respectively (OR = 3.89 [95CI 0.91-16.73]; p=0.060).

Conclusion(s): The prevalence of cryptorchidism is high in patients with Down syndrome. We can show a strong association between smoking parents and the occurrence of cryptorchidism, especially when it comes to maternal smoking.

PMC Identifier

28977107 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28977107>]

Status

Embase

Institution

(De Rezende Duarte, Mrad, Tibirica, De Sa Camargo, Vieira, Araujo, De Figueiredo, De Bastos-Netto) Universidade Federal de Juiz de for A, Juiz de Fora, MG, Brazil (De Bessa) Universidade Estadual de Feira de Santana, Feira de Santana, BA, Brazil

(De Bastos-Netto) Hospital Maternidade Therezinha de Jesus, Faculdade de Ciencias Medicas e da Saude de Juiz de for A, Juiz de Fora, MG, Brazil

Publisher

Associacao Medica Brasileira

Year of Publication

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344.

Comparative study of surgical results with and without ligation of hernia sac in orchiopexy of pediatric patients with undescended testis.

Amanollahi O., Kashanian Z.

Embase

Journal of Kermanshah University of Medical Sciences. 21 (2) (pp 69-72), 2017. Date of Publication: Summer 2017.

[Article]

AN: 619469032

Introduction: Undescended Testis (UDT) is a congenital anomaly and its common complications include testicular malignancy, infertility, testicular torsion, risk of trauma. Treatment of UDT is surgical (orchiopexy), and since it is always with a hernia sac, one stage of the surgery is separating, cutting and ligation of the hernia sac, in order to avoid an inguinal hernia. In this study, we investigated whether surgical procedure without inguinal hernia sac ligation and only with separating and cutting it at the deep ring level could be a more appropriate alternative to the classic surgical procedure.

Method(s): This clinical trial was conducted in march 2012-2013 in kermanshah university of medical sciences, on children of 2 months to 11 years old with UDT. The patients were followed up for 3to 4years.

Result(s): A total of 109 patients underwent orchiopexy during the study. Among them, 38.33% underwent orchiopexy with ligation of the hernia sac and 61.46% underwent orchiopexy without ligation of the hernia sac and with separating and cutting sac at the deep inguinal ring. No cases of an inguinal hernia were observed in the two groups during the follow-up period of 4-5 years. Other complications such as wound infection, hydrocele,, hematoma, and hemorrhage at the surgical site also did not differ significantly between the two groups.

Conclusion(s): Regarding the findings of this study, it appears that the use of the non-ligation technique does not increase inguinal hernia, and can be a good alternative to the classical method of hernia sacligation.

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Institution

(Amanollahi) Dept. of Surgery, School of Medicine, Kermanshah University of Medical Sciences, Kermanshah, Iran, Islamic Republic of (Kashanian) Kermanshah University of Medical Sciences, Kermanshah, Iran, Islamic Republic of

Publisher

Kermanshah University of Medical Sciences (E-mail: ahmadiar1012@yahoo.com)

Year of Publication

2017

345.

Treatment for incarcerated indirect hernia with "Cross-Internal Ring" inguinal oblique incision in children.

Yan X.-Q., Yang J., Zheng N.-N., Kuang H.-F., Duan X.-F., Bian H.-Q.

Embase

Journal of Research in Medical Sciences. 22 (9) (no pagination), 2017. Article Number: 106. Date of Publication: September 2017.

[Article]

AN: 619449406

Background: This study aims to evaluate the utility of the "Cross-Internal Ring" inguinal oblique incision for the surgical treatment of incarcerated indirect hernia (IIH) complicated with severe abdominal distension.

Material(s) and Method(s): Patients of IIH complicated with severe abdominal distension were reviewed retrospectively. All patients received operation through the "Cross-Internal Ring" inguinal oblique incision.

Result(s): There were totally 13 patients were included, male to female ratio was 9-4. The time for patients to resume oral feeding varying from 2 to 5 days after operation, no complications include delayed intestinal perforation, intra-abdominal abscess, and incision infection happened. Average postoperative hospital stay was 5.2 days. All cases were followed up for 6-18 months. No recurrence or iatrogenic cryptorchidism happened.

Conclusion(s): "Cross-Internal Ring" inguinal oblique incision is a simple, safe, and reliable surgical method to treat pediatric IIH complicated with severe abdominal distension.

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Institution

(Yan, Yang, Zheng, Kuang, Duan, Bian) Department of Pediatric Surgery, Tongji Medical College, Wuhan Children's Hospital, Huazhong University of Science and Technology, Wuhan 430016, China

Publisher

Isfahan University of Medical Sciences(IUMS) (Hezar Jerib Avenue, P.O. Box 81745-319, Isfahan, Iran, Islamic Republic of)

Year of Publication

2017

346.

Advances in paediatric urology.

Diamond D.A., Chan I.H.Y., Holland A.J.A., Kurtz M.P., Nelson C., Estrada C.R., Bauer S., Tam P.K.H.

Embase

The Lancet. 390 (10099) (pp 1061-1071), 2017. Date of Publication: 9 - 15 September 2017.

[Review]

AN: 619146569

Paediatric urological surgery is often required for managing congenital and acquired disorders of the genitourinary system. In this Series paper, we highlight advances in the surgical management of six paediatric urological disorders. The management of vesicoureteral reflux is evolving, with advocacy ranging from a less interventional assessment and antimicrobial prophylaxis to surgery including endoscopic injection of a bulking agent and minimally invasive ureteric reimplantation. Evidence supports early orchidopexy to improve fertility and reduce malignancy in boys with undescended testes. A variety of surgical techniques have been developed for hypospadias, with excellent outcomes for distal but not proximal hypospadias. Pelvi-ureteric junction obstruction is mostly detected prenatally; indications for surgery have been refined with evidence, and

minimally invasive pyeloplasty is now standard. The outlook for patients with neurogenic bladder has been transformed by a combination of clean intermittent catheterisation, algorithms of diagnostic investigations, and innovative medical and surgical therapies. Posterior urethral valves are associated with considerable mortality; fetal diagnosis allows stratification of candidates for intervention, but ongoing bladder dysfunction in patients after valve ablation remains a cause of long-term morbidity.

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(Diamond, Kurtz, Nelson, Estrada, Bauer) Department of Urology, Boston Children's Hospital, Harvard Medical School, Boston, MA, United States (Chan, Tam) Department of Surgery, The University of Hong Kong, Queen Mary Hospital, Hong Kong

(Holland) Department of Paediatric Surgery, The Children's Hospital at Westmead, Westmead, NSW, Australia

(Holland) Sydney Medical School, University of Sydney, Sydney, NSW, Australia

Publisher

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Year of Publication

2017

347.

Inguinal exploration for the management of impalpable undescended testes.

Thapa B., Pun M.S.

Embase

Journal of Nepal Paediatric Society. 37 (1) (pp 36-40), 2017. Date of Publication: January-April 2017.

[Article]

AN: 619098637

Introduction: The primary goal of surgical intervention with an impalpable testis is to locate and reposition the gonad. There has been much debate in the management of impalpable

undescended testes. Many centres still advocate the role of open inguinal exploration in impalpable testes.

Material(s) and Method(s): This retrospective study included 35 male patients. The clinical notes were reviewed for details of age at operation, side, location and condition of testes intraoperatively and the type of operation performed. Standard approach of inguinal explorations was performed under general anaesthesia. Follow up in first week and after 3 months was done.

Result(s): 25(71.42%) impalpable testes were in left side, 9(25.71%) were on right side and 1(2.85%) was bilateral. At exploration 17(47.22%) were intracanalicular, nine (25%) were intraabdominal with seven low and two high, 49(11.11%) were scrotal and six (16.66%) were absent, seven (41.17%) canalicular testes underwent orchidopexy and 10(58.22%) underwent gonadectomy for atrophied testes. Seven (77.77%) low abdominal testes were brought down to scrotum and two (22.22%) high abdominal were brought down to lower inguinal area as a first stage surgery. All 4(100%) scrotal testes were atrophied and gonadectomy performed. Six patients were diagnosed anorchia, 14 (87.5%) of impalpable testes that underwent orchidopexy were in a follow up with excellent results. One high first stage orchidopexy and another canalicular testes were found to have atrophied who did orchidectomy in follow-up.

Conclusion(s): Inguinal exploration is a safe, reliable and successful surgical procedure for the management of impalpable testes including intraabdominal testes without procedure related complications.

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Status

Embase

Institution

(Thapa, Pun) Department of Paediatric Surgery, Kanti Children's Hospital, Maharajgunj, Kathmandu, Nepal

Publisher

Nepal Paediatric Society (NEPAS) (E-mail: nepas@healthnet.org.np)

Year of Publication

2017

A Three years audit of surgical management of un-descended testis - Experience at King Fahad Hospital, al Baha, KSA.

Sharif M., Hafiz A.S.B., Bashir T., Elsiddig I.E., Ibrahim M.

Embase

Pakistan Journal of Medical and Health Sciences. 11 (3) (pp 829-831), 2017. Date of Publication: July-September 2017.

[Article]

AN: 619052363

Aims: To collect the data regarding age, preoperative, operative findings & postoperative outcomes and complications. To compare our results with national and international literatures.

Duration: 3 years (January, 2011 to December, 2013) Study Design: Retrospective analysis.

Setting(s): Department of Pediatric Surgery, King Fahad Hospital, Al Baha, KSA.

Method(s): A retrospective study was conducted from Jan 2011 to Dec 2013. The data of all operated cases of UDT was retrieved from Operation Theatre Register, Inpatient Department, OPD and from medical record office. The files of these patients were reviewed regarding age at presentation, preoperative and postoperative findings and outcomes. All operative cases were included in the study except with incomplete record and missing file.

Result(s): A total of 141 patients were operated but due to incomplete records or missing files, only 116 patients were analyzed for results. The operated patients have age ranging from 8 months to 11 years. 68(58.6%) patients have age up to 2 years and 48(41.3%) were more > 2 years age. The mean age at surgery was 34 months (almost 3 years). 58(50%) were operated as daycare surgery and discharged on same day while 55(47.4%) were admitted as inpatient and discharged on first postoperative day. Three patients of undescended testis presented in emergency as case of testicular torsion.

Conclusion(s): We conclude that majority of the patients of UDT presented or operated late and also in late operated cases, testis was small size, which has bad impact on fertility in these children. So there is need to run awareness program through symposiums and media among general practitioners, pediatrician and public about early diagnosis, preoperative timing and early surgical management these children to prevent complications like infertility, sub-fertility, malignancy and psychological stress.

Status

Embase

Institution

(Sharif, Hafiz, Bashir, Elsiddig, Ibrahim) Department of Pediatric Surgery, King Fahad Hospital, Al Baha, Saudi Arabia

Publisher

Lahore Medical And Dental College (Lahore Medical & Dental College, Lahore x, Pakistan. E-mail: nayyar_salam@yahoo.com)

Year of Publication

2017

349.

DMRTC2, PAX7, BRACHYURY/t and tert are implicated in male germ cell development following curative hormone treatment for cryptorchidism-induced infertility.

Gegenschatz-Schmid K., Verkauskas G., Demougin P., Bilius V., Dasevicius D., Stadler M.B., Hadziselimovic F.

Embase

Genes. 8 (10) (no pagination), 2017. Article Number: 267. Date of Publication: 11 Oct 2017.

[Article]

AN: 618783974

Defective mini-puberty results in insufficient testosterone secretion that impairs the differentiation of gonocytes into dark-type (Ad) spermatogonia. The differentiation of gonocytes into Ad spermatogonia can be induced by administration of the gonadotropin-releasing hormone agonist, GnRH α (Buserelin, INN)). Nothing is known about the mechanism that underlies successful GnRH α treatment in the germ cells. Using RNA-sequencing of testicular biopsies, we recently examined RNA profiles of testes with and without GnRH α treatment. Here, we focused on the expression patterns of known gene markers for gonocytes and spermatogonia, and found that DMRTC2, PAX7, BRACHYURY/T, and TERT were associated with defective mini-puberty and were responsive to GnRH α . These results indicate novel testosterone-dependent genes and provide valuable insight into the transcriptional response to both defective mini-puberty and curative GnRH α treatment, which prevents infertility in man with one or both undescended (cryptorchid) testes.

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Embase

Institution

(Gegenschatz-Schmid, Hadziselimovic) Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, Liestal 4410, Switzerland (Verkauskas, Bilius) Children's Surgery Centre, Faculty of Medicine, Vilnius of University, Vilnius 01513, Lithuania

(Demougin) Biozentrum, Life Sciences Training Facility, University of Basel, Basel 4001, Switzerland

(Dasevicius) Institute for Pathology, National Centre of Pathology, Affiliate of Vilnius University Hospital Santariskiu Klinikos, Vilnius 08406, Lithuania

(Stadler) Friedrich Miescher Institute for Biomedical Research, Basel 4058, Switzerland

(Stadler) Swiss Institute of Bioinformatics, Basel, Switzerland

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350.

Focal dermal hypoplasia (Goltz Syndrome): A cross-sectional study from Eastern India.

Ghosh S.K., Dutta A., Sarkar S., Nag S.S., Biswas S.K., Mandal P.

Embase

Indian Journal of Dermatology. 62 (5) (pp 498-504), 2017. Date of Publication: September-October 2017.

[Article]

AN: 618525138

Introduction: Focal dermal hypoplasia (Goltz syndrome), is an extremely rare genetic disorder characterized by distinct skin manifestations and a wide range of abnormalities involving the ocular, dental, skeletal, urinary, gastrointestinal, cardiovascular, and central nervous systems.

The objective of the present series is to emphasize the different typical as well as unusual features of this rare syndrome. Methodology: This cross-sectional observational study was performed over a period of 8 years in a tertiary care hospital of Eastern India. Consecutive patients with the clinical diagnosis of Goltz syndrome were studied.

Result(s): A total of 8 patients with Goltz syndrome were evaluated. Out of them, one patient was a boy and the rest were girl. The age ranged from 3 days to 9 years. There was no family history. A characteristic Blaschkoid hypo- and hyper-pigmented skin lesions, congenital nodular fat

herniation, and skin atrophy were present in all patients. Congenital cutaneous aplasia was present in 50% of the patients. Facial asymmetry and ear deformity (megalopinna and low-set ears) were seen in 37.5% and 12.5% of patients, respectively. Cutaneous telangiectasia was noticed in 37.5% of patients. Freckle- and lentiginous-like pigmentation within the hypopigmented macules was found in 25% of patients. Raspberry-like papillomas around mouth were documented in 6 (75%) patients. Dysplastic nail changes with ridging were seen in 7 (87.5%) patients. Genital abnormality in the form of bilateral undescended testes and microphthalmia with aniridia were found in one patient each. Limb defects were present in all patients. Left-sided renal agenesis was found in one patient. The patient also had multiple cortical cysts of the right kidney. Limitation(s): Genetic testing could not be performed in the present series.

Conclusion(s): Our case series showed a few unusual or extremely rare manifestations such as undescended testes, dermal sinus, kyphoscoliosis, aniridia, unilateral kidney agenesis, and renal cortical cysts among others.

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Institution

(Ghosh, Biswas, Mandal) Department of Dermatology, Venereology and Leprosy, R.G.Kar Medical College, 1, Khudiram Bose Road, Kolkata, West Bengal 700 004, India (Dutta, Nag) Department of Pediatrics, North Bengal Medical College, West Bengal, India (Sarkar) Department of Psychiatry, Calcutta National Medical College, Kolkata, West Bengal, India

Publisher

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351.

Risk factors for cryptorchidism.

Gurney J.K., Mcglynn K.A., Stanley J., Merriman T., Signal V., Shaw C., Edwards R., Richiardi L., Hutson J., Sarfati D.

Embase

Nature Reviews Urology. 14 (9) (pp 534-548), 2017. Date of Publication: 01 Sep 2017.

[Review]

AN: 618211027

Undescended testis-known as cryptorchidism-is one of the most common congenital abnormalities observed in boys, and is one of the few known risk factors for testicular cancer. The key factors that contribute to the occurrence of cryptorchidism remain elusive. Testicular descent is thought to occur during two hormonally-controlled phases in fetal development-between 8-15 weeks (the first phase of decent) and 25-35 weeks gestation (the second phase of descent); the failure of a testis to descend permanently is probably caused by disruptions to one or both of these phases, but the causes and mechanisms of such disruptions are still unclear. A broad range of putative risk factors have been evaluated in relation to the development of cryptorchidism but their plausibility is still in question. Consistent evidence of an association with cryptorchidism exists for only a few factors, and in those cases in which evidence seems unequivocal the factor is likely to be a surrogate for the true causal exposure. The relative importance of each risk factor could vary considerably between mother-son pairs depending on an array of genetic, maternal, placental and fetal factors-all of which could vary between regions. Thus, the role of causative factors in aetiology of cryptorchidism requires further research.

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Embase

Institution

(Gurney, Stanley, Signal, Shaw, Edwards, Sarfati) Department of Public Health, University of Otago, 23a Mein St, Newtown, Wellington 6037, New Zealand (Mcglynn) Division of Cancer Epidemiology and Genetics, National Cancer Institute, 9609 Medical Center Drive, Rockville, MD 20850, United States

(Merriman) Department of Biochemistry, University of Otago, 710 Cumberland Street, North Dunedin, Dunedin 9016, New Zealand

(Richiardi) Cancer Epidemiology Unit, University of Turin, Via Giuseppe Verdi, 8, Torino 10124, Italy

(Hutson) The Royal Childrens Hospital, 50 Flemington Rd, Parkville, Melbourne, VIC 3052,

Australia

Publisher

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352.

KBG syndrome: An Australian experience.

Murray N., Burgess B., Hay R., Colley A., Rajagopalan S., McGaughran J., Patel C., Enriquez A., Goodwin L., Stark Z., Tan T., Wilson M., Roscioli T., Tekin M., Goel H.

Embase

American Journal of Medical Genetics, Part A. 173 (7) (pp 1866-1877), 2017. Date of Publication: July 2017.

[Article]

AN: 616837459

In 2011, heterozygous mutations in the ANKRD11 gene were identified in patients with KBG syndrome. Since then, 100 cases have been described with the expansion of the clinical phenotype. Here we present 18 KBG affected individuals from 13 unrelated families, 16 with pathogenic mutations in the ANKRD11 gene. Consistent features included intellectual disability, macrodontia, and the characteristic broad forehead with hypertelorism, and a prominent nasal bridge. Common features included hand anomalies, cryptorchidism, and a large number of palate abnormalities. Distinctive findings in this series included malrotation of the abdominal viscera, bilateral inguinal herniae in two patients, basal ganglia calcification and the finding of osteopenia in three patients. Nine novel heterozygous variants were found and the genotype-phenotype correlation was explored. This report highlights the need for thorough examination and investigation of the dental and skeletal systems. The results confirm the specificity of ANKRD11 mutations in KBG and further evidence for this transcription repressor in neural, cardiac, and skeletal development. The description of further cases of KBG syndrome is needed to further delineate this condition, in particular the specific neurological and behavioral phenotype.

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Status

Embase

Institution

(Murray, Burgess, Hay, Goel) Hunter Genetics, Waratah, NSW, Australia (Colley, Rajagopalan)

Department of Clinical Genetics, Liverpool Hospital, NSW, Australia

(McGaughran, Patel) Genetic Health Queensland c/-Royal Brisbane and Women's Hospital

Herston, QLD, Australia

(Enriquez) Department of Clinical Genetics, Children's Hospital Westmead, Westmead, NSW,

Australia

(Goodwin) Department of Genetics Southblock, Nepean Hospital, Penrith, Australia

(Stark) Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Parkville,

VIC, Australia

(Tan) Murdoch Children's Research Institute Melbourne, VIC, Australia

(Wilson) Department of Clinical Genetics, Sydney Children's Hospital Network, NSW, Australia

(Roscioli) Garvan Institute of Medical Research, NSW, Australia

(Tekin) Division of Paediatric Genetics, Ankara University, Ankara, Turkey

(Tekin) Dr John T McDonald Foundation, Department of Human Genetics, Miami, FL, United

States

(Tekin) University of Miami School of Medicine, Miami, FL, United States

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353.

Corrigendum to "Management of undescended testes: European Association of Urology/European Society for Paediatric Urology Guidelines" (Journal of Pediatric Urology (2016) 12(6) (335-343) (S1477513116302443) (10.1016/j.jpuro.2016.07.014)).

Radmayr C., Dogan H.S., Hoebeke P., Kocvara R., Nijman R., Silay S., Stein R., Undre S., Tekgul S.

Embase

Journal of Pediatric Urology. 13 (2) (pp 239), 2017. Date of Publication: April 2017.

[Erratum]

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The authors regret that in the abovementioned article, the author list was incomplete. The correct author listing appears above.

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Institution

(Radmayr) Paediatric Urology, Medical University of Innsbruck, Innsbruck, Austria (Dogan, Tekgul) Hacettepe University, Faculty of Medicine, Department of Urology, Division of Paediatric Urology, Ankara, Turkey

(Hoebeke) Department of Urology, Ghent University Hospital, Gent, Belgium

(Kocvara) Department of Urology, General Teaching Hospital in Praha, Charles University 1st Faculty of Medicine, Praha, Czechia

(Nijman) Department of Urology, Division of Paediatric Urology, University of Groningen, Groningen, Netherlands

(Stein) Division of Paediatric Urology, Department of Urology, Mainz University Medical Centre, Johannes Gutenberg University, Mainz, Germany

(Undre) Department of Paediatric Urology, Great Ormond Street Hospital for Sick Children, London, United Kingdom

(Silay) Division of Pediatric Urology, Department of Urology, Istanbul Medeniyet University, Istanbul, Turkey

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354.

Semen quality of young men from the general population in Baltic countries.

Erenpreiss J., Punab M., Zilaitiene B., Hlevicka S., Zayakin P., Matulevicius V., Preiksa R.T., Jorgensen N.

Embase

Human Reproduction. 32 (6) (pp 1334-1340), 2017. Date of Publication: 01 Jun 2017.

[Article]

AN: 617718292

STUDY QUESTION: What are the parameters of semen quality in Baltic men? SUMMARY

ANSWER: Combined parameters of sperm concentration, motility and morphology revealed that 11-15% of men had low semen quality, 37-50% intermediate and 38-52% high semen quality.

WHAT IS KNOWN ALREADY: Previous studies have revealed regional differences in semen parameters, and semen quality of Baltic men has been suggested to be better than that of other European men. STUDY DESIGN, SIZE, DURATION: This was a cross-sectional study of 1165 men aged 16-29 years from Estonia (N = 573), Latvia (N = 278) and Lithuania (N = 314) conducted in 2003-2004. PARTICIPANTS/MATERIALS SETTING METHODS: Men from the general population, median age 19.8 years, provided one semen sample each, had blood samples taken, had testis size determined, and provided information on lifestyle. Based on combined data of sperm concentration, sperm motility and morphology the cohort was classified into three categories: low, intermediate or high semen quality. Comparisons between groups (including subgroups of Estonian men of Russian versus Estonian ethnicity) were tested, adjusting for ejaculation abstinence and age. MAIN RESULTS AND THE ROLE OF CHANCE: The median sperm concentration of the Estonian, Latvian and Lithuanian populations of Baltic men was 63 mill/ml. Low semen quality was detected in 11-15% of the men, intermediate in 37-50% and high in 38-52%. No crucial differences between national subgroups were detected, except that a higher percentage (9.6%) of the subgroup of Russian Estonians reported having had cryptorchidism compared to the other men (2.5-3.6%, $P < 0.001$). Smoking had an adverse impact on both sperm concentration and total sperm counts ($P < 0.001$). LIMITATIONS REASONS FOR CAUTION: The semen quality data were collected >10 years ago. Thus, a recent change in semen quality cannot be excluded. Owing to the study design, it is assumed, but unproven, that the men were representative of the general populations. Some men were very young (16 years), however, this was also the case for other European studies of similar populations. Assessment of sperm motility is associated with inter-observer variation, and no quality control was undertaken for sperm motility assessment to account for that. Thus, estimates of sperm motility should be interpreted with caution. WIDER IMPLICATIONS OF THE FINDINGS: Analysis of the semen variables separately did not identify that a considerable percentage of Baltic men had low semen quality. The combined analysis, however, showed that more than one out of nine men had semen quality at a level indicating reduced fertility chances. We suggest that future studies of semen quality should be carried out reporting both results of single semen parameters and estimates that combine the most frequently assessed variables.

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Status

Embase

Institution

(Erenpreiss, Zayakin) Latvian Biomedical Research and Study Center, Riga LV-1067, Latvia

(Punab) Andrology Center, Tartu University Hospital, Tartu 50406, Estonia

(Zilaitiene, Matulevicius, Preiksa) Institute of Endocrinology, Lithuanian University of Health Sciences, Kaunas LT-50009, Lithuania

(Hlevicka) Clinic 'Piramida', Riga LV-1012, Latvia

(Jorgensen) Department of Growth and Reproduction, Rigshospitalet, University of Copenhagen, Blegdamsvej 9, Copenhagen DK-2100, Denmark

(Jorgensen) Intl. Ctr. for Res. and Res. Training in Endocrine Disruption of Male Reproduction and Child Health, Rigshospitalet, Blegdamsvej 9, Copenhagen DK-2100, Denmark

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355.

Copy number variants of Ras/MAPK pathway genes in patients with isolated cryptorchidism.

Rodriguez F., Vallejos C., Giraudo F., Unanue N., Hernandez M.I., Godoy P., Celis S., Martin-Arenas R., Palomares-Bralo M., Heath K.E., Lopez M.T., Cassorla F.

Embase

Andrology. 5 (5) (pp 923-930), 2017. Date of Publication: September 2017.

[Article]

AN: 617453727

Cryptorchidism is the most common congenital disorder in boys, but the cause for most cases remains unknown. Patients with Noonan Syndrome are characterized by a typical face, growth retardation, congenital heart defects, learning disabilities and cryptorchidism. Copy number variations of Ras/MAPK pathway genes are unusual in patients with several clinical features of Noonan Syndrome; however, they have not been studied in patients with only one feature of this

condition, such as cryptorchidism. Our aim was to determine whether patients with isolated cryptorchidism exhibit Ras/MAPK pathway gene copy number variations (CNVs). Fifty-nine patients with isolated cryptorchidism and negative for mutations in genes associated with Noonan Syndrome were recruited. Determination of Ras/MAPK pathway gene CNVs was performed by Comparative Genome Hybridization array. A CNV was identified in two individuals, a ~175 kb microduplication at 3p25.2, partially including RAF1. A similar RAF1 microduplication has been observed in a patient with testicular aplasia. This suggests that some patients with isolated cryptorchidism may harbor Ras/MAPK pathway gene CNVs.

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Author NameID

Rodriguez F.; ORCID: <http://orcid.org/0000-0003-0461-7393>

Institution

(Rodriguez, Vallejos, Giraudo, Unanue, Hernandez, Cassorla) Institute of Maternal and Child Research, School of Medicine, University of Chile, Santiago, Chile (Godoy) Pediatric Service, Hospital Base San Jose, Osorno, Chile

(Celis, Lopez) Pediatric Urology Department, Hospital Clinico San Borja - Arriaran, Santiago, Chile

(Martin-Arenas, Palomares-Bralo, Heath) Institute of Medical and Molecular Genetics (INGEMM), Hospital Universitario La Paz, IdiPAZ, UAM and CIBERER, ISCIII, Madrid, Spain

Publisher

Blackwell Publishing Ltd

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2017

356.

Early childhood development of boys with genital anomalies.

Schneuer F.J., Bentley J.P., Holland A.J.A., Lain S.J., Jamieson S.E., Badawi N., Nassar N.

Embase

Birth Defects Research. 109 (8) (pp 535-542), 2017. Date of Publication: 2017.

[Article]

AN: 617403395

Background: Male genital anomalies often require surgery in early life to address functional and cosmetic consequences. However, there has been little assessment of developmental outcomes of affected boys.

Method(s): We conducted a population-based cohort study of all boys born in New South Wales, Australia, and undergoing school-entry developmental assessment in 2009 or 2012. Health and developmental information was obtained by means of record-linkage of birth, hospital and Australian Early Development Census data. Boys with hypospadias or undescended testis (UDT) were compared with those without. Developmental outcomes were assessed in five domains (physical health, emotional maturity, communication, cognitive skills, and social competence), and boys were categorized as vulnerable (<10th centile of national scores), developmentally high risk (DHR; vulnerable in 21 domains), and special needs.

Result(s): We included 420 boys with hypospadias, 873 with UDT, and 77,176 unaffected boys. There was no difference in the proportion of boys developmentally vulnerable in any domain or DHR between boys with hypospadias (DHR: n 5 49; 13.1%; p 5 0.9), UDT (n 5 116; 15.2%; p 5 0.06), and unaffected boys (n 5 9278; 12.9%). Compared with unaffected boys (n 5 4826; 6.3%), boys with hypospadias (n 5 43; 10.2%; p < 0.001) or UDT (n 5 105; 12.0%; p < 0.001) were more likely to have special needs. Stratified analyses revealed that only boys with UDT and coexisting anomalies had increased risk of being DHR (odds ratio: 2.65; 95% confidence interval, 1.61-4.36) or special needs (odds ratio: 2.91; 95% confidence interval, 2.00-4.22).

Conclusion(s): We found no increased risk of poor development among boys with hypospadias or UDT. However, boys with UDT and coexisting anomalies were more likely to have poorer development and special needs.

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Institution

(Schneuer, Bentley, Lain, Nassar) Menzies Centre for Health Policy, School of Public Health, University of Sydney, Sydney, Australia (Holland) Discipline of Paediatrics and Child Health, The Children's Hospital at Westmead, Sydney Medical School, The University of Sydney, NSW, Australia

(Jamieson) Telethon Kids Institute, The University of Western Australia, Crawley, WA, Australia

(Badawi) Grace Centre for Newborn Care, The Children's Hospital at Westmead, Sydney, Australia

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Year of Publication

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357.

Congenital hypogonadotropic hypogonadism, functional hypogonadotropism or constitutional delay of growth and puberty? An analysis of a large patient series from a single tertiary center.

Varimo T., Miettinen P.J., Kansakoski J., Raivio T., Hero M.

Embase

Human Reproduction. 32 (1) (pp 147-153), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 616972959

STUDY QUESTION What diagnoses underlie delayed puberty (DP) and predict its outcome?

SUMMARY ANSWER A multitude of different diagnoses underlie DP, and in boys a history of cryptorchidism, small testicular size and slow growth velocity (GV) predict its clinical course.

WHAT IS KNOWN ALREADY DP is caused by a variety of underlying etiologies. Hormonal markers can be used in the differential diagnosis of DP but none of them have shown complete diagnostic accuracy.

STUDY DESIGN, SIZE, DURATION Medical records of 589 patients evaluated for DP in a single tertiary care center between 2004 and 2014 were retrospectively reviewed.

PARTICIPANTS/MATERIALS, SETTING, METHODS Clinical and biochemical data of 174 boys and 70 girls who fulfilled the criteria of DP were included in the analyses. We characterized the frequencies of underlying conditions and evaluated the predictive efficacy of selected clinical and hormonal markers.

MAIN RESULTS AND THE ROLE OF CHANCE Thirty etiologies that underlie DP were identified. No markers of clinical value could be identified in the girls, whereas a history of cryptorchidism in the boys was associated with an increase in the risk of permanent hypogonadism (odds ratio 17.2 (95% CI; 3.4-85.4, $P < 0.001$)). The conditions that cause functional hypogonadotropic hypogonadism were more frequent in boys with a GV below 3 cm/yr than in those growing faster (19% vs 4%, $P < 0.05$). In this series, the most effective markers to discriminate the prepubertal boys with constitutional delay of growth and puberty

(CDGP) from those with congenital hypogonadotropic hypogonadism (CHH) were testicular volume (cut-off 1.1 ml with a sensitivity of 100% and a specificity of 91%), GnRH-induced maximal LH (cut-off 4.3 IU/L; 100%, 75%) and basal inhibin B (INHB) level (cut-off 61 ng/L; 90%, 83%). LIMITATIONS, REASONS FOR CAUTION The main limitation of the study is the retrospective design. WIDER IMPLICATIONS OF THE FINDINGS Prior cryptorchidism and slow GV are two important clinical cues that may help clinicians to predict the clinical course of DP in boys, whereas markers of similar value could not be identified in girls. In prepubertal boys, testicular size appeared as effective as INHB and GnRH-induced LH levels in the differential diagnosis between CHH and CDGP.

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Institution

(Varimo, Miettinen, Raivio, Hero) Children's Hospital, Pediatric Research Center, University of Helsinki, Helsinki, Finland (Miettinen) Research Programs Unit, Molecular Neurology, and Biomedicum Stem Cell Center, University of Helsinki, Helsinki, Finland

(Kansakoski, Raivio) Faculty of Medicine, Department of Physiology, University of Helsinki, Helsinki, Finland

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358.

Maternal use of mild analgesics during pregnancy associated with reduced anogenital distance in sons: A cohort study of 1027 mother-child pairs.

Lind D.V., Main K.M., Kyhl H.B., Kristensen D.M., Toppari J., Andersen H.R., Andersen M.S., Skakkebaek N.E., Jensen T.K.

Embase

Human Reproduction. 32 (1) (pp 223-231), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 616972953

STUDY QUESTION Is maternal use of mild analgesics in pregnancy associated with anogenital distance (AGD) - the distance from the anus to the genitals - in the offspring? **SUMMARY**

ANSWER Maternal use of mild analgesics [especially simultaneous use of paracetamol and nonsteroidal anti-inflammatory drugs (NSAIDs)] during pregnancy was associated with a shorter AGD in boys whereas no effect was found in girls. **WHAT IS KNOWN ALREADY** Mild analgesics including paracetamol (acetaminophen) and NSAIDs (e.g. ibuprofen and acetyl salicylic acid) have endocrine disrupting properties and in utero exposure reduces AGD in male rats. In humans, maternal exposure has been associated with cryptorchidism and hypospadias in male offspring but no studies have examined AGD. **STUDY DESIGN, SIZE, DURATION** A prospective birth cohort study. Between 2010 and 2012, 2500 pregnant women were recruited from the Odense Child Cohort. Children were examined 3 months after the expected date of birth.

PARTICIPANTS/MATERIALS, SETTING, METHODS Pregnant women were asked about use of medication including mild analgesics (paracetamol and NSAID) during pregnancy at recruitment (gestational age (GA) week 10-27) and at GA week 28. AGD and penile width were measured 3 months after expected date of birth by trained personnel. A total of 1027 women answered both questionnaires and their children were examined. Associations between prenatal exposure to mild analgesics and AGD and penile width were estimated using multivariable linear regression adjusting for age and weight-for-age SD score. **MAIN RESULTS AND THE ROLE OF CHANCE** A total of 40% of the women reported use of paracetamol and/or NSAIDs (4.4%) during the first 28 weeks of pregnancy. Exposure to analgesics during pregnancy was associated with a reduced AGD in boys, although statistically significant only for NSAIDs. The association was significant among 20 boys exposed to both paracetamol and NSAIDs (AGD -4.1 mm; CI 95%: -6.4; -1.7). Maternal intake of analgesics did not show any clear association with AGD in female offspring.

No effect on penile width was found. **LIMITATIONS REASONS FOR CAUTION** Only 27 boys and 18 girls were exposed to NSAIDs and most of them were also exposed to paracetamol. This makes it impossible to distinguish between exposures to NSAIDs alone and a potential mixture effect. Moreover, use of mild analgesics was self-reported up to 2 months after intake, which could have caused misclassification of exposure but is probably not associated with AGD as this was unknown to the women at time of reply to the questionnaire thereby underestimating the association. Confounding by indication may also explain our findings, as the condition for which the analgesic was taken may be associated with a reduction in AGD, rather than the use of the analgesic medication. This is the first study to report such an association in humans and further studies are needed to confirm our findings. **WIDER IMPLICATIONS OF THE FINDINGS** A negative association was observed between exposure to analgesics during pregnancy and AGD

in boys, suggesting disruption of androgen action. The health implications of a shorter AGD are still uncertain, but in cross-sectional studies among adult men a shorter AGD is associated with poorer semen quality and lower testosterone. As 41% of the women used these painkillers the finding are of public health importance and pregnant women should be advised about the potentially harmful effects of painkiller use.

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Embase

Institution

(Lind, Andersen, Jensen) Department of Environmental Medicine, Institute of Public Health, University of Southern Denmark, Odense, Denmark (Main, Skakkebaek) Department of Growth and Reproduction, Rigshospitalet, University of Copenhagen, Blegdamsvej 9, Copenhagen 2100, Denmark

(Kyhl, Jensen) Odense University Hospital, Hans Christian Andersen Children's Hospital, Sdr. Boulevard 29, Odense C 5000, Denmark

(Kyhl) Odense Patient data Exploratory Network (OPEN), Sdr. Boulevard 29, Odense C 5000, Denmark

(Kristensen) Laboratory of Genomic and Molecular Biomedicine, Department of Biology, University of Copenhagen, Universitetsparken 13, Copenhagen DK-2100, Denmark

(Toppari) Department of Physiology, Institute of Biomedicine, University of Turku, Kiinamylynkatu 10, Turku FI-20520, Finland

(Toppari) Department of Pediatrics, Turku University Hospital, Kiinamylynkatu 10, Turku FI-20520, Finland

(Andersen) Odense University Hospital, Endocrinological Department, Odense C 5000, Denmark

Publisher

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Causes of male infertility: A 9-year prospective monocentre study on 1737 patients with reduced total sperm counts.

Punab M., Poolamets O., Paju P., Vihljajev V., Pomm K., Ladva R., Korrovits P., Laan M.

Embase

Human Reproduction. 32 (1) (pp 18-31), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 616972952

STUDY QUESTION What are the primary causes of severe male factor infertility? SUMMARY ANSWER Although 40% of all patients showed primary causes of infertility, which could be subdivided into three groups based on the severity of their effect, ~75% of oligozoospermia cases remained idiopathic. WHAT IS KNOWN ALREADY There are few large-scale epidemiological studies analyzing the causes of male factor infertility. STUDY DESIGN, SIZE, DURATION A prospective clinical-epidemiological study was conducted at the Andrology Centre, Tartu University Hospital between 2005 and 2013, recruiting male partners of couples failing to conceive a child for over ≥ 12 months. Among 8518 patients, 1737 (20.4%) were diagnosed with severe male factor infertility. A reference group of fertile controls was comprised of 325 partners of pregnant women. PARTICIPANTS/MATERIALS, SETTING, METHODS The mean age of infertility patients and fertile controls was 33.2 +/- 7.3 and 31.7 +/- 6.3 years, respectively. All participants were examined using a standardized andrology workup, accompanied by a structured medical interview. Hormonal analysis included serum FSH, LH and testosterone. Semen quality was determined in accordance to the World Health Organization recommendations. Cases with spermatozoa concentrations of ≤ 5 million/ml were screened for chromosomal aberrations and Y-chromosomal microdeletions. MAIN RESULTS AND THE ROLE OF CHANCE The primary cause of infertility was defined for 695 of 1737 patients (~40%). The analyzed causal factors could be divided into absolute (secondary hypogonadism, genetic causes, seminal tract obstruction), severe (oncological diseases, severe sexual dysfunction) and plausible causal factors (congenital anomalies in uro-genital tract, acquired or secondary testicular damage). The latter were also detected for 11 (3.4%) men with proven fertility (diagnoses: Unilateral cryptorchidism, testis cancer, orchitis, mumps orchitis). The causal factors behind the most severe forms of impaired spermatogenesis were relatively well understood; causes were assigned: For aspermia in 46/46 cases (100%), for azoospermia in 321/388 cases (82.7%), and for cryptozoospermia in 54/130 cases (41.5%). In contrast, 75% of oligozoospermia cases remained unexplained. The main cause of aspermia was severe sexual dysfunction (71.7% of aspermia patients). Azoospermia patients accounted for 86.4% of all cases diagnosed with secondary hypogonadism and 97.1% of patients with seminal tract obstruction. Of patients with a known genetic factor, 87.4% had extreme infertility (azoo-, crypto- or aspermia). The prevalence

of congenital anomalies in the uro-genital tract was not clearly correlated with the severity of impaired sperm production. Previously defined 'potential contributing factors' varicocele and leukocytospermia were excluded as the primary causes of male infertility. However, their incidence was >2-fold higher (31.0 vs 13.5% and 16.1 vs 7.4%; $P < 0.001$) in the idiopathic infertility group compared to controls. In addition, the proportions of overweight (or obese) patients and patients suffering from a chronic disease were significantly increased in almost all of the patient subgroups. LIMITATIONS REASONS FOR CAUTION The study included only subjects with reduced total spermatozoa counts. Thus, these findings cannot be automatically applied to all male factor infertility cases. WIDER IMPLICATIONS OF THE FINDINGS The novel insights and improved clarity achieved in the comprehensive analysis regarding the absolute, causative and plausible factors behind male infertility, as well as the 'potential contributing factors', will be valuable tools in updating the current clinical guidelines. The study highlights knowledge gaps and reiterates an urgent need to uncover the causes and mechanisms behind, and potential treatments of, oligozoospermic cases, representing the majority of idiopathic infertility patients (86.3%).

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Embase

Institution

(Punab, Poolamets, Paju, Vihljajev, Pomm, Ladva, Korrovits) Andrology Center, Tartu University Hospital, Tartu 50406, Estonia (Laan) Human Molecular Genetics Research Group, Institute of Molecular and Cell Biology, University of Tartu, Tartu 51010, Estonia

Publisher

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2017

Analgesia use during pregnancy and risk of cryptorchidism: A systematic review and meta-analysis.

Gurney J., Richiardi L., McGlynn K.A., Signal V., Sarfati D.

Embase

Human Reproduction. 32 (5) (pp 1118-1129), 2017. Date of Publication: 01 May 2017.

[Review]

AN: 616190001

Study Question Are boys who are born to mothers who use analgesics during pregnancy at increased risk of cryptorchidism compared to those born to mothers who do not take analgesia?
Summary Answer In this systematic review and meta-analysis of 10 published studies, we observed only weak evidence of an association between analgesia use during pregnancy and risk of cryptorchidism in the son. **What is Known Already** Concentrations of analgesia relevant to human exposure have been implicated as causing endocrine disturbances in the developing foetal testis. However, when viewed collectively there appears to be conflicting evidence regarding an association between maternal use of analgesics and development of cryptorchidism.
Study Design, Size, Duration A systematic review and meta-analysis of studies on analgesia use during pregnancy and risk of cryptorchidism was performed. The search terms used were (analges* OR paracetamol OR acetaminophen) AND (cryptorchidism OR cryptorchism OR undescended test* OR non-descended test* OR non descended test*) for the databases Ovid Medline, Embase, Scopus and Web of Science. The search included all published articles up until 23 May 2016 and no limits were set in terms of language. **Participants/Materials, Setting, Methods** Abstracts were screened by one reviewer to remove irrelevant studies, with a 10% random sample of these verified by a second reviewer. The full text of all remaining papers was assessed by two reviewers. Abstracts included in the final analysis were studies which reported associations between the exposure (analgesia) and the outcome (cryptorchidism). Studies were only included if data were provided from which summary associations (odds ratios (ORs) or relative risks) and their 95% CIs could be calculated, or if summary associations were provided by the authors themselves. For each included study, two reviewers independently extracted study meta-data in line with PRISMA recommendations. We assessed study quality and potential for bias using the criteria outlined in the Newcastle-Ottawa Quality Assessment Scale, but did not determine a quality score. Two reviewers independently assessed study quality against these criteria. **Main Results and The Role of chance** After screening 350 manuscripts, 10 were included in our review (5 case-control studies, 5 cohort studies). We observed weak evidence of an association between ever use of analgesia and risk of cryptorchidism (pooled crude OR: 1.11, 95% CI: 1.00-1.23), with case-control studies revealing a marginally stronger association (1.23, 95% CI: 0.85-1.78) than cohort studies (1.09, 95% CI: 0.97-1.22). We observed weak evidence of a dose-response relationship between increasing weeks of analgesia exposure and risk of

cryptorchidism, as well as weak evidence of an effect of timing on analgesia exposure and risk of cryptorchidism. Assessment of study quality via the Newcastle-Ottawa criteria revealed little (if any) evidence of substantial bias that may have meaningfully affected a given study's results. Limitations, Reasons For Caution While confounding does not appear to be important, misclassification of the exposure is possibly an important source of measurement error in this context. The systematic review is open to reporting bias. Owing to scant data, no meta-analyses for two key questions (relating to dose-response and timing of exposure) could be performed. Medications were grouped based on their common effect and this offers little insight into the relation between specific types of analgesia and cryptorchidism. Finally, there are limitations in assuming that analgesia use reported by mothers is synonymous with actual intrauterine exposure. Wider Implications of The Findings The ubiquity of analgesia use during pregnancy makes this exposure particularly important from a population health perspective. About 9 of the 10 studies were conducted in Europe or USA, limiting generalizability of our observations. While the observations from our systematic review and meta-analysis suggest that analgesia use during pregnancy is not strongly associated with cryptorchidism development in the son, they also highlight the need for further detailed assessments of this relationship. Study Funding/Competing Interest(S) This study was funded by the Health Research Council of New Zealand (reference #: 14/052).

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Embase

Institution

(Gurney, Signal, Sarfati) Department of Public Health, University of Otago, 23a Mein St, Newtown, Wellington, New Zealand (Richiardi) Cancer Epidemiology Unit, University of Turin, Turin, Italy

(McGlynn) Division of Cancer Epidemiology and Genetics, National Cancer Institute, MS, United States

Publisher

Oxford University Press

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2017

361.

Maternal endometriosis and genital malformations in boys: a Danish register-based study.

Arendt L.H., Lindhard M.S., Henriksen T.B., Forman A., Olsen J., Ramlau-Hansen C.H.

Embase

Fertility and Sterility. 108 (4) (pp 687-693), 2017. Date of Publication: October 2017.

[Article]

AN: 618057208

Objective To investigate the association between maternal endometriosis and occurrence of the genital anomalies cryptorchidism and hypospadias in sons. Design Population-based cohort study. Setting Not applicable. Patient(s) All live-born singleton boys born from 1978 to 2012. Intervention(s) None. Main Outcome Measure(s) Cryptorchidism and hypospadias in boys based on information from the Danish National Patient Register. Result(s) The study included 1,073,026 live-born singleton boys. A total of 6,443 boys were sons of women diagnosed with endometriosis before pregnancy. Altogether, 27,342 boys were diagnosed with cryptorchidism, of whom 16,446 had corrective surgery. Hypospadias was diagnosed in 4,853 boys. As compared with unexposed boys, a tendency towards a slightly higher occurrence of cryptorchidism was observed among boys of women with endometriosis (adjusted hazard ratio [aHR] 1.18; 95% confidence interval [CI], 0.97, 1.44). When stratified by medically assisted reproduction (MAR) technologies, the association was slightly stronger among boys born to women with endometriosis who had conceived via MAR, yet it remained moderate and statistically insignificant (aHR 1.27; 95% CI, 0.97, 1.70). When women who conceived with MAR were excluded, the association between endometriosis and cryptorchidism disappeared. For hypospadias, we observed no association, either in the main analysis or the stratified analysis. Conclusion(s) The findings from this register-based study do not provide strong evidence for a higher occurrence of the studied genital anomalies among boys of women with endometriosis.

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Status

Embase

Author NameID

Arendt, Linn Hakonsen; ORCID: <http://orcid.org/0000-0001-9692-3501> Olsen, Jorn; ORCID:

<http://orcid.org/0000-0001-7462-2740>

Institution

(Arendt, Ramlau-Hansen) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus, Denmark (Arendt, Lindhard, Henriksen) Perinatal Epidemiology Research Unit, Department of Child and Adolescent Health, Aarhus University Hospital, Aarhus, Denmark (Forman) Department of Gynaecology and Obstetrics, Aarhus University Hospital, Aarhus, Denmark (Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark
Publisher
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Year of Publication
2017

362.

Maternal Overweight and Obesity and Genital Anomalies in Male Offspring: A Population-Based Swedish Cohort Study.

Arendt L.H., Ramlau-Hansen C.H., Lindhard M.S., Henriksen T.B., Olsen J., Yu Y., Cnattingius S.
Embase

Paediatric and Perinatal Epidemiology. 31 (4) (pp 317-327), 2017. Date of Publication: July 2017.
[Article]

AN: 617012803

Background: Overweight and obese pregnant women face higher risk of several critical birth outcomes, including an overall increased risk of congenital abnormalities. Only few studies have focused on associations between maternal overweight and the genital anomalies in boys, cryptorchidism and hypospadias, and results are inconclusive.

Method(s): We performed a population-based cohort study and assessed the associations between maternal body mass index (BMI) in early pregnancy and occurrence of cryptorchidism and hypospadias. All live-born singleton boys born in Sweden from 1992 to 2012 were included. From the Swedish Patient Register, information on cryptorchidism and hypospadias was available. Data were analysed using Cox proportional hazards regression adjusted for potential confounders. Mediation analyses were performed to estimate how much of the association between BMI and genital anomalies were mediated through obesity-related diseases.

Result(s): Of the 1 055 705 live-born singleton boys born from 1992 to 2012, 6807 (6.4 per 1000) were diagnosed with hypospadias and 16 469 (15.6 per 1000) were diagnosed with

cryptorchidism, of which 9768 (9.3 per 1000) underwent corrective surgery for cryptorchidism. We observed dose-response associations between maternal BMI and hypospadias and cryptorchidism. Boys of mothers with BMI ≥ 40.0 kg/m² had the highest adjusted hazard ratios for hypospadias (HR 1.35, 95% confidence interval [CI] 1.04, 1.76) and cryptorchidism (HR 1.25, 95% CI 1.00, 1.58). A substantial proportion of the associations between BMI and the genital anomalies were mediated through preeclampsia.

Conclusion(s): This large register-based study adds to the current literature and indicates that the occurrence of hypospadias and cryptorchidism increase with maternal overweight and obesity severity.

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Author NameID

Arendt, Linn Hakonsen; ORCID: <http://orcid.org/0000-0001-9692-3501> Cnattingius, Sven;

ORCID: <http://orcid.org/0000-0002-0805-8093>

Institution

(Arendt, Ramlau-Hansen, Olsen) Department of Public Health, Section for Epidemiology, Aarhus University, Aarhus, Denmark (Arendt, Lindhard, Henriksen) Perinatal Epidemiology Research Unit, Department of Child and Adolescent Health, Aarhus University Hospital, Aarhus, Denmark (Olsen, Yu) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark (Cnattingius) Clinical Epidemiology Unit, Department of Medicine, Karolinska University Hospital, Stockholm, Sweden

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363.

Rural versus urban compliance in the management of cryptorchidism: is there a difference?.

Point D., Morley C., Turchi A., Reddy S., Sirisreetreerux P., Gearhart J., Al-Omar O.

Embase

European Journal of Pediatrics. 176 (8) (pp 1067-1073), 2017. Date of Publication: 01 Aug 2017.

[Article]

AN: 616957113

The authors compared the age and referral patterns of pediatric patients undergoing surgical intervention for cryptorchidism at a rural, West Virginia University, versus urban, Johns Hopkins University, tertiary center. A retrospective review of patients undergoing surgical evaluation for cryptorchidism was performed. Patients treated for reasons unrelated to cryptorchidism or referred for multiple urologic diagnoses were excluded. The patients at each institution were then divided into four groups based on their corrected gestational age at time of surgery. Referral times and provider specialties were also obtained. A total of 131 cases at the urban center and 100 cases at the rural center were identified. At the rural center, the average age of referral and surgery were 48.3 and 53.8 months, respectively, compared to 59.6 and 65.2 months at the urban center. Only 40% of patients at the rural site and 29% at the urban institution underwent intervention at less than 18 months of age. There was no significant difference in time of referral to surgery between the institutions. The majority of referrals were made by private practice pediatricians.

Conclusion(s): In this study, a pattern of delayed referral and intervention was observed at both institutions despite differing geographic regions and heterogeneous patient populations. It is important that referring providers realize that scrotal U/S does not change management of UDT and should not delay prompt referral. What is known: * Significant referral delay is a challenging issue in the management of cryptorchidism. * Ultrasound is not a valid method for the detection of cryptorchidism. What is new: * The rural and urban management of cryptorchidism is not that different. * More emphasis should be put on the detection management of cryptorchidism.

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Institution

(Point, Morley, Al-Omar) Department of Urology, West Virginia University School of Medicine, Morgantown, WV, United States (Tourchi, Reddy, Sirisreerux, Gearhart) Division of Pediatric Urology, James Buchanan Brady Urological Institute, The Johns Hopkins University School of Medicine, Baltimore, MD, United States

(Al-Omar) Department of Surgery, Division of Urology, West Virginia University Robert C. Byrd Sciences Center, P.O. Box 9238, 6300 HSS, Morgantown, WV 26506-9238, United States

Publisher

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Year of Publication

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364.

Choice of Repairing Inguinal Hernia in Children: Open Versus Laparoscopy.

Raveenthiran V., Agarwal P.

Embase

Indian Journal of Pediatrics. 84 (7) (pp 555-563), 2017. Date of Publication: 01 Jul 2017.

[Article]

AN: 616479734

Inguinal hernia is a common disorder of childhood that requires surgical repair at diagnosis. Traditionally, it is operated upon by open inguinal incision. However, with the introduction of laparoscopic repair in 1990, opinion of scientific community is divided concerning the best method of pediatric herniotomy. Educated parents, who long to have the choicest of the best, often gather information from internet and prefer to discuss their concerns with primary care physicians. This descriptive review is intended to provide practicing pediatricians with updated evidence-based information which will enable them to counsel parents regarding the choice of hernia repair. Based on careful analysis of current literature, unacceptable standards are defined in this paper and rationalized recommendations are proposed. Laparoscopy appears to be beneficial in bilateral hernia of girls, giant hernia, recurrence following failed open repair and in hernia associated with undescended testis or ambiguous genitalia. On the other hand, open herniotomy appears to be advantageous in male inguinal hernia, unilateral female hernia, premature newborns, failed laparoscopic repair and in hernia associated with serious co-morbidity.

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Status

Embase

Institution

(Raveenthiran) Department of Pediatric Surgery, Sri Ramasamy Memorial (SRM) Medical College, Kattankulathur, Chennai, Tamil Nadu, India (Agarwal) Department of Pediatric Surgery, Sri Ramachandra Medical College, Porur, Chennai, Tamil Nadu, India

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365.

Association of reproductive disorders and male congenital anomalies with environmental exposure to endocrine active pesticides.

Gonzalez J.G., Miranda M.I.V., Mullor M.R., Jerez A.F.H., Carreno T.P., Rodriguez R.A.

Embase

Reproductive Toxicology. 71 (pp 95-100), 2017. Date of Publication: 01 Aug 2017.

[Article]

AN: 616107670

There is growing evidence that environmental exposure to pesticides may increase the risk of developing reproductive and developmental disorders. This study determined the prevalence and risk of developing gestational disorders and male congenital genitourinary malformations in areas with distinct exposure to pesticides, many of them with potential endocrine disrupting properties. A population-based case-control study was carried out on pregnant women and male children living in ten health districts of Andalusia classified as areas of high and low environmental exposure to pesticides according to agronomic criteria. The study population included 45,050 cases and 950,620 controls matched for age and health district. Data were collected from computerized hospital records between 1998 and 2005. Prevalence rates and risk of miscarriage, low birth weight, hypospadias, cryptorchidism and micropenis were significantly greater in areas with higher use of pesticides in relation to those with lower use, thus supporting and extending previous information.

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Institution

(Gonzalez) Rafael Mendez Hospital Lorca, Murcia (Spain) and University of Lorca, Spain

(Miranda, Mullor, Carreno, Rodriguez) University of Almeria, Department of Neurosciences and Health Sciences, Almeria, Spain

(Jerez) University of Granada School of Medicine, Granada, Spain

(Carreno) Andalusian Council of Health at Almeria Province, Almeria, Spain

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366.

INSL3 and AMH in patients with previously congenital or acquired undescended testes.

van Brakel J., de Muinck Keizer-Schrama S.M.P.F., Hazebroek F.W.J., Dohle G.R., de Jong F.H.

Embase

Journal of Pediatric Surgery. 52 (8) (pp 1327-1331), 2017. Date of Publication: August 2017.

[Article]

AN: 616031326

Background In previous reports no differences in Leydig and Sertoli cell function were found between congenital undescended testis (CU DT) and acquired UDT (AUDT) on the basis of serum levels of LH, testosterone, FSH or inhibin B. This study tried to detect differences in Leydig and Sertoli cell function between CU DT and AUDT using insulin-like peptide 3 (INSL3) and anti-Mullerian hormone (AMH). Method 118 men with a history of UDT (CU DT N = 55 (6/55 bilateral), AUDT N = 63 (15/63 bilateral)) were investigated. Differences between CU DT and AUDT, influence of age at surgery in CU DT, and effect of spontaneous descent or orchiopexy in AUDT were evaluated. Results For INSL3, no significant differences were found. AMH levels in bilateral CU DT were significantly lower compared with bilateral AUDT (6.4 (1.7-11.4) vs 13.2 (6.1-30.1) mug/l, p = 0.02). AMH levels in unilateral CU DT were significantly higher than in bilateral CU DT (12.1 (2.4-43.7) vs. 6.4 (1.7-11.4) mug/l, p = 0.02). Conclusion No differences in Leydig cell function on the basis of INSL3 levels between the different UDT groups were found. Sertoli cell

function evaluated by AMH, was more negatively affected in bilateral CUDT in comparison with bilateral AUDT and unilateral CUDT. Level of evidence rating Level III Treatment Study.

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Institution

(van Brakel, Dohle) Department of Urology, Erasmus MC, P.O. Box 2040, Rotterdam 3000 CA, Netherlands (de Muinck Keizer-Schrama) Department of Paediatrics, Endocrinology, Erasmus MC-Sophia Children's Hospital, P.O. Box 2040, Rotterdam 3000 CA, Netherlands

(Hazebroek) Department of Paediatric Surgery, Erasmus MC-Sophia Children's Hospital, P.O. Box 2040, Rotterdam 3000 CA, Netherlands

(de Jong) Department of Internal Medicine, Section of Endocrinology, Erasmus MC, P.O. Box 2040, Rotterdam 3000 CA, Netherlands

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367.

Blind ending vessels on diagnostic laparoscopy for nonpalpable testis: Is a nubbin present?.

Sturm R., Kurzrock E., Amend G., Shannon R., Gong E., Cheng E.

Embase

Journal of Pediatric Urology. 13 (4) (pp 392.e1-392.e6), 2017. Date of Publication: August 2017.

[Article]

AN: 617221074

Introduction The traditional management paradigm for nonpalpable testis (NPT) has been that inguinal or scrotal exploration for a nubbin may be omitted when blind ending vessels are observed during diagnostic laparoscopy. Our aim was to examine whether blind ending vessels excluded the presence of a nubbin in a series of boys who underwent exploration in this setting.

Materials and methods Using a surgical database and chart review, pre-pubertal boys (<=12

years) with the diagnosis of undescended or atrophic testis who underwent a diagnostic laparoscopy for unilateral NPT between 2000 and 2015 were retrospectively identified. Physical exam, procedural and pathologic findings were confirmed by chart review. Results 595 boys underwent diagnostic laparoscopy for NPT by 11 surgeons. Of these, 318 had an intra-abdominal testis and 18 underwent diagnostic laparoscopy alone. Of the remaining 259, 32 had an open internal ring and inguinal or scrotal exploration was performed. The remaining 227 with a closed ring comprised the cohort for our analysis, of whom 188 had vessels entering the ring, 36 had blind ending vessels, and in three the vessel status was unavailable. In the 188 boys with vessels entering the ring, 164 (87%) had a nubbin excised during inguinal or scrotal exploration, of which 93% were grossly identified as an atrophic testis. Pathology confirmed the presence of hemosiderin in 44% and calcifications in 54%. In the 36 boys with blind ending vessels, 26 (72%) had a nubbin excised during inguinal or scrotal exploration, of which 96% were grossly identified as an atrophic testis. Pathology confirmed hemosiderin in 54% and calcifications in 58%. All seven cases with both blind ending vas and vessels had an atrophic testis grossly identified. Of all 207 excised remnants in this series, nubbins with viable testicular elements (seminiferous tubules in 11, germ cells in two) were only excised during cases that reported a non-atretic vas or any vessels entering the internal ring. Conclusion In this large multi-institutional series, blind ending vessels were associated with a nubbin noted during inguinal or scrotal exploration in the majority of cases. Based on this series if the surgeon's goal is to remove all nubbins, exploration is necessary regardless of vessel appearance. However, viable testicular elements were rarely identified and only when either a non-atretic vas or any vessels were observed to enter the ring.[Figure presented]

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Author NameID

Sturm, Renea; ORCID: <http://orcid.org/0000-0001-8030-5637>

Institution

(Sturm, Shannon, Gong, Cheng) Division of Urology, Ann and Robert H. Lurie Children's Hospital, Chicago, IL, United States (Kurzrock, Amend) Department of Urology, University of California Davis, Sacramento, CA, United States

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368.

Practical approach to evaluating testicular status in infants and children. Approche pratique a l'evaluation des testicules chez les nourrissons et les enfants <Approche pratique a l'evaluation des testicules chez les nourrissons et les enfants.>

Mau E.E., Leonard M.P.

Embase

Canadian Family Physician. 63 (6) (pp 432-435 and 442-445), 2017. Date of Publication: 01 Jun 2017.

[Review]

AN: 616835380

Objective To review the differences between normal, retractile, ectopic, ascended, and undescended testes and to describe the optimal way to perform a testicular examination to distinguish one from the other, as well as to demonstrate that ultrasound imaging is not necessary and to clarify when to consider specialist referral. **Sources of information** This paper is based on selected findings from a MEDLINE search on undescended testes and orchiopexy referrals, and on our experience at the Urology Clinic at the Children's Hospital of Eastern Ontario in Ottawa, including review of referrals to our clinic for undescended testes and the resultant findings of normal variants versus surgical cases. **The MeSH headings used in our MEDLINE search included** undescended testicle, retractile testicle, ectopic testicle, ascended testicle, referral and consultation, and orchiopexy. **Main message** An undescended testis is defined as the true absence of one testis (or both testes) from normal scrotal position. Ectopic and ascended testes will likewise be absent from the scrotum, the latter having been present at one point in development. Differentiating among testicular examination findings is important, as descended and retractile testes are managed conservatively, while prompt surgical intervention should be offered for ascended, ectopic, and undescended testes. Uncertainty surrounding the diagnosis of an undescended testis causes anxiety, might lead to unwarranted imaging, and might increase the wait list for specialty assessment. For this reason, avoidance of ultrasound in the evaluation of undescended testes was included in the recent Choosing Wisely Canada campaign. We seek to clarify the physical examination findings in the evaluation of possible undescended testes, the suggested referral parameters, and the subsequent management. **Conclusion** Undescended testes and their variants are common. As decision for referral is based on the primary care

physician's physical examination findings, we clarify distinguishing between normal and abnormal findings on testicular examination to aid in appropriate referral for subspecialist evaluation.

Consultation, if needed, should be sought at 6 months' corrected gestational age, or at detection if later than 6 months, without delay for ultrasound imaging, as surgical management is recommended for those patients with undescended, ectopic, or ascended testes.

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Institution

(Mau) Pediatric Urology, Royal University Hospital, Saskatoon, Sask, Canada (Leonard)
Department of Surgery in Pediatric Urology, Children's Hospital of Eastern Ontario, Ottawa, Canada

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369.

Health Disparities in the Appropriate Management of Cryptorchidism.

Savoie K.B., Bachier-Rodriguez M., Schurtz E., Tolley E.A., Giel D., Feliz A.

Embase

Journal of Pediatrics. 185 (pp 187-192.e1), 2017. Date of Publication: June 2017.

[Article]

AN: 615342262

Objective To assess regional practices in management of cryptorchidism with regard to timely fixation by the current recommended age of 18 months. Study design A retrospective study was performed. Charts of all patients who underwent surgical correction for cryptorchidism by a pediatric general surgeon or urologist within a tertiary pediatric hospital in an urban setting were systematically reviewed. Results We identified 1209 patients with cryptorchidism. The median age of surgical correction was 3.7 years (IQR: 1.4, 7.7); only 27% of patients had surgical correction before 18 months of age. Forty-six percent of our patients were white, 40% were

African American, and 8% were Hispanic. African American and Hispanic patients were less likely to undergo timely repair ($P = .01$), as were those with public or no insurance ($P < .0001$). A majority (72%) of patients had no diagnostic imaging prior to surgery. A majority of patients had palpable testes at operation (85%) and underwent inguinal orchiopexy (76%); 82% were operated on by a pediatric urologist. Only 35 patients (3%) experienced a complication; those repaired late were significantly less likely to develop a complication ($P = .03$). There were no differences in age at time of surgery by surgeon type. Conclusions A majority of our patients were not referred for surgical intervention in a timely manner, which may reflect poor access to care in our region. Public and self-pay insurance status was associated with delayed repair. Education of community physicians and families could be potentially beneficial.

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Status

Embase

Author NameID

Savoie, Kate B.; ORCID: <http://orcid.org/0000-0002-0960-333X>

Institution

(Savoie, Bachier-Rodriguez, Feliz) Department of General Surgery, University of Tennessee Health Science Center, Memphis, TN, United States (Savoie, Bachier-Rodriguez) Le Bonheur

Foundation, Le Bonheur Children's Hospital, Memphis, TN, United States

(Schurtz, Giel) Department of Urology, University of Tennessee Health Science Center, Memphis, TN, United States

(Tolley) Department of Preventive Medicine, University of Tennessee Health Science Center, Memphis, TN, United States

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A population-based study of prevalence trends and geospatial analysis of hypospadias and cryptorchidism compared with non-endocrine mediated congenital anomalies.

Lane C., Boxall J., MacLellan D., Anderson P.A., Dodds L., Romao R.L.P.

Embase

Journal of Pediatric Urology. 13 (3) (pp 284.e1-284.e7), 2017. Date of Publication: June 2017.

[Article]

AN: 615003876

Introduction Several reports have suggested an increase in the prevalence of hypospadias and cryptorchidism over the last few decades. Endocrine disruption caused by exposure to environmental chemicals has been postulated as a possible cause. **Objectives** The objectives of our study were: 1) to determine whether the prevalence of hypospadias and cryptorchidism is increasing compared with other congenital anomalies not known to be mediated by endocrine factors; and 2) to perform a geospatial analysis of these congenital malformations looking for clustering that could offer insight into environmental risk factors. **Material and methods** Data were obtained from the Nova Scotia ATLEE Perinatal Database containing the perinatal records of all live births in Nova Scotia, Canada since 1988. Records from 1988 to 2013 defined the study cohort. Overall prevalence rates and prevalence trends by year were calculated for hypospadias, cryptorchidism, gastroschisis, and clubfoot. County of residence was collected and spatial autocorrelation testing for clustering was performed for each of the congenital anomalies. **Results** There were 258,147 live births during the study period. Overall prevalence rates for the four malformations over the study period were: hypospadias 78 per 10,000 male births, cryptorchidism 75 per 10,000 male births, clubfoot 24 per 10,000 total births, and gastroschisis 4 per 10,000 total births. Incidence rate ratios per year for hypospadias, cryptorchidism, clubfoot, and gastroschisis were 1.00 (0.99-1.01), 0.99 (0.98-1.00), 0.98 (0.97-0.99), and 1.04 (1.04-1.07), respectively. During the study period, the prevalence rates in the region were unchanged for hypospadias, slightly reduced for cryptorchidism and clubfoot, and rising for gastroschisis (Figure). Spatial autocorrelation testing revealed statistically significant clustering for hypospadias ($p = 0.03$) and cryptorchidism ($p = 0.03$), while no spatial autocorrelation was observed for the other malformations. **Discussion** Contrary to previous studies we show that hypospadias and cryptorchidism prevalence rates are not increasing over time in our region. Nonetheless, rates for these conditions in our area are high compared with other regions of the world. Local clustering of these congenital anomalies without clustering of the control, non-endocrine mediated congenital malformations supports a possible unique spatial distribution associated with environmental exposure. The hotspots identified for hypospadias and cryptorchidism are associated with intense agricultural activity. **Conclusions** Our study found no increase in hypospadias and cryptorchidism prevalence over a 26-year period compared with other congenital anomalies not known to be

associated with endocrine factors. Geospatial analysis supports high clustering for hypospadias and cryptorchidism in areas of intense agricultural activity.[Figure presented]

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Embase

Institution

(Lane, MacLellan, Anderson, Romao) Division of Urology, Department of Surgery, IWK Health Centre, Department of Urology, Faculty of Medicine, Dalhousie University, Halifax, NS, Canada

(Boxall) Department of Earth Sciences, Dalhousie University, Halifax, NS, Canada

(Dodds) Perinatal Research Epidemiology Unit, IWK Health Centre, Department of Pediatrics and Obstetrics/Gynecology, Dalhousie University, Halifax, NS, Canada

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371.

Scrotal testis size in unilateral non-palpable cryptorchidism, what it can and cannot tell: Study of a Middle Eastern population.

Shadpour P., Kashi A.H., Arvin A.

Embase

Journal of Pediatric Urology. 13 (3) (pp 268.e1-268.e6), 2017. Date of Publication: June 2017.

[Article]

AN: 614487928

Background Predicting the fate of a unilateral non-palpable testis based on its scrotal counterpart has been recommended by some, yet disputed by others, and the question remains open.

Objective To investigate the accuracy of contralateral testis hypertrophy in predicting the absence of a unilateral non-palpable testis in a Middle Eastern population. Study design This retrospective study included all patients referred to the present institution with unilateral non-palpable testis between June 2010 and August 2014, who had undergone laparoscopy. The scrotal testis was

examined by sonography for size and volume, and diagnostic laparoscopy was utilized to determine the state of the cryptorchid testis. Results Of the 135 referred patients, 64 were aged ≤ 8 years, 29 were 9-18 years, and 42 were >18 years old. Diagnostic laparoscopy revealed 63 intra-abdominal testes, 20 small intra-inguinal testes, 32 vanished testes, and 20 nubbins or aplasia (Summary fig.). Scrotal testis volume was only a modest predictor for absence of the contralateral gonad in adult patients in whom a 22 ml cut-off yielded 64.3% sensitivity and 92.9% specificity. For those aged <18 years, overall accuracy was poor and dropped below 60%. Relative enlargement of contralateral testis in decreasing order of size was observed in patients with primary monorchism, followed by those with secondarily atrophic or nubbin testis, and then those with normal sized inguinal or abdominal testis. Discussion Unlike some previous series, which based their conclusions upon open exploration and mostly studied pre-pubertal subjects, the present results exclusively from laparoscopic exploration suggested that contralateral testis volume is a poor and inconsistent predictor of monorchism in children, and marginally predictive for young adults with unilateral non-palpable testis. This study comprised a reasonably large overall sample size compared with preceding reports; however, the number of patients within in each age group was limited. Greater numbers could allow for statistical comparison stratified by age group, for which this study was not powered. Conclusion Contralateral testis volume predicts, with modest accuracy, monorchism in adults with unilateral non-palpable testis. In younger patients, the overall predictive accuracy of scrotal testis size is poor and not consistently dependable.[Figure presented]

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Status

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Author NameID

Shadpour P.; ORCID: <http://orcid.org/0000-0003-4171-4168> Kashi A.H.; ORCID:

<http://orcid.org/0000-0002-7165-7101>

Institution

(Shadpour, Kashi, Arvin) Hasheminejad Kidney Center (HKC), Hospital Management Research Center (HMRC), Iran University of Medical Sciences (IUMS), Tehran, Iran, Islamic Republic of

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372.

Positive Oct-3/4 and D2-40 Immunohistochemical Expression in Germ Cells and Suspected Histology Pattern of Intratubular Germ Cell Neoplasia in Boys with Cryptorchidism Vanish after the Age of 2 Years.

Thorup J., Clasen-Linde E., Cortes D.

Embase

European Journal of Pediatric Surgery. 27 (4) (pp 313-318), 2017. Date of Publication: 01 Aug 2017.

[Article]

AN: 612123582

Introduction Intratubular germ cell neoplasia (ITGCN) is a precursor to testicular germ cell cancer. Adult germ cell cancer immunohistochemical markers may fail to detect ITGCN in prepubertal boys with congenital cryptorchidism, because positive immunohistochemistry is commonly seen in boys younger than the age of 2 years, where most orchiopexies are performed. The aim of the study was to evaluate the diagnostic challenge to differentiate between a histological pattern of ITGCN and a histological pattern with some atypical germ cells and all positive cancer immunohistochemical markers, but no increased risk of malignancy. Materials and Methods Histology sections from 373 testicular biopsies from 289 boys aged 1 month to 2 years operated for cryptorchidism were incubated with primary antibodies including anti-placental-like-alkaline phosphatase, anti-Oct-3/4, anti-C-kit, anti-D2-40, and in case of repeat biopsy with anti-stem cell factor (SCF) receptor. Results The prevalence of Oct-3/4 and D2-40-positive staining of germ cells in testicular biopsies were in age groups less than 6 months, 100% and 50%; 6-12 months, 60% and 17%; and 1-2 years, 12% and 4%. A 1 year, 1-month-old boy with Prader-Willi syndrome treated with growth hormone had ITGCN in both cryptorchid testes. In another three bilateral nonsyndromic cases, 8 months, 8 months and 1-year-old, a histological pattern in accordance with ITGCN was found. These three boys had a repeat biopsy from both testes performed at the age of 3 years, 4 months, 3.5 years, and 3 years, 10months, respectively. In all cases, the Oct-3/4 and D2-40 positive germ cells turned negative and the histological pattern normalized completely. The primary biopsies had SCF negative germ cells. Conclusion This study is valuable in identifying the age-related change in Oct-3/4 or D2-40 immunopositive germ cells in seminiferous tubules. An ITGCN-like histological pattern in nonsyndromic cryptorchidism will vanish after the age of 3 years. Even when immunohistochemistry is applied, prepubertal

ITGCN is so rarely demonstrated in cryptorchid testes, that it is not plausible that ITGCN generally originates during fetal development in cryptorchidism.

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Institution

(Thorup, Cortes) Department of Pediatric Surgery, Rigshospitalet, University of Copenhagen, Copenhagen DK-2100, Denmark (Thorup, Cortes) Institute for Clinical Medicine, Faculty of Health and Medical Science, University of Copenhagen, Denmark

(Clasen-Linde) Department of Pathology, Rigshospitalet, Copenhagen, Denmark

(Cortes) Section of Endocrinology, Department of Pediatrics, Hvidovre Hospital, Copenhagen, Denmark

Publisher

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Year of Publication

2017

373.

Sperm DNA damage measured by sperm chromatin structure assay in men with a history of undescended testes.

van Brakel J., Dinkelman-Smit M., de Muinck Keizer-Schrama S.M.P.F., Hazebroek F.W.J., Dohle G.R.

Embase

Andrology. 5 (4) (pp 838-843), 2017. Date of Publication: July 2017.

[Article]

AN: 616983031

The aim of this study was to compare sperm DNA damage between men with a history of congenital undescended testis (UDT) and men with a history of acquired UDT. A long-term follow-up study of men with previous UDT was performed. Fifty men with congenital UDT who had undergone orchiopexy at childhood age, 49 men with acquired UDT after a 'wait-and-see'-

protocol (e.g. awaiting spontaneous descent until puberty and perform an orchiopexy in case of non-descent), and 22 healthy proven fertile men were included. The DNA fragmentation index (DFI) using sperm chromatin structure assay (SCSA) was used to express the level of sperm DNA damage. Decreased fertility potential was considered if DFI was above 30%. Sperm DNA damage was not statistically different between cases of congenital and acquired UDT. DFI was significantly more often >30% in the complete group of men with congenital UDT (9/50; 18%) and in the subgroup with bilateral congenital UDT (3/7; 43%) in comparison with the controls (none) (p-value 0.049 and 0.01, respectively). Age at orchiopexy in congenital UDT had no statistical effect on DNA damage. In men with acquired UDT, DFI did not statistically differ between those having undergone orchiopexy and those experiencing spontaneous descent. This study supports the hypothesis that UDT is a spectrum representing both congenital UDT and acquired UDT. Sperm DNA damage at adult age is not influenced by age at orchiopexy in congenital UDT cases and by orchiopexy or spontaneous descent in acquired UDT cases.

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Status

Embase

Author NameID

van Brakel J.; ORCID: <http://orcid.org/0000-0002-0394-4723>

Institution

(van Brakel, Dinkelman-Smit, Dohle) Urology, Erasmus MC, Rotterdam, Netherlands (de Muinck Keizer-Schrama) Pediatrics, Endocrinology, Erasmus MC-Sophia Children's Hospital, Rotterdam, Netherlands

(Hazebroek) Paediatric Surgery, Erasmus MC-Sophia Children's Hospital, Rotterdam, Netherlands

Publisher

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Altered secretion of Sertoli cells hormones in 2-year-old prepubertal cryptorchid boys: a cross-sectional study.

Hamdi S.M., Almont T., Galinier P., Mieusset R., Thonneau P.

Embase

Andrology. 5 (4) (pp 783-789), 2017. Date of Publication: July 2017.

[Article]

AN: 616410237

In cryptorchid boys, failures in germ cell development have been clearly established. Some studies reported some abnormalities in Sertoli cells morphology but the results regarding their endocrine secretion remain controversial. To compare testicular hormone levels in young boys with and without cryptorchidism, we performed a cross-sectional hospital-based study. From surgery appointment records, we identified a case group of boys with unilateral or bilateral cryptorchidism and a control group undergoing dental care, minor osteoarticular or dermal surgery. Blood samples were withdrawn during the surgical procedure to perform testosterone, inhibin B and anti-mullerian hormone (AMH) immunoassays. We included 27 cryptorchid boys and 27 controls aged of 26.6 vs. 24.2 months, respectively ($p = 0.172$) far from the post-natal mini-puberty and the corresponding hormonal surges. Age-adjusted AMH and inhibin B levels were significantly lower in cryptorchid than in control boys (AMH: 87 ng/mL vs. 135 ng/mL; $p = 0.009$, inhibin B: 97 pg/mL vs. 133 pg/mL; $p = 0.019$, respectively). Moreover, AMH and inhibin B levels were significantly lower in the bilateral cryptorchid subgroup, being 50% lower than in the controls ($p = 0.011$ and 0.019 , respectively) and while both hormones levels were independent in controls, they became strongly correlated in bilateral cryptorchid boys ($R^2 = 0.75$, $p = 0.001$). In addition, testosterone levels were still detectable in some boys, with significantly lower levels in cryptorchid group than in controls. Overall, 2-year-old cryptorchid patients presented a simultaneous and significant drop in AMH and inhibin B levels, suggesting a functional defect of Sertoli cells. This deficiency appeared more pronounced in bilateral cryptorchidism and thus, regarding the pivotal role of Sertoli cells in germ cell development, it may explain the compromised fertility found later in men born with such a malformation.

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Status

Embase

Author NameID

Hamdi S.M.; ORCID: <http://orcid.org/0000-0002-2434-3441>

Institution

(Hamdi, Almont, Mieusset, Thonneau) EA 3694 Human Fertility Research Group, CHU de Toulouse, University of Toulouse, Toulouse, France (Hamdi) Laboratory of Biochemistry and Hormonology, CHU de Toulouse, University of Toulouse, Toulouse, France

(Galiniere) Department of Paediatric Surgery, CHU de Toulouse, Université de Toulouse, Toulouse, France

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375.

Band-like calcification with simplified gyration and polymicrogyria: Report of 10 new families and identification of five novel OCLN mutations.

Abdel-Hamid M.S., Abdel-Salam G.M.H., Issa M.Y., Emam B.A., Zaki M.S.

Embase

Journal of Human Genetics. 62 (5) (pp 553-559), 2017. Date of Publication: 01 Apr 2017.

[Article]

AN: 615976371

Band-like calcification with simplified gyration and polymicrogyria (BLC-PMG) is an extremely rare autosomal recessive disorder with distinctive clinical and neuroimaging findings. To date, only 17 patients from 9 unrelated families with BLC-PMG have been reported worldwide. Herein, we describe a series of 13 new patients derived from 10 unrelated Egyptian families. Patients presented at early life with the classic phenotype including severe microcephaly, failure to acquire developmental skills, growth failure and the distinguished calcification patterns involving the cortex, thalami, basal ganglia and pons. Additional features not reported before included calcification of the cerebellum (eight patients: 61.5%) and imperforate anus and undescended testis in a single patient. Molecular studies of the OCLN gene (NM-001205254) identified six distinct candidate mutations. Interestingly, the deletion mutation of the transmembrane domain in exons 3 and 4 (c.51-?-730-?del, p.Lys18-Glu243) was found in five unrelated families (50%), suggesting a founder mutation in our population. On the other hand, five novel truncating mutations (c.809delA (p.K270Rfs*62), c.858-861delTTAT (p.I286Mfs*45), c.1037+5G>C, c.1169C>G (p.S390*) and c.1180delG (p.E394Sfs*91)) were detected, each in one family. To our

knowledge, this is the largest series of patients with BLC-PMG. Cerebellum calcification is an additional relevant finding in our series, thus expanding the neuroradiological phenotype of this syndrome.

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Institution

(Abdel-Hamid) Division of Human Genetics and Genome Research, Department of Medical Molecular Genetics, National Research Centre, Cairo, Egypt (Abdel-Salam, Issa, Emam, Zaki) Division of Human Genetics and Genome Research, Department of Clinical Genetics, National Research Centre, Eltahrir Street, Dokki, Cairo 12311, Egypt

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376.

The cremasteric reflex and its muscle - a paragon of ongoing scientific discussion: A systematic review.

Schwarz G.M., Hirtler L.

Embase

Clinical Anatomy. 30 (4) (pp 498-507), 2017. Date of Publication: May 2017.

[Article]

AN: 615632880

The technique of triggering the cremasteric reflex and its respective signaling pathway is not described uniformly throughout the literature. As this reflex is a useful sign in diagnosing testicular torsion, orchitis, varicocele, and undescended testis, it seems desirable to identify and define the correct mechanism. Our aim was to investigate how the cremasteric reflex and its signaling pathway are described in the current literature and how the variability of the innervation of the inguinal region could affect the frequency of this reflex. Thirty-five original articles and 18 current

textbooks were included after searching PubMed (MEDLINE) and Scopus for the terms "cremaster muscle," "cremasteric reflex," and "genitofemoral nerve" and after applying all exclusion criteria. This systematic review was performed according to the PRISMA Statement Rules. Eliciting the cremasteric reflex was defined either as "rubbing of the upper inner thigh" or "rubbing of the skin under the inguinal ligament." Four different afferent pathways among studies and three different pathways among textbooks were described and the frequency of an intact reflex ranged between 42.7 and 92.5% in newborns and between 61.7 and 100% in boys between 24 months and 12 years. Owing to the huge differences among the studies investigated and the lack of convincing results, it is not possible to define the correct way to elicit the cremasteric reflex. Four hypotheses about the afferent pathway are proposed on the basis of the literature. Further studies should be performed, concentrating on the afferent pathway(s) with respect to the individual innervation of the inguinal region. Clin. Anat. 30:498-507, 2017. © 2017 Wiley Periodicals, Inc.

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Status

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Author NameID

Hirtler, Lena; ORCID: <http://orcid.org/0000-0001-5194-9118>

Institution

(Schwarz, Hirtler) Division of Anatomy, Center for Anatomy and Cell Biology, Medical University of Vienna, Vienna, Austria

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Year of Publication

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377.

The management of boys under 3 months of age with an inguinal hernia and ipsilateral palpable undescended testis.

Wright N.J., Davidson J.R., Major C., Durkin N., Tan Y.-W., Jobson M., Ade-Ajayi N., Hall N.J., Bouhadiba N.

Embase

Journal of Pediatric Surgery. 52 (7) (pp 1108-1112), 2017. Date of Publication: July 2017.

[Article]

AN: 614749788

Aims The optimal management for boys under 3 months of age with an indirect inguinal hernia (IIH) and ipsilateral palpable undescended testis (IPUDT) is unknown. We aimed to: 1) determine the current practice for managing these boys across the UK, and 2) compare outcomes of different treatment strategies. **Methodology** We undertook two studies. Firstly, we completed a National Survey of all surgeons on the British Association of Paediatric Surgeons email list in 2014. Subsequently, we undertook a multi-centre, retrospective, 10-year (2005-2015) review across 4 pediatric surgery centers of boys under 3 months of age with concomitant IIH and IPUDT. Primary outcome was testicular atrophy. Secondary outcomes included need for subsequent orchidopexy, testicular ascent and hernia recurrence. Data are presented as median (range). Chi-squared test and multivariate binomial logistic regression analysis were used for analysis; $p < 0.05$ was considered significant. **Results** Survey: Consultant practice varies widely across the UK, with a tendency towards performing concurrent orchidopexy at the time of herniotomy under 3 months of age. Concurrent orchidopexy is favored less in cases where the hernia is symptomatic. **Case Series Review:** Forty-one boys with 43 concomitant IIH and IPUDT were identified, and all included. 32 (74%) hernias were reducible, 11 (26%) were symptomatic requiring urgent or emergency repair. Post-conceptual age at surgery was 45 weeks (36-65). Primary operations included: 29 (67%) open hernia repair and standard orchidopexy, 8 (19%) open hernia repair with future orchidopexy if required, 4 (9%) laparoscopic hernia repair with future orchidopexy if required, 2 (5%) open hernia repair and suturing of the testis to the inverted scrotum without scrotal incision. Variation in atrophy rate between different surgical approaches did not reach statistical significance ($p = 0.42$). Overall atrophy rate was 18%. If hernia repair alone was undertaken (8 open and 4 laparoscopic), the testis did not descend in 8 patients, requiring subsequent orchidopexy (67%); if orchidopexy was undertaken at the time of hernia repair, 1 in 29 required a repeat orchidopexy (3%) ($p = 0.0001$). No hernia recurred. **Conclusion** This study suggests that orchidopexy at the time of inguinal herniotomy does not increase the risk of testicular atrophy in boys under 3 months of age.

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Status

Embase

Institution

(Wright, Major, Bouhadiba) Department of Paediatric Surgery, Evelina Children's Hospital, Guy's and St. Thomas' NHS Trust, London, United Kingdom (Wright, Davidson, Bouhadiba)

Department of Paediatric Surgery, University Hospital Lewisham, London, United Kingdom

(Durkin, Tan, Ade-Ajayi) Department of Paediatric Surgery, King's College Hospital, London, United Kingdom

(Jobson, Hall) Department of Paediatric Surgery and Urology, Southampton Children's Hospital, Southampton, United Kingdom

(Hall) Faculty of Medicine, University of Southampton, Southampton, United Kingdom

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378.

Cryptorchidism and pesticides: Is there a connection?.

Fratric I., Varga J., Vukmirovic S., Sudji J., Zivkovic D.

Embase

Journal of Pediatric Surgery. 52 (7) (pp 1166-1168), 2017. Date of Publication: July 2017.

[Article]

AN: 613978142

Introduction The aim of our study was to compare the level of the most common organophosphate metabolite, dimethyl phosphate, in urine of women giving birth to both boys with cryptorchidism (study group), and healthy boys (control group), as well as to compare the level of dimethyl phosphate in our population with the results obtained in other populations.
Material and methods After the ethical approval we included thirty women in both study and control groups. All newborns were born between 38 and 42 weeks' gestation. Urine samples were taken on 3rd postpartal day. Gas chromatography with flame photometric detection was used to analyze dimethyl phosphate in urine following the method of Wu et al. Statistical analysis was done using Mann-Whitney test to compare the results in the two groups. **Results** Geometric mean of dimethyl phosphate in the study group was 7.18 +/- 8.26 mug/L and the creatinine-corrected level was 5.63 +/- 5.95 mug/L, and in the control group, the values are 7.98 +/- 6.75 mug/L and 6.15 +/- 7.01 mug/L, respectively. There was not a statistically significant difference in levels of

dimethyl phosphate between these two groups ($p = 0.72786$). Dimethyl phosphate levels obtained in similar studies are: 14.4 mug/L in Israel, 3.7 mug/L in Palestine, 10.3 mug/L in Jerusalem, 1.60 mug/L in Caribbean islands and 2.60 mug/L in Canada. Conclusions Pregnant women in our country are exposed to organophosphate pesticides, but a correlation between the exposure to organophosphate pesticides and cryptorchidism was not found. Level of evidence I. Type of Study: Prognostic study, prospective study.

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Status

Embase

Author NameID

Fratric, Ivana; ORCID: <http://orcid.org/0000-0001-7124-8087>

Institution

(Fratric) Department of Pediatric Surgery, Faculty of Medicine, University of Novi Sad, Institute for Children and Youth Health Care of Vojvodina, Hajduk Veljkova 10, Novi Sad 21000, Serbia

(Varga) Clinic for Pediatric Surgery, Department of Urology, Institute for Children and Youth Health Care of Vojvodina, Faculty of Medicine, University of Novi Sad, Novi Sad, Serbia

(Vukmirovic) Department of Pharmacology and Toxicology, Faculty of Medicine, University of Novi Sad, Novi Sad, Serbia

(Sudji) Institute of Occupational Health, Futoska 121, Novi Sad 21000, Serbia

(Zivkovic) Department of Health Care, Faculty of Medicine, University of Novi Sad, Department of Urology, Institute for Children and Youth Health Care of Vojvodina, Novi Sad, Serbia

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379.

Staged Fowler-Stephens and Single-stage Laparoscopic Orchiopexy for Intra-abdominal Testes: Is There a Difference? A Single Institution Experience.

Alam A., Delto J.C., Blachman-Braun R., Wayne G., Mittal A.G., Castellan M., Kozakowski K., Labbie A., Gosalbez R.

Embase

Urology. 101 (pp 104-110), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 613950980

Objective To compare single-stage laparoscopic orchiopexy (SSLO) and staged Fowler-Stephens (SFS) procedures in the management of intra-abdominal undescended testes, and to analyze postoperative atrophy and malpositioning as end points. **Materials and Methods** A retrospective chart review identified laparoscopic orchiopexy patients with intra-abdominal testes between November 2006 and November 2014. Of 167 patients who had laparoscopic orchiopexy, 73 (85 testes) were identified as having laparoscopic orchiopexy. Baseline characteristics, as well as testicular scrotal position and size at follow-up, were recorded. Regression analysis was performed to compare outcomes between patients who underwent SFS and SSLO. **Results** Of the 85 laparoscopic orchiopexies, 35 underwent SFS and 50 had SSLO. Patient demographics were comparable in both groups. The median age at surgery was 12 months (5-151 months), and the average follow-up was 17.3 months. On follow-up, there were 0 recorded cases of SFS patients with abnormally positioned testes postoperatively, whereas there were 10 (20.0%) SSLO patients who had abnormally positioned testes (odds ratio: 0.05, 95% confidence interval: 0.01-0.44). Differences in atrophy rates were not significant. **Conclusion** These results suggest that there may be no difference between the 2 approaches in terms of postoperative atrophy. However, the SFS appears to be more successful in securing a favorable scrotal position. Atrophy does not seem to be associated with other patient factors. Prospective, randomized studies are indicated to further explore outcome differences between the 2 approaches.

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Author NameID

Blachman-Braun, Ruben; ORCID: <http://orcid.org/0000-0001-9700-0805>

Institution

(Alam, Blachman-Braun, Mittal, Castellan, Kozakowski, Labbie, Gosalbez) Division of Pediatric Urology, Nicklaus Children's Hospital, Miami, FL, United States (Delto, Wayne) Urology

Department, Mount Sinai Medical Center, Miami Beach, FL, United States

(Mittal) Department of Pediatric Urology, Texas Children's Hospital and Baylor School of Medicine, Houston, TX, United States

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380.

Does varicocelectomy improve semen analysis outcomes in adolescents without testicular asymmetry?.

Chu D.I., Zderic S.A., Shukla A.R., Srinivasan A.K., Tasian G.E., Weiss D.A., Long C.J., Canning D.A., Kolon T.F.

Embase

Journal of Pediatric Urology. 13 (1) (pp 76.e1-76.e5), 2017. Date of Publication: 01 Feb 2017.

[Article]

AN: 613738331

Purpose The main indications for adolescent varicocelectomy are testicular hypotrophy or pain. However, we have previously shown that both serial total testicular volume and volume differential are weakly associated with semen quality. The ultimate patient goal is paternity, but semen analysis is critical to appropriate management of varicocele. We hypothesize that varicocelectomy improves total motile count (TMC) among patients who only have abnormal semen analysis (SA) parameters, but not among those with potential hormonal dysfunction such as Klinefelter syndrome or cryptorchidism. **Methods** We retrospectively reviewed our registry of adolescent males followed with a clinical left varicocele. For this study, subjects without sustained testicular asymmetry, who were Tanner V, and gave at least one preoperative SA were included. Subjects were excluded if they had embolization for their varicocele or no postoperative SA. **Primary outcome** was change in TMC before and after surgery, compared using the Wilcoxon signed rank test after stratifying by surgical indication. **Secondary outcomes** included rates of improved TMC and normalized TMC (>20 million) after surgery, compared across covariates using the Fisher exact test. **Results** Seventeen patients met the eligibility criteria, 11 of whom underwent repair for only abnormal preoperative TMC. Overall, median age (interquartile range [IQR]) at first preoperative SA was 17.6 (15.9-17.9) years. The median preoperative TMC across all SA was 2.8 (0.7-7.4) million. The median age at surgery was 18.2 (16.8-18.9) years. Postoperatively, the median TMC across all SA increased to 18.2 (3.6-18.2) million (Wilcoxon

signed rank test, $p < 0.01$; see Figure). The improvement in TMC occurred primarily in the group who only had abnormal preoperative TMC (82% improved, 55% normalized); lack of improvement was seen in patients who had a history of Klinefelter or orchiopexy for cryptorchidism.

Conclusions Adolescent varicocele patients should undergo SA after development of Tanner V. Varicocelectomy has a high success rate for improving TMC in adolescent or young adult males who only have abnormal TMC and no history of cryptorchidism.[Figure presented]

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Author NameID

Chu, David I.; ORCID: <http://orcid.org/0000-0002-7972-4671> Zderic, Stephen A.; ORCID:

<http://orcid.org/0000-0003-3839-7106>

Kolon, Thomas F.; ORCID: <http://orcid.org/0000-0002-8740-5021>

Institution

(Chu, Zderic, Shukla, Srinivasan, Tasian, Weiss, Long, Canning, Kolon) Division of Urology, The Children's Hospital of Philadelphia, Philadelphia, PA, United States

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381.

The natural history of semen parameters in untreated asymptomatic adolescent varicocele patients: A retrospective cohort study.

Chu D.I., Zderic S.A., Shukla A.R., Srinivasan A.K., Tasian G.E., Weiss D.A., Long C.J., Canning D.A., Kolon T.F.

Embase

Journal of Pediatric Urology. 13 (1) (pp 77.e1-77.e5), 2017. Date of Publication: 01 Feb 2017.

[Article]

AN: 613695119

Introduction Adolescent males with varicoceles present a dilemma for surveillance and treatment. Testicular volumetrics have not been shown to predict SA outcomes. Serial SAs are thus recommended in asymptomatic adolescent males with varicoceles and normal testicular development, but the natural history of semen parameters is unknown. Objective To explore the natural history of semen parameters in adolescent boys with a left varicocele under active surveillance. Study design Adolescents with an asymptomatic unilateral left varicocele, Tanner V development, normal testicular volumes, and an initial SA were retrospectively reviewed in a single-institution prospectively followed cohort. Total motile count (TMC) was calculated. A cutoff of TMC \geq 20 million was used to dichotomize SA results into "normal" or "poor." Those with poor SA were offered repeat SA. Cumulative probabilities of normal TMC over successive rounds of SA were calculated. Bivariate models were used to explore associations of a second consecutive poor TMC with age and varicocele grade. Results A total of 216 patients provided an initial SA between 1992 and 2015. We excluded 17 for a history of cryptorchidism or incomplete SA data for a final cohort of 199 patients with median follow-up of 3.3 years (interquartile range 1.5-5.6 years). The mean age at initial SA was 17.9 years (range 14.8-21.8 years). One hundred and nine out of 199 had an initial normal TMC. Of the 90 out of 199 with an initially poor TMC, 51 had repeat SA and 24 of the 51 patients improved to normal TMC. Of the 27 patients with two consecutive poor TMCs, 15 had a third SA and five out of 15 improved to normal TMC. Thus, cumulatively, 55%, 67%, and 69% of all patients had a normal TMC after an initial, second, and third SA, respectively. However, fewer patients in each round of SA normalized their TMC (Figure). Neither age nor varicocele grade was associated with a second consecutive poor TMC. Discussion Two-thirds of Tanner V boys with an uncorrected varicocele and normal testicular volumes achieve a normal TMC regardless of varicocele grade or age. Despite Tanner V development, 47% with an initial "poor" SA will improve to normal status without surgery. However, a small subgroup of patients will have persistently poor TMC and thus should be targeted in future research for timely intervention. Conclusion Semen parameters improve over time. SA should be followed and repeated at least once in symptomatic Tanner V boys with varicoceles.[Figure presented]

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Author NameID

Chu, David I.; ORCID: <http://orcid.org/0000-0002-7972-4671> Zderic, Stephen A.; ORCID:

<http://orcid.org/0000-0003-3839-7106>

Canning, Douglas A.; ORCID: <http://orcid.org/0000-0003-4998-2669>

Kolon, Thomas F.; ORCID: <http://orcid.org/0000-0002-8740-5021>

Institution

(Chu, Zderic, Shukla, Srinivasan, Tasian, Weiss, Long, Canning, Kolon) Division of Urology, The Children's Hospital of Philadelphia, Philadelphia, PA, United States

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382.

Ascending testis after repair of pediatric inguinal hernia and hydrocele: A misunderstood operative complication.

Wang F., Zhong H., Zhao J.

Embase

Journal of Pediatric Urology. 13 (1) (pp 53.e1-53.e5), 2017. Date of Publication: 01 Feb 2017.

[Article]

AN: 612930012

Introduction Ascending testis (AT) is a rare complication after repair of an inguinal hernia/hydrocele. However, there has been some controversy concerning the AT following laparoscopic and open procedures. Objective To review the experience of, and discuss the associated mechanisms with, testicular ascent after pediatric inguinal hernia/hydrocele surgery. Study design A retrospective review of the medical records of male children who underwent inguinal hernia/hydrocele repair at the present hospital between January 2000 and December 2014. Those who underwent subsequent orchiopexies due to ipsilateral cryptorchidism were identified. The ATs that were misdiagnosed, caused by improper operation, and retractile testes were excluded. The Poisson distribution evaluated the incidences of subgroups. Results A total of 17,295 inguinal hernias and hydroceles were repaired on 12,849 males; of whom, 10 testes (0.058%) developed ATs on nine individuals postoperatively (Summary Table). The difference of AT incidences between subgroups was insignificant ($P > 0.05$), except for that grouped by the age at initial operation (cutoff = 1 year, $P = 0.008$; cutoff = 2 years, $P = 0.012$). During orchiopexy, extensive adhesions were found in the inguinal canal only in the two cases following open repair. The hernia sac/processus vaginalis remained intact in the canal of AT after laparoscopic repair,

and partial after open herniotomy. Discussion It was generally assumed that testicular ascent after repair of an inguinal hernia/hydrocele was caused by adhesion of the spermatic cord. However, the cord was not dissected during laparoscopic procedure, so adhesion was not the major reason for AT following laparoscopic surgery. The sac/processus were partially excised during open repair, but kept intact in laparoscopic procedure. Therefore, remnants of the sac/processus might play a greater role in postoperative testicular ascent than adhesions. Furthermore, it was found that AT incidence after the repair was not higher than that in 'normal' males. Ascending testis was probably not an operative complication, but a natural descent process of testis independent of the operation. Moreover, the testis descended further due to dissection of the cord and excision of the sac/processus, so the AT incidence was extremely low following open operation. From this point of view, open repair of an inguinal hernia/hydrocele was probably a protective factor for preventing testicular ascent. Conclusion Ascending testis is rare in male pediatric patients who have had repair of an inguinal hernia/hydrocele. Currently, the mechanism of testicular ascent is still unclear, and should be further investigated in the future.[Table presented]

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Status

Embase

Institution

(Wang, Zhong, Zhao) Department of Pediatric Urology, Ningbo Women & Children's Hospital, Cicheng New Town, Jiangbei District, Ningbo, Zhejiang, China

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383.

Postnatal risk factors for testicular cancer: The EPSAM case-control study.

Moirano G., Zugna D., Grasso C., Mirabelli D., Lista P., Ciuffreda L., Segnan N., Merletti F., Richiardi L.

Embase

International Journal of Cancer. 141 (9) (pp 1803-1810), 2017. Date of Publication: 01 Nov 2017.

[Article]

AN: 617580978

Testicular cancer is considered to originate from an impaired differentiation of fetal germ cells, but puberty could represent another time window of susceptibility. Our study aimed at investigating the association between environmental exposures acting during puberty/adolescence (13-19 years of age) and the risk of testicular cancer. We used data of the EPSAM study, a case-control study on germ-cell testicular cancer conducted in the province of Turin, Italy, involving cases diagnosed between 1997 and 2008. Histologically confirmed cases (n = 255) and controls (n = 459) completed a postal questionnaire focusing in particular on the pubertal period (namely age 13 years) with questions on physical activity (competitive sports, gardening), lifestyle (alcohol consumption, smoking), occupational history and medical conditions. All analyses were adjusted for the matching variables, cryptorchidism and educational level. Having done at least one competitive sport during puberty (odds ratio [OR]: 0.72, 95% confidence interval: 0.52-1.00), gardening activities during puberty (OR: 0.62, 0.42-0.94) and having a lower weight than peers during puberty (OR: 0.64, 0.42-0.97) were all inversely associated with the risk of testicular cancer. No evidence of association between smoking or alcohol consumption during puberty and the risk of testicular cancer was observed. Regarding agriculture-related occupations, we found an association with the risk of testicular cancer both for occasional jobs during puberty (OR: 2.40, 95% CI: 1.08-5.29) and ever employment in adolescence (OR: 2.59, 95% CI: 0.83-8.10). Our results suggest that postnatal exposures could play a role in testicular cancer aetiology, at least when acting in puberty or adolescence.

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Author NameID

Moirano, Giovenale; ORCID: <http://orcid.org/0000-0001-8748-3321>

Institution

(Moirano, Zugna, Grasso, Mirabelli, Merletti, Richiardi) Cancer Epidemiology Unit-CERMS, Department of Medical Sciences, University of Turin and CPO-Piemonte, Torino, Italy (Lista, Ciuffreda) Medical Oncology Division 1, University Hospital "Citta' della Salute e della Scienza", Turin, Italy

(Segnan) Department of Cancer Screening and Unit of Cancer Epidemiology, WHO Collaborative Center for Cancer Early Diagnosis and Screening, CPO Piedmont and University Hospital "Citta' della Salute e della Scienza", Turin, Italy

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Year of Publication

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384.

Demographics and co-occurring conditions in a clinic-based cohort with Down syndrome in the United Arab Emirates.

Corder J.P., Al Ahbabi F.J.S., Al Dhaheri H.S., Chedid F.

Embase

American Journal of Medical Genetics, Part A. 173 (9) (pp 2395-2407), 2017. Date of Publication: September 2017.

[Article]

AN: 617259192

The majority of studies describing demographics and co-occurring conditions in cohorts with Down syndrome come from regions outside of the Middle East, mainly from Europe and North America. This paper describes demographics and co-occurring conditions in a hospital-based cohort of individuals with Down syndrome living in the Middle Eastern country of the United Arab Emirates (UAE). The first dedicated Down syndrome clinic in the UAE was established in 2012 at Tawam Hospital in Al Ain. This paper describes a clinic-based cohort of 221 participants over 4 years from the Gulf Down Syndrome Registry, a new Down syndrome database and contact registry created at Tawam Hospital. Key demographic findings include mean maternal age of 37 years, among the highest described in the literature. Sixty-two percent of mothers are >35 years. Over 90% of mothers received post-natal diagnosis of Down syndrome. High sex ratio, parental consanguinity, and large family size also characterize the group. The spectrum of many co-occurring conditions mirrors that of previously described populations, with some notable differences. Cardiovascular malformations are well represented, however, atrioventricular canal is not the most common. Genitourinary conditions are common, as evidenced by 12% of males with hypospadias and 15% with undescended testes. Glucose-6-phosphate dehydrogenase

deficiency, alpha thalassemia trait, hypovitaminosis D, and dental caries are common in our cohort. This study describes a large hospital-based group with Down syndrome presenting to a new dedicated Down syndrome clinic in the UAE, highlighting unique demographic and co-occurring conditions found in that population.

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Author NameID

Corder, Jennifer Price; ORCID: <http://orcid.org/0000-0002-3629-566X>

Institution

(Corder, Al Ahbabi, Al Dhaheri) Department of Pediatrics, Tawam Hospital in Affiliation With Johns Hopkins International, Al Ain, United Arab Emirates (Chedid) Department of Neonatology, Al Jalila Children's Specialty Hospital, Dubai, United Arab Emirates

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385.

Genes Involved in Long-Term Memory Are Expressed in Testis of Cryptorchid Boys and Respond to GnRHa Treatment.

Hadziselimovic F., Gegenschatz-Schmid K., Verkauskas G., Demougin P., Bilius V., Dasevicius D., Stadler M.B.

Embase

Cytogenetic and Genome Research. 152 (1) (pp 9-15), 2017. Date of Publication: 01 Aug 2017.

[Article]

AN: 617340062

It has been known for many years that boys with unilateral or bilateral undescended testis (cryptorchidism) tend to have a low IQ, and those who belong to the high infertility risk (HIR) group perform less well at school than low infertility risk (LIR) patients. However, the molecular

biological processes underlying this phenomenon are not understood. In this study, we report the outcome of testicular RNA profiling for genes involved in long-term memory formation. We analyzed the histology and the transcriptome of testicular biopsies from bilateral HIR cryptorchid boys, comparing those who received GnRHa treatment for 6 months after the first surgery with those who did not receive GnRHa before the second surgery. We found that GnRHa treatment alters the testicular mRNA levels of neuronal genes that are involved in long-term memory and testosterone synthesis. These data highlight a possible molecular link between cryptorchidism, impaired mini-puberty, and diminished cognitive functions. Our results are consistent with the hypothesis that hypogonadotropic hypogonadism in cryptorchid boys with altered mini-puberty may affect neuronal genes important for memory and learning, which could help explaining the negative correlation between cryptorchidism and intellectual abilities.

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Institution

(Hadziselimovic, Gegenschatz-Schmid) Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, Bahnhofplatz 11, Liestal CH-4410, Switzerland (Demougin) Biozentrum, Life Sciences Training Facility, University of Basel, Basel, Switzerland

(Stadler) Friedrich Miescher Institute for Biomedical Research, Basel, Switzerland

(Stadler) Swiss Institute of Bioinformatics, Basel, Switzerland

(Verkauskas, Bilius) Children's Surgery Centre, Faculty of Medicine, University of Vilnius, Vilnius, Lithuania

(Dasevicius) Institute for Pathology, National Centre of Pathology, Affiliate of Vilnius University Hospital Santariskiu Klinikos, Vilnius, Lithuania

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2017

Mutations in the human ROBO1 gene in pituitary stalk interruption syndrome.

Bashamboo A., Bignon-Topalovic J., Moussi N., McElreavey K., Brauner R.

Embase

Journal of Clinical Endocrinology and Metabolism. 102 (7) (pp 2401-2406), 2017. Date of

Publication: 01 Jul 2017.

[Article]

AN: 617314617

Context: Pituitary stalk interruption syndrome (PSIS) is characterized by a thin or absent pituitary stalk usually in association with an ectopic posterior pituitary and hypoplasia/aplasia of the anterior pituitary. Associated phenotypes include varied ocular anomalies, hypoglycemia, micropenis/ cryptorchidism, growth failure, or combined pituitary hormone deficiencies. Although genetic causes have been identified, they explain only around 5% of PSIS cases.

Objective(s): To identify genetic causes of PSIS by exome sequencing.

Design(s): Exon enrichment was performed using the Agilent SureSelect Human All Exon V4. Pairedend sequencing was performed on the Illumina HiSeq2000 platform with an average sequencing coverage of 350.

Patient(s): Patients with unexplained PSIS were included in the study.

Result(s): In five cases of unexplained PSIS including two familial cases, we identified a novel heterozygous frameshift and nonsense and missense mutations in the ROBO1 gene (p.Ala977Glnfs*40, two affected sibs; p.Tyr1114Ter, sporadic case, and p.Cys240Ser, affected child and paternal aunt) that controls embryonic axon guidance, and branching in the nervous system. Interestingly, four of the five cases of PSIS also presented with ocular anomalies, including hypermetropia with strabismus as well as ptosis.

Conclusion(s): These data suggest that mutations in ROBO1 contribute to PSIS and associated ocular anomalies.

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Embase

Institution

(Bashamboo, Bignon-Topalovic, Moussi, McElreavey) Human Developmental Genetics, Institut Pasteur, 25 rue du Dr Roux, Paris Cedex 15 75724, France (Brauner) Universite Paris Descartes and Pediatric Endocrinology Unit, Fondation Ophtalmologique Adolphe de Rothschild, Paris 75019, France

Publisher

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Year of Publication

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387.

Mutations in the leukemia inhibitory factor receptor (LIFR) gene and Lifr deficiency cause urinary tract malformations.

Kosfeld A., Brand F., Weiss A.-C., Kreuzer M., Goerk M., Martens H., Schubert S., Schafer A.-K., Riehmer V., Hennies I., Brasen J.H., Pape L., Amann K., Krogvold L., Bjerre A., Daniel C., Kispert A., Haffner D., Weber R.G.

Embase

Human Molecular Genetics. 26 (9) (pp 1716-1731), 2017. Article Number: ddx086. Date of Publication: 01 May 2017.

[Article]

AN: 616106177

Congenital anomalies of the kidneys and urinary tract (CAKUT) are the most common cause of chronic kidney disease in children. As CAKUT is a genetically heterogeneous disorder and most cases are genetically unexplained, we aimed to identify new CAKUT causing genes. Using whole-exome sequencing and trio-based de novo analysis, we identified a novel heterozygous de novo frameshift variant in the leukemia inhibitory factor receptor (LIFR) gene causing instability of the mRNA in a patient presenting with bilateral CAKUT and requiring kidney transplantation at one year of age. LIFR encodes a transmembrane receptor utilized by IL-6 family cytokines, mainly by the leukemia inhibitory factor (LIF). Mutational analysis of 121 further patients with severe CAKUT yielded two rare heterozygous LIFR missense variants predicted to be pathogenic in three unrelated patients. LIFR mutants showed decreased half-life and cell membrane localization resulting in reduced LIF-stimulated STAT3 phosphorylation. LIFR showed high expression in human fetal kidney and the human ureter, and was also expressed in the developing murine urogenital system. Lifr knockout mice displayed urinary tract malformations including hydronephrosis, hydroureter, ureter ectopia, and, consistently, reduced ureteral lumen and muscular hypertrophy, similar to the phenotypes observed in patients carrying LIFR variants. Additionally, a form of cryptorchidism was detected in all Lifr^{-/-} mice and the patient carrying the LIFR frameshift mutation. Altogether, we demonstrate heterozygous novel or rare LIFR mutations

in 3.3% of CAKUT patients, and provide evidence that Lifr deficiency and deactivating LIFR mutations cause highly similar anomalies of the urogenital tract in mice and humans.

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Institution

(Kosfeld, Brand, Martens, Schubert, Riehmer, Weber) Department of Human Genetics, Hannover Medical School, Hannover, Germany (Weiss, Schafer, Kispert) Institute of Molecular Biology, Hannover Medical School, Hannover, Germany

(Kreuzer, Hennies, Pape, Haffner) Department of Pediatric Kidney, Liver and Metabolic Diseases, Hannover Medical School, Hannover, Germany

(Goerk, Amann, Daniel) Department of Nephropathology, Friedrich-Alexander University of Erlangen-Nurnberg, Erlangen, Germany

(Brasen) Department of Pathology, Hannover Medical School, Hannover, Germany

(Krogvold, Bjerre) Division of Paediatric and Adolescent Medicine, Oslo University Hospital, Oslo, Norway

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388.

Comparative safety of anti-epileptic drugs during pregnancy: A systematic review and network meta-analysis of congenital malformations and prenatal outcomes.

Veroniki A.A., Cogo E., Rios P., Straus S.E., Finkelstein Y., Kealey R., Reynen E., Soobiah C., Thavorn K., Hutton B., Hemmelgarn B.R., Yazdi F., D'Souza J., MacDonald H., Tricco A.C.

Embase

BMC Medicine. 15 (1) (no pagination), 2017. Article Number: 95. Date of Publication: 05 May 2017.

[Article]

AN: 615882610

Background: Pregnant women with epilepsy frequently experience seizures related to pregnancy complications and are often prescribed anti-epileptic drugs (AEDs) to manage their symptoms. However, less is known about the comparative safety of AED exposure in utero. We aimed to compare the risk of congenital malformations (CMs) and prenatal outcomes of AEDs in infants/children who were exposed to AEDs in utero through a systematic review and Bayesian random-effects network meta-analysis.

Method(s): MEDLINE, EMBASE, and Cochrane CENTRAL were searched from inception to December 15, 2015. Two reviewers independently screened titles/abstracts and full-text papers for experimental and observational studies comparing mono- or poly-therapy AEDs versus control (no AED exposure) or other AEDs, then abstracted data and appraised the risk of bias. The primary outcome was incidence of major CMs, overall and by specific type (cardiac malformations, hypospadias, cleft lip and/or palate, club foot, inguinal hernia, and undescended testes).

Result(s): After screening 5305 titles and abstracts, 642 potentially relevant full-text articles, and 17 studies from scanning reference lists, 96 studies were eligible (n = 58,461 patients). Across all major CMs, many AEDs were associated with higher risk compared to control. For major CMs, ethosuximide (OR, 3.04; 95% CrI, 1.23-7.07), valproate (OR, 2.93; 95% CrI, 2.36-3.69), topiramate (OR, 1.90; 95% CrI, 1.17-2.97), phenobarbital (OR, 1.83; 95% CrI, 1.35-2.47), phenytoin (OR, 1.67; 95% CrI, 1.30-2.17), carbamazepine (OR, 1.37; 95% CrI, 1.10-1.71), and 11 polytherapies were significantly more harmful than control, but lamotrigine (OR, 0.96; 95% CrI, 0.72-1.25) and levetiracetam (OR, 0.72; 95% CrI, 0.43-1.16) were not.

Conclusion(s): The newer generation AEDs, lamotrigine and levetiracetam, were not associated with significant increased risks of CMs compared to control, and were significantly less likely to be associated with children experiencing cardiac malformations than control. However, this does not mean that these agents are not harmful to infants/children exposed in utero. Counselling is advised concerning teratogenic risks when the prescription is written for a woman of childbearing age and before women continue with these agents when considering pregnancy, such as switching from polytherapy to monotherapy with evidence of lower risk and avoiding AEDs, such as valproate, that are consistently associated with CMs. These decisions must be balanced against the need for seizure control. Systematic Review Registration: PROSPERO

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Institution

(Veroniki, Cogo, Rios, Straus, Kealey, Reynen, Soobiah, Yazdi, D'Souza, MacDonald, Tricco) St. Michael's Hospital, Knowledge Translation Program, Li Ka Shing Knowledge Institute, 209 Victoria Street, East Building, Toronto, ON M5B 1W8, Canada (Straus) University of Toronto, Department of Geriatric Medicine, 27 King's College Circle, Toronto, ON M5S 1A1, Canada (Finkelstein) The Hospital for Sick Children, 555 University Avenue, Toronto, ON M5G 1X8, Canada

(Finkelstein) University of Toronto, Department of Paediatrics, 172 St. George Street, Toronto, ON M5R 0A3, Canada

(Finkelstein) University of Toronto, Department of Pharmacology and Toxicology, Medical Sciences Building, 1 King's College Circle, Toronto, ON M5S 1A8, Canada

(Soobiah) University of Toronto, Institute for Health Policy Management and Evaluation, 4th Floor, 155 College Street, Toronto, ON M5T 3M6, Canada

(Thavorn, Hutton) University of Ottawa, School of Epidemiology, Public Health and Preventive Medicine, Faculty of Medicine, Roger-Guindon Building, 451 Smyth Road, Ottawa, ON K1H 8M5, Canada

(Thavorn) The Ottawa Hospital, Clinical Epidemiology Program, Ottawa Hospital Research Institute, 501 Smyth Road, Ottawa, ON K1H 8L6, Canada

(Thavorn) Institute of Clinical and Evaluative Sciences (ICES uOttawa), 1053 Carling Ave, Ottawa, ON K1Y 4E9, Canada

(Hutton) Center for Practice Changing Research, The Ottawa Hospital - General Campus, Ottawa Hospital Research Institute, 501 Smyth Road, PO Box 201B, Ottawa, ON K1H 8L6, Canada

(Hemmelgarn) University of Calgary, Departments of Medicine and Community Health Sciences, TRW Building, 3rd Floor, 3280 Hospital Drive NW, Calgary, AB T2N 4Z6, Canada

(Tricco) University of Toronto, Epidemiology Division, Dalla Lana School of Public Health, 6th Floor, 155 College Street, Toronto, ON M5T 3M7, Canada

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Constitutional bone impairment in Noonan syndrome.

Baldassarre G., Mussa A., Carli D., Molinatto C., Ferrero G.B.

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American Journal of Medical Genetics, Part A. 173 (3) (pp 692-698), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 614507841

Noonan syndrome (NS) is an autosomal dominant trait characterized by genotypic and phenotypic variability. It belongs to the Ras/MAPK pathway disorders collectively named Rasopathies or neurocardiofaciocutaneous syndromes. Phenotype is characterized by short stature, congenital heart defects, facial dysmorphisms, skeletal and ectodermal anomalies, cryptorchidism, mild to moderate developmental delay/learning disability, and tumor predisposition. Short stature and skeletal dysmorphisms are almost constant and several studies hypothesized a role for the RAS pathway in regulating bone metabolism. In this study, we investigated the bone quality assessed by phalangeal quantitative ultrasound (QUS) and the metabolic bone profiling in a group of patients with NS, to determine whether low bone mineralization is primary or secondary to NS characteristics. Thirty-five patients were enrolled, including 20 males (55.6%) and 15 females (44.5%) aged 1.0-17.8 years (mean 6.4 +/- 4.5, median 4.9 years). Each patients was submitted to clinical examination, estimation of the bone age, laboratory assays, and QUS assessment. Twenty-five percent of the cohort shows reduced QUS values for their age based on bone transmission time. Bone measurement were adjusted for multiple factors frequently observed in NS patients, such as growth retardation, delayed bone age, retarded puberty, and reduced body mass index, potentially affecting bone quality or its appraisal. In spite of the correction attempts, QUS measurement indicates that bone impairment persists in nearly 15% of the cohort studied. Our results indicate that bone impairment in NS is likely primary and not secondary to any of the phenotypic traits of NS, nor consistent with metabolic disturbances. © 2017 Wiley Periodicals, Inc.

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Institution

(Baldassarre, Carli, Molinatto, Ferrero) Department of Pediatric and Public Health Sciences, University of Turin, Turin, Italy

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390.

The challenging sonographic inguinal canal evaluation in neonates and children: an update of differential diagnoses.

Sameshima Y.T., Yamanari M.G.I., Silva M.A., Neto M.J.F., Funari M.B.G.

Embase

Pediatric Radiology. 47 (4) (pp 461-472), 2017. Date of Publication: 01 Apr 2017.

[Article]

AN: 613215783

Bulging of the inguinal region is a frequent complaint in the pediatric population and sonographic findings can be challenging for radiologists. In this review we update the sonographic findings of the most common disorders that affect the inguinal canal in neonates and children, with a focus on the processus vaginalis abnormalities such as congenital hydroceles, indirect inguinal hernias and cryptorchidism, illustrated with cases collected at a quaternary hospital during a 7-year period. We emphasize the importance of correctly classifying different types of congenital hydrocele and inguinal hernia to allow for early surgical intervention when necessary. We have systematically organized and illustrated all types of congenital hydrocele and inguinal hernias based on embryological, anatomical and pathophysiological findings to assist readers in the diagnosis of even complex cases of inguinal canal ultrasound evaluation in neonates and children. We also present rare diagnoses such as the abdominoscrotal hydrocele and the herniation of uterus and ovaries into the canal of Nuck.

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Embase

Institution

(Sameshima, Yamanari, Silva, Neto, Funari) Diagnostic Imaging Department, Hospital Israelita Albert Einstein, Av. Albert Einstein, 627/701, Sao Paulo, SP 05651-901, Brazil

Publisher

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Year of Publication

2017

391.

Long-Term Follow-Up of Testicular Microlithiasis in Children and Adolescents: Multicenter Prospective Cohort Study of the Italian Society of Pediatric Urology.

Marte A., Pintozzi L., Creti G., Chiesa P.L., Renzo D.D., Gasparella M., Maggio G.D., Bagnara V., Merlini E., Tadini B., Caldarulo E., Sangiorgio L., Battaglino G., Nappo S.G., Caione P.

Embase

European Journal of Pediatric Surgery. 27 (2) (pp 155-160), 2017. Article Number: 153592oa.

Date of Publication: 01 Apr 2017.

[Article]

AN: 609135791

Introduction Testicular microlithiasis (TM), characterized by the presence of intratubular calcifications in a single or both the gonads, is an uncommon entity with unknown etiology and outcome in pediatric and adolescent age. In this study, the results of a multicenter long-term survey are presented. **Materials and Methods** From 11 units of pediatric urology/surgery, patients with TM were identified and yearly, followed up in a 7-year period, adopting a specific database. The recorded items were: age at diagnosis, presenting symptoms/associated abnormalities, ultrasonographic finding, surgery and histology at biopsy, if performed. **Results** Out of 85 patients, 81 were evaluated yearly (4 patients lost to follow-up). TM was bilateral in 66.6% of the patients. Associate genital abnormalities were present in 90%, more frequently undescended/retractile testis (23.4%) and varicocele (22.2%). TM remained unchanged at 4.7 years follow-up in 77 patients (93.8%) and was reduced in 4 patients after 1 to 5 years of inguinoscrotal surgery. Orchiectomy was performed in three patients (3.7%), one for severe testicular hypoplasia and two for seminoma (2.5%), respectively, concurrent and metachronous to diagnosis of TM. Tumorectomy with parenchymal sparing surgery was performed in a teratoma associated with TM. **Conclusion** TM is a controversial entity, often associated with several inguinogenital features, which rarely can recover. Testicular malignancy, although present in TM, has not proven definitively associated to microliths. Proper counseling, yearly ultrasound, and self-examination are long-term recommended.

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Institution

(Marte, Pintozzi) Division of Pediatric Surgery, Second University of Naples, Largo Madonna delle Grazie 1, Naples 80138, Italy (Creti) Division of Pediatric Urology, Casa Sollievo della Sofferenza Hospital, IRCCS-San Giovanni Rotondo, Italy

(Chiesa, Renzo) Division Pediatric Surgery, Santo Spirito Hospital-G. D'Annunzio University-Chieti-Pescara, Pescara, Italy

(Gasparella) Division of Pediatric Surgery, Hospital of Treviso-Padua University, Treviso, Italy

(Maggio) Department of Medical Sciences, Surgery and Neuroscience, Siena University, Siena, Italy

(Bagnara) Unit of Pediatric Surgery, G.B. Morgagni Hospital, Catania, Italy

(Merlini, Tadini) Division of Pediatric Urology, S. Anna Hospital, Torino, Italy

(Caldarulo) Division of Pediatric Surgery OO.RR., Hospital of Foggia, Foggia, Italy

(Sangiorgio) Unit Pediatric Urology, SS. Antonio e Biagio e Cesare Arrigo Hospital, Alessandria, Italy

(Battaglino) Division of Pediatric Surgery, San Bortolo Hospital, Vicenza, Italy

(Nappo, Caione) Division of Pediatric Urology, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

Publisher

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2017

392.

Torsed and Nontorsed Inguinal Undescended Testis: Comparison of Computed Tomography Findings.

Klang E., Kanaan N., Soudack M., Kleinbaum Y., Heiman Z., Raskin S., Amitai M.M.

Embase

Journal of Computer Assisted Tomography. 41 (4) (pp 633-637), 2017. Date of Publication: 01 Jul 2017.

[Article]

AN: 614612032

Objective The aim of this study was to compare the computed tomography imaging features of a torsed inguinal testis with nontorsed inguinal testes. **Methods** Computed tomography scans of patients with undescended testes were retrospectively collected (2011-2016). Imaging features of nontorsed undescended testis were compared with a case of an inguinal torsed testis.

Observations included location of the undescended testis, size (length x width) and texture of each testis, peritesticular findings, position of testicular vessels, and enhancement patterns.

Results Twelve nontorsed inguinal undescended testes were compared with 1 torsed undescended testicle. Torsed testis was larger than nontorsed (44 x 27 mm vs 32.9 +/- 6.1 x 22.9 +/- 4.9 mm), surrounded by fat stranding and fluid, with heterogeneous texture, enhancement of its outer layers, and an upward kink of its vessels. **Conclusions** Because torsed undescended testis can mimic a groin abscess and because torsion is a medical emergency, radiologists should be aware of this entity and its distinguishing imaging features. Color Doppler examination can ascertain absence/reduction of blood flow.

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Institution

(Klang, Kanaan, Soudack, Kleinbaum, Heiman, Raskin, Amitai) Department of Radiology, Chaim Sheba Medical Center, Tel Aviv University, Tel Aviv, Israel

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

Year of Publication

2017

393.

Testicular torsion in undescended testis: A persistent challenge.

Naouar S., Braiek S., El Kamel R.

Embase

Asian Journal of Urology. 4 (2) (pp 111-115), 2017. Date of Publication: April 2017.

[Article]

AN: 613179479

Objective To evaluate the management and outcomes of patients who presented with torsion of an undescended testis and review the reported series in the literature. **Methods** The case records of 13 patients operated for testicular torsion involving undescended testis were retrospectively reviewed. The medical records included age at presentation, medical history, physical examination, operative findings and the results of follow-up. The diagnosis of torsion of undescended testis was made clinically and confirmed by inguinal exploration. **Results** In six cases the testis was preserved and orchiopexy was performed, while in seven cases orchidectomy was performed due to testicular gangrene in six patients and testicular tumor discovered peroperatively in one case. Mean duration of symptoms at time of surgery in the orchiopexy group was 6.5 h and in the orchidectomy group was 21.2 h. From six patients treated by orchiopexy, two patients suffered from testicular atrophy at a mean of 24 months. **Conclusion** Testicular torsion in undescended testis is still diagnosed with delay which may affect testicular salvage. The importance of examination of external genital organs is highlighted which should be routinely included by emergency physicians in physical examination for abdominal or groin pain.

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Institution

(Naouar, Braiek, El Kamel) Urology Department, Les Aghlabites Surgical Division, Ibn El Jazzar Teaching Hospital, Kairouan, Tunisia

Publisher

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2017

Does the internal inguinal ring need closure during laparoscopic orchiopexy with Prentiss manoeuvre?.

Narayanan S.K., Puthenvariath J.N., Somnath P., Mohanan A.

Embase

International Urology and Nephrology. 49 (1) (pp 13-15), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 612906477

Background: Undescended testis is a common problem, which is prevalent in 3 % of male infants. This study aimed to determine the effect of leaving the deep inguinal ring (DIR) without closure during laparoscopic orchiopexy (LO), with regard to post-operative hernia formation and other outcomes.

Method(s): From 2012 to 2014, 63 testicular units were managed with laparoscopy for non-palpable testis (NPT). Diagnostic laparoscopy was performed for all NPTs, and when they were intra-abdominal (42 testicular units), the DIR was left open after mobilization of the testis into the scrotum medial to the inferior epigastric vessels (Prentiss manoeuvre). We followed up these cases to check for hernia formation.

Result(s): The ages ranged from 10 months to 11 years with mean age at 3.7 years. Clinically, no cases presented with hernia, hydrocele or any other complications during a mean follow-up period of 34.4 months.

Conclusion(s): Closing the peritoneum over the DIR might be omitted in LO with Prentiss manoeuvre, saving operative time and effort. By doing so, there is no risk of hernia formation.

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Institution

(Narayanan, Puthenvariath, Somnath, Mohanan) Department of Pediatric Surgery, Institute of Maternal and Child Health, Government Medical College, Kozhikode, Kerala 673008, India

Publisher

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2017

395.

Age at referral for undescended testes: Has anything changed in a decade?.

Bajaj M., Upadhyay V.

Embase

New Zealand Medical Journal. 130 (1457) (pp 45-49), 2017. Date of Publication: 16 Jun 2017.

[Article]

AN: 616912508

AIM: Undescended testis (UDT) affects 1-6% of males and is one of the most common disorders in paediatric surgery. Updated consensus guidelines now recommend surgical management of UDT by 18 months. We compare the age at referral and subsequent timing of orchiopexy with data published from 1996-1998 at our institution, prior to the advent of updated guidelines.

METHOD(S): A retrospective review of all patients undergoing an orchiopexy for UDT from 2014 to 2016 was conducted. The age at time of first referral, first outpatient review and age at date of surgery were recorded. Calculations were made for time between referral and clinic visit (T-1) and between clinic visit and surgery (T-2). Data are reported as median (range).

RESULT(S): In the 2014-2016 group (n=216), the median age at time of referral was 5.3 (range 0-182) months. Following referral, children were seen in the clinic at a median interval 1.84 (T-1: range 0.16-17) months. The median interval between the clinic visit and operation was 2.95 (T-2: range 0-30.7) months. The median age at time of surgery was 12.6 (range 4.6-191.3) months. Compared to the data from 1996-1998 (n=325), there was a drop in the median ages both at time of referral (23 months vs 5.3) and at time of operation (38.8 months vs 12.6). In this cohort, 66% (n=143) of boys had surgery before eighteen months of age. The median times between referral and clinic visit (T-1: 1.7 months vs 1.84) and between clinic and operation (T-2: 3.3 months vs 2.95) were essentially unchanged.

CONCLUSION(S): Our second snapshot in time (2014-2016) shows improvements in median age at referral (under six months) and age at time of operation (at 12.6 months) when compared to the older snapshot (1996-1998). These timings are more in keeping with recommendations for orchiopexy.

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Institution

(Bajaj, Upadhyay) Paediatric Surgery Department, Starship Children's Hospital, 2 Park Road, Grafton, Auckland 1023, New Zealand

Publisher

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2017

396.

Testicular growth and spermatogenesis: new goals for pubertal hormone replacement in boys with hypogonadotropic hypogonadism? -a multicentre prospective study of hCG/rFSH treatment outcomes during adolescence-.

Rohayem J., Hauffa B.P., Zacharin M., Kliesch S., Zitzmann M., Wusthof A., Tittel B., Morlot M., Vorhoff W., Muller-Rossberg E., Wagner V., Konrad K., Salzgeber K., Fricke-Otto S., Wolfle J., Willig P., Karges B., Lankes E., Droste M., Richter-Unruh A., Liesenkotter K.-P., Hubner A., Zichner S., Welzel M., Dunstheimer D., Potelajew U., Heger S., Hammer E., Oevering R., Schmidt S., Nieschlag E.

Embase

Clinical Endocrinology. 86 (1) (pp 75-87), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 612108612

Context/objective: Testosterone treatment for pubertal induction in boys with hypogonadotropic hypogonadism (HH) provides virilization, but does not induce testicular growth or fertility. Larger studies evaluating the outcomes of gonadotropin replacement during adolescence have not been reported to date; whether previous testosterone substitution affects testicular responses is unresolved. We aimed to assess the effects of human chorionic gonadotropin (hCG) and recombinant FSH (rFSH) in boys and adolescents with HH with respect to a) testicular growth, b) spermatogenesis, c) quality of life (QoL) and to identify factors influencing therapeutic success.

Design/setting: A prospective case study was conducted in 26 paediatric endocrine centres.

Patients/interventions: HCG and rFSH were administered until cessation of testicular growth and plateauing of spermatogenesis to (1) prepubertal HH boys with absent or early arrested puberty

(group A) and to (2) HH adolescents who had previously received full testosterone replacement (group B). Outcome measures: Bi-testicular volumes (BTVs), sperm concentrations and QoL. Result(s): Sixty (34 A/26 B) HH patients aged 14-22 years were enrolled. BTVs rose from 5 +/- 5 to 34 +/- 3 ml in group A vs 5 +/- 3 to 32 +/- 3 ml in group B, with normal final BTVs (≥ 24 ml) attained in 74%/70% after 25/23 months in A/B, respectively. Sperm in the ejaculate were found in 21/23(91%)/18/19(95%), with plateauing concentrations after 31/30 months of hCG and 25/25 months of combined treatment in A/B. Sperm concentrations were normal (≥ 15 mill/ml) in 61%/32%, with mean concentrations of 40 +/- 73 vs 19 +/- 38 mill/ml in A/B (n.s.). Outcomes were better in patients without bilateral cryptorchidism, with non-congenital HH causes, higher baseline BTVs, and higher baseline inhibin B and AMH levels. QoL increased in both groups. Conclusion(s): HCG/rFSH replacement during adolescence successfully induces testicular growth and spermatogenesis, irrespective of previous testosterone replacement, and enhances QoL.

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Institution

(Rohayem, Kliesch, Zitzmann) Center of Reproductive Medicine and Andrology, Clinical Andrology, University of Munster, Muenster, Germany (Hauffa) Children's Hospital Essen, Department of Paediatric Endocrinology and Diabetes, University Duisburg-Essen, Essen, Germany

(Zacharin) The Royal Children's Hospital, Melbourne, VIC, Australia

Publisher

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Year of Publication

2017

397.

Sphingosine-1-phosphate lyase mutations cause primary adrenal insufficiency and steroid-resistant nephrotic syndrome.

Prasad R., Hadjidemetriou I., Maharaj A., Meimaridou E., Buonocore F., Saleem M., Hurcombe J., Bierzynska A., Barbagelata E., Bergada I., Cassinelli H., Das U., Sgene G., Krone R., Hacıhamdioglu B., Sari E., Yesilkaya E., Storr H.L., Clemente M., Fernandez-Cancio M., Camats N., Ram N., Achermann J.C., Van Veldhoven P.P., Guasti L., Braslavsky D., Guran T., Metherell L.A.

Embase

Journal of Clinical Investigation. 127 (3) (pp 942-953), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 614961819

Primary adrenal insufficiency is life threatening and can present alone or in combination with other comorbidities. Here, we have described a primary adrenal insufficiency syndrome and steroid-resistant nephrotic syndrome caused by loss-of-function mutations in sphingosine-1-phosphate lyase (SGPL1). SGPL1 executes the final decisive step of the sphingolipid breakdown pathway, mediating the irreversible cleavage of the lipid-signaling molecule sphingosine-1-phosphate (S1P). Mutations in other upstream components of the pathway lead to harmful accumulation of lysosomal sphingolipid species, which are associated with a series of conditions known as the sphingolipidoses. In this work, we have identified 4 different homozygous mutations, c.665G>A (p.R222Q), c.1633-1635delTTC (p.F545del), c.261+1G>A (p.S65Rfs6), and c.7dupA (p.S3Kfs11), in 5 families with the condition. In total, 8 patients were investigated, some of whom also manifested other features, including ichthyosis, primary hypothyroidism, neurological symptoms, and cryptorchidism. Sgpl1^{-/-} mice recapitulated the main characteristics of the human disease with abnormal adrenal and renal morphology. Sgpl1^{-/-} mice displayed disrupted adrenocortical zonation and defective expression of steroidogenic enzymes as well as renal histology in keeping with a glomerular phenotype. In summary, we have identified SGPL1 mutations in humans that perhaps represent a distinct multisystemic disorder of sphingolipid metabolism.

Status

Embase

Institution

(Prasad, Hadjidemetriou, Maharaj, Meimaridou, Storr, Guasti, Metherell) Centre for Endocrinology, William Harvey Research Institute, John Vane Science Centre, Queen Mary University of London, Charterhouse Square, London EC1M 6BQ, United Kingdom (Buonocore, Sgene, Achermann) Genetics and Genomic Medicine, UCL Great Ormond Street Institute of Child Health, University College London, London, United Kingdom (Saleem, Hurcombe, Bierzynska) Children's and Academic Renal Unit, University of Bristol, Bristol, United Kingdom

(Barbagelata) Servicio de Nefrologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina

(Bergada, Cassinelli, Braslavsky) Ctro. de Invest. Endocrinologicas Dr. Cesar Bergada- CONICET-FEI-Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina

(Das) Alderhey Children's Hospital NHS Foundation Trust, Eaton Road, Liverpool, United Kingdom

(Sgene) Center Is Detailed in the Supplemental Acknowledgments, United Kingdom

(Krone) Birmingham Children's Hospital, Birmingham, United Kingdom

(Hacihamdioglu) Health Sciences University, Suleymaniye Maternity and Children's Training and Research Hospital, Department of Paediatric Endocrinology and Diabetes, Istanbul, Turkey

(Sari, Yesilkaya) Gulhane Military Medical School Department of Paediatric Endocrinology and Diabetes, Ankara, Turkey

(Clemente, Fernandez-Cancio, Camats) Growth and Development Research Unit, Vall d'Hebron Research Institute (VHIR), Hospital Vall d'Hebron, CIBERER, Instituto de Salud Carlos III, Barcelona, Spain

(Ram) Department of Medicine, Aga Khan University Hospital, Karachi, Pakistan

(Van Veldhoven) Laboratory of Lipid Biochemistry and Protein Interactions (LIPIT), Campus Gasthuisberg, KU Leuven, Leuven, Belgium

(Guran) Marmara University, Department of Paediatric Endocrinology and Diabetes, Istanbul, Turkey

Publisher

American Society for Clinical Investigation (E-mail: rhickam@the-jci.org)

Year of Publication

2017

398.

Advantage of early orchiopexy for undescended testis: Analysis of testicular growth percentage ratio in patients with unilateral undescended testicle.

Tseng C.-S., Chiang I.-N., Hong C.-H., Lu Y.-C., Hong J.-H., Chang H.-C., Huang K.-H., Pu Y.-S.

Embase

Scientific reports. 7 (1) (pp 17476), 2017. Date of Publication: 12 Dec 2017.

[Article]

AN: 628364071

This study reports the experience of our tertiary referral center and proposes a new indicator, the growth percentage ratio (GPR), for determining the optimal timing of surgical intervention. A retrospective review of boys who underwent orchiopexy for undescended testis from 2001 to 2013 was conducted. We analyzed testicular volumes in different age groups using the UDT to normally descended testis ratio and testicular GPR. A total of 134 boys with unilateral undescended testicle underwent regular ultrasonography follow-up examinations for more than a mean of 3.9 years. Forty-five (33.4%) of them underwent orchiopexy before the age of one year. Orchiopexy at this age resulted in a GPR (2.02 ± 0.40) that was significantly higher than the GPRs in the second ($1 < \text{age} \leq 2$ years, 1.25 ± 0.13 , $p=0.004$) and third ($\text{age} > 2$ years, 1.24 ± 0.14 , $p=0.008$) age groups. The undescended testicle grew faster when orchiopexy was performed before one year of age. Orchiopexy performed within one year from birth significantly accelerates the growth of the UDT, as determined using the GPR, compared to other age groups. The present clinical evidence indicates that orchiopexy should be performed before one year of age.

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Author NameID

Tseng, Chi-Shin; ORCID: <http://orcid.org/0000-0002-1971-8552>

Institution

(Tseng, Chiang, Lu, Hong, Chang, Huang, Pu) Department of Urology, National Taiwan University Hospital, Taipei, Taiwan (Republic of China) (Hong) Graduate Institute of Electronics Engineering, National Taiwan University, Taipei, Taiwan (Republic of China)

Publisher

NLM (Medline)

Year of Publication

2017

399.

Experimentally induced testicular dysgenesis syndrome originates in the masculinization programming window.

van den Driesche S., Kilcoyne K.R., Wagner I., Rebourcet D., Boyle A., Mitchell R., McKinnell C., Macpherson S., Donat R., Shukla C.J., Jorgensen A., Meyts E.R.-D., Skakkebaek N.E., Sharpe R.M.

Embase

JCI insight. 2 (6) (pp e91204), 2017. Date of Publication: 23 Mar 2017.

[Article]

AN: 626580861

The testicular dysgenesis syndrome (TDS) hypothesis, which proposes that common reproductive disorders of newborn and adult human males may have a common fetal origin, is largely untested. We tested this hypothesis using a rat model involving gestational exposure to dibutyl phthalate (DBP), which suppresses testosterone production by the fetal testis. We evaluated if induction of TDS via testosterone suppression is restricted to the "masculinization programming window" (MPW), as indicated by reduction in anogenital distance (AGD). We show that DBP suppresses fetal testosterone equally during and after the MPW, but only DBP exposure in the MPW causes reduced AGD, focal testicular dysgenesis, and TDS disorders (cryptorchidism, hypospadias, reduced adult testis size, and compensated adult Leydig cell failure). Focal testicular dysgenesis, reduced size of adult male reproductive organs, and TDS disorders and their severity were all strongly associated with reduced AGD. We related our findings to human TDS cases by demonstrating similar focal dysgenetic changes in testes of men with preinvasive germ cell neoplasia (GCNIS) and in testes of DBP-MPW animals. If our results are translatable to humans, they suggest that identification of potential causes of human TDS disorders should focus on exposures during a human MPW equivalent, especially if negatively associated with offspring AGD.

PMC Identifier

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Institution

(van den Driesche, Kilcoyne, Wagner, Rebourcet, Boyle, Mitchell, McKinnell, Macpherson, Sharpe) MRC Centre for Reproductive Health, Queen's Medical Research Institute, University of Edinburgh, Edinburgh, United Kingdom (Donat, Shukla) Edinburgh Urological Cancer Group, Department of Urology, Western General Hospital, Edinburgh, United Kingdom (Jorgensen, Meyts, Skakkebaek) Department of Growth & Reproduction, Copenhagen University Hospital (Rigshospitalet), Copenhagen, Denmark

Publisher

NLM (Medline)

Year of Publication

2017

400.

Laparoscopy in the management of impalpable testis (Series of 64 Cases).

Ismail K.A., Ashour M.H.M., El-Afifi M.A., Hashish A.A., El-Dosouky N.E., Negm M., Hashish M.S.

Embase

African journal of paediatric surgery : AJPS. 14 (4) (pp 65-69), 2017. Date of Publication: 01 Oct 2017.

[Article]

AN: 626126005

Background: The undescended testis represents one of the most common disorders of childhood. Laparoscopy has been widely used both in the diagnosis and treatment of non-palpable testis. In this study, we investigated and evaluated the usefulness of laparoscopy in the diagnosis and treatment of no palpable testis.

Patients and Methods: From January 2003 to January 2008, we used laparoscopy in the management of 64 patients with 75 impalpable testes. Their ages varied from 1 to 15 years (median age = 4.6 years). The site and the size of the testes were localised by abdominopelvic ultrasonography in all 64 children for accurate diagnosis. One stage laparoscopic orchiopexy was performed in 26 testes, staged Fowler-Stephens orchiopexy was underwent in 17 testes, while laparoscopic orchidectomy was done in 5 testes. Follow-up by clinical examination and colour Doppler ultrasound was performed in every patient who underwent orchiopexy.

Result(s): There were 11 patients with bilateral non-palpable testes. The overall diagnostic agreement of ultrasound with laparoscopy was seen in only 16 out of 75 testes (21.3%). The results of diagnostic laparoscopy were varied and showed various pathological. Conditions and positioned of the testes, such as 20 low intra-abdominal testes (26.6%), 17 testes were high intra-abdominal (22.7%), and 18 testes (24%) entered the inguinal canal. Associated inguinal hernia was present in 4 patients. After a mean follow-up period of 26 months (6 months - 5 years), all testes were in the bottom of the scrotum except 3 testes were retracted to the neck of the scrotum and atrophy of the testis occurred in 2 patients (2.7%).

Conclusion(s): Laparoscopy has proven to be the only diagnostic modality where the findings provide a clear dependable direction for the definitive management of impalpable testes, so it allows an accurate diagnosis and definitive treatment in the same sitting.

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Institution

(Ismail, Ashour, El-Affi, Hashish, El-Dosouky, Negm, Hashish) Department of General Surgery,
Section of Pediatric Surgery, Tanta University Hospital, Tanta, Egypt

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401.

Establishing disability weights for congenital pediatric surgical conditions: A multi-modal approach.

Poenaru D., Pemberton J., Frankfurter C., Cameron B.H., Stolk E.

Embase

Population Health Metrics. 15 (1) (no pagination), 2017. Article Number: 8. Date of Publication: 04 Mar 2017.

[Article]

AN: 614636414

Background: Burden of disease (BoD) as measured by Disability-Adjusted Life Years (DALYs) is one of the criteria for priority-setting in health care resource allocation. DALYs incorporate disability weights (DWs), which are currently expert-derived estimates or non-existent for most pediatric surgical conditions. The objective of this study is to establish DWs for a subset of key pediatric congenital anomalies using a range of health valuation metrics with caregivers in both high- and low-resource settings.

Method(s): We described 15 health states to health professionals (physicians, nurses, social workers, and therapists) and community caregivers in Kenya and Canada. The health states summaries were expert- and community-derived, consisting of a narrated description of the disease and a functional profile described in EQ-5D-5 L style. DWs for each health state were elicited using four health valuation exercises (preference ranking, visual analogue scale (VAS), paired comparison (PC), and time trade-off (TTO)). The PC data were anchored internally to the TTO and externally to existing data to yield DWs for each health state on a scale from 0 (health) to 1 (dead). Any differences in DWs between the two countries were analyzed.

Result(s): In total, 154 participants, matched by profession, were recruited from Kijabe, Kenya (n = 78) and Hamilton, Canada (n = 76). Overall calculated DWs for 15 health states ranged from 0.13 to 0.77, with little difference between countries (intra-class coefficient 0.97). However, DWs generated in Kenya for severe hypospadias and undescended testes were higher than Canadian-derived DWs ($p = 0.04$ and $p < 0.003$, respectively).

Conclusion(s): We have derived country-specific DWs for pediatric congenital anomalies using several low-cost methods and inter-professional and community caregivers. The TTO-anchored PC method appears best suited for future use. The majority of DWs do not appear to differ significantly between the two cultural contexts and could be used to inform further work of estimating the burden of global pediatric surgical disease. Care should be taken in comparing the DWs obtained in the current study to the existent list of DWs because methodological differences may impact on their compatibility.

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Institution

(Poenaru, Pemberton, Frankfurter, Cameron) McMaster University, McMaster Pediatric Surgery Research Collaborative, Dept. of Surgery, Hamilton, Canada (Poenaru) MyungSung Medical College, Addis Ababa, Ethiopia

(Poenaru) Montreal Children's Hospital, McGill University, Department of Surgery, 16-4200 Sherbrooke West, Montreal, Westmount, QC H3Z 1C4, Canada

(Pemberton, Cameron) McMaster University, Dept. of Clinical Epidemiology and Biostatistics, Hamilton, Canada

(Stolk) Erasmus Medical Center Rotterdam, Institute for Medical Technology Assessment, Rotterdam, Netherlands

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2017

Human exposure to environmental contaminants and congenital anomalies: a critical review.

Foster W.G., Evans J.A., Little J., Arbour L., Moore A., Sauve R., Andres Leon J., Luo W.

Embase

Critical Reviews in Toxicology. 47 (1) (pp 59-84), 2017. Date of Publication: 02 Jan 2017.

[Review]

AN: 614413436

Congenital anomalies are an important cause of infant mortality and disability. Developmental exposure to environmental contaminants is thought to increase the risk for congenital anomalies. Herein, we describe a critical review of the literature conducted between February and March 2014 yielding 3057 references from which 97 unique relevant articles published from 2003 through 2014 were evaluated. Common congenital anomalies including hypospadias, cryptorchidism, anogenital distance (AGD), congenital heart defects and oral clefts were well represented in the literature whereas other outcomes such as neural tube defects, limb deficiency defects and gastroschisis were rarely described. While definitions used for congenital anomalies and methods of ascertainment were usually consistent across studies, inconsistencies were frequently found in grouping of different congenital heart defects. Despite strong links between some congenital anomalies and parental occupation, these studies are unable to provide clear insight into the specific chemicals responsible owing to lack of direct measures of exposure. In comparison, data are mixed for contaminant exposures at concentrations representative of results from contemporary biomonitoring studies. Of the environmental contaminants studied, the association between phthalate exposures and developmental abnormalities of the male reproductive tract received the greatest attention. Important limitations of the literature studied relate to adequacy of sample size, absence of or weaknesses in exposure assessment methodologies, failure to account for biological plausibility and grouping of congenital anomalies with divergent mechanisms. We conclude that the literature is inadequate at this time to support a conclusion that exposure to environmental contaminants are or are not associated with increased risks for congenital anomalies in the general population.

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Status

Embase

Institution

(Foster) Department of Obstetrics & Gynecology, McMaster University, Hamilton, ON, Canada

(Evans) Department of Biochemistry and Medical Genetics, University of Manitoba, Winnipeg,

MB, Canada

(Little) School of Epidemiology, Public Health and Preventive Medicine, Faculty of Medicine,
University of Ottawa, Ottawa, ON, Canada

(Arbour) Department of Medical Genetics, University of British Columbia, Vancouver, BC, Canada

(Moore) Department of Paediatrics, University of Toronto and the Hospital for Sick Children,
Toronto, ON, Canada

(Sauve) Department of Community Health Sciences, University of Calgary, Calgary, AB, Canada

(Andres Leon, Luo) Maternal, Child and Youth Health Surveillance and Epidemiology Division,
Centre for Chronic Disease Prevention, Public Health Agency of Canada, Ottawa, ON, Canada

Publisher

Taylor and Francis Ltd (E-mail: healthcare.enquiries@informa.com)

Year of Publication

2017

403.

Delayed treatment of undescended testes may promote hypogonadism and infertility.

Rohayem J., Luberto A., Nieschlag E., Zitzmann M., Kliesch S.

Embase

Endocrine. 55 (3) (pp 914-924), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 614021669

Context: Undescended testes at birth may be caused by testosterone deficiency during fetal development. It is unclear whether the process of failed descent contributes to permanent endocrine impairment.

Objective(s): To evaluate the impact of age at treatment of undescended testes on endocrine and spermatogenic testicular function in middle-aged men.

Patients and Methods: Reproductive hormone and semen data of 357 men with previously undescended testes were evaluated with respect to age at correction of testicular position and compared to those of 709 controls with eutopic testes at birth and normozoospermia.

Result(s): Men with undescended testes had higher mean Luteinizing Hormone levels ($p < 0.0001$) and lower mean testosterone levels ($p = 0.003$) compared to controls. They also had lower bi-testicular volumes, higher Follicle Stimulating Hormone levels, and lower sperm concentrations (all $p < 0.0001$). Lowest mean sperm concentrations were found in subjects with

bilateral undescended testes. Normal sperm concentrations were found in 21 % of cases (in 27 % of men with unilateral and in 12 % with bilateral undescended testes), while oligozoospermia was diagnosed in 44 %, and azoospermia in 35 % (in 28 % with unilateral, 46 % with bilateral undescended testes). Subjects with reduced semen quality had higher gonadotropin levels than those with normozoospermia. Age at correction (median: 6 years (1-39)) was inversely correlated with bi-testicular volumes and sperm concentrations, and positively correlated with FSH and LH, but not with serum testosterone.

Conclusion(s): Latent, rarely decompensated hypogonadism is a potential long-term consequence of undescended testes, besides infertility and testicular cancer, preferentially affecting subjects with delayed or unsuccessful correction of testicular position. Impaired Leydig cell function is likely to contribute to compromised fertility. These observations support correction of cryptorchidism during early infancy.

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Status

Embase

Institution

(Rohayem, Luberto, Nieschlag, Zitzmann, Kliesch) Center of Reproductive Medicine and Andrology, Department of Clinical Andrology, University of Muenster, Albert-Schweitzer-Campus 1, Building D11, Muenster D-48149, Germany (Luberto) Unit of Endocrinology, Department of Biomedical, Metabolic and Neural Sciences, University of Modena and Reggio Emilia, Via Universita, 4, Modena, MO 41121, Italy

Publisher

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Year of Publication

2017

404.

Treatment of impalpable testis - One clinic's experience.

Osemlak P., Zadkowski T., Rogowski B., Cielecki C., Wieczorek A., Wozniak M., Jedrzejewski G., Ben-Skowronek I., Nachulewicz P.

Embase

Wideochirurgia I Inne Techniki Maloinwazyjne. 12 (2) (pp 166-171), 2017. Date of Publication: June 2017.

[Article]

AN: 617761633

Introduction: Undescended testes are recognised in 1% to 2% of boys during the first year of life, and about 20% of them are impalpable. Ultrasonography (US) may establish the localisation of the testis but the final diagnosis is usually determined laparoscopically.

Aim(s): To evaluate long-term results of laparoscopic treatment of boys with impalpable testes and sensitivity of preoperative ultrasound.

Material(s) and Method(s): Between 2011 and 2015, we operated on 545 boys with undescended testes. Sixty-two of them with 65 impalpable testes were treated laparoscopically - the study group. Mean age was 3.5 years. The study group was divided into 5 groups according to type of treatment. The volume and position of the operated gonad were assessed manually and by ultrasound.

Result(s): In group 1 testicular agenesis was observed in 19 patients. In group 2 revision of the inguinal canal revealed testicular agenesis in 7 and atrophy in 4 patients. In group 3 conversion to classic orchiopexy was performed in 10 patients. In group 4 one-stage orchiopexy was performed in 9 patients on 12 testes. In group 5 a two-stage F-S procedure was performed in 13 patients. Ten testes in group 4 had a volume in the normal range (84%) and also 10 testes in group 5 (77%).

Conclusion(s): Laparoscopy in impalpable testes is the procedure of choice and allows definitive management, even when conversion to open procedure is necessary. Sensitivity of preoperative ultrasound is generally about 60% for true intra-Abdominal testes, so diagnostic laparoscopy is necessary.

Status

Embase

Institution

(Osemlak, Zadkowski, Rogowski, Cielecki, Nachulewicz) Department of Paediatric Surgery and Traumatology, Medical University of Lublin, 6 Prof. A. Gebali St, Lublin 20-093, Poland

(Wieczorek, Wozniak, Jedrzejewski) Department of Paediatric Radiology, University Paediatric Hospital, Lublin, Poland

(Ben-Skowronek) Department of Paediatric Endocrinology and Diabetology with Endocrine - Metabolic Laboratory, Medical University of Lublin, Lublin, Poland

Publisher

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405.

Canadian Urological Association-Pediatric Urologists of Canada (CUA-PUC) guideline for the diagnosis, management, and followup of cryptorchidism.

Braga L.H., Lorenzo A.J., Romao R.L.P.

Embase

Canadian Urological Association Journal. 11 (7) (pp E251-E260), 2017. Date of Publication: July 2017.

[Article]

AN: 617349283

Status

Embase

Institution

(Braga) Division of Urology, McMaster Children's Hospital, McMaster University, Hamilton, ON, Canada (Lorenzo) Division of Urology, Hospital for Sick Children, University of Toronto, Toronto, ON, Canada

(Romao) Division of Urology, IWK Health Centre, Dalhousie University, Halifax, NS, Canada

Publisher

Canadian Urological Association (E-mail: josephine.sciortino@cua.org)

Year of Publication

2017

406.

Reproductive function in the sons of women who experienced stress due to bereavement before and during pregnancy: a nationwide population-based cohort study.

Plana-Ripoll O., Li J., Kesmodel U.S., Parner E., Olsen J., Basso O.

Embase

Fertility and Sterility. 107 (1) (pp 189-197.e8), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 613819931

Objective To estimate the association between prenatal exposure to maternal stress and reproductive disorders in Danish men, where prenatal stress exposure was defined as the mother's loss of a close relative during pregnancy or in the 12 months before conception. **Design** Population-based cohort study. **Setting** Not applicable. **Patient(s)** All males born in Denmark between 1973 and 2008 (n = 1,217,576) and observed for up to 39 years. **Intervention(s)** None. **Main Outcome Measure(s)** Male reproductive function, defined using a composite outcome including congenital malformations of genital organs, testicular cancer, diagnosis of male infertility, or assisted conception use due to male factor infertility. **Result(s)** In total, 28,986 men (2.4%) had been exposed to prenatal stress, and 62,929 (5.2%) experienced the composite outcome during the follow-up period. Prenatal exposure to stress was associated with an elevated risk of reproductive problems (hazard ratio [HR] 1.09; 95% CI, 1.04-1.15). The association was stronger when the exposure occurred during the first trimester of pregnancy, and for congenital malformations of genital organs. When focusing on infertility alone, we saw no evidence of increased risk (HR 0.90; 95% CI, 0.77-1.06). In addition, the probability of marrying a woman was lower for exposed men (HR 0.93; 95% CI, 0.89-0.98). **Conclusion(s)** Prenatal stress in the form of the mother's bereavement during the first trimester of pregnancy is associated with a higher risk of reproductive disorders from congenital malformations of the genital organs in the male offspring. The lack of an association between maternal bereavement and later infertility in the exposed male offspring may be due in part to the men's lower probability of attempting to have children.

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Status

Embase

Institution

(Plana-Ripoll) Section of Epidemiology, Department of Public Health, Aarhus University, Aarhus, Denmark (Li, Olsen) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

(Kesmodel) Department of Obstetrics and Gynecology, Herlev and Gentofte Hospital, Herlev, Denmark

(Parner) Section of Biostatistics, Department of Public Health, Aarhus University, Aarhus, Denmark

(Olsen) Department of Epidemiology, Fielding School of Public Health, University of California, Los Angeles, California, United States

(Basso) Department of Obstetrics and Gynecology, Research Institute of the McGill University Health Centre (MUHC), Montreal, Quebec, Canada

(Basso) Department of Epidemiology, Biostatistics and Occupational Health, McGill University, Montreal, Quebec, Canada

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407.

Pediatric Scrotal Pathology.

Emr B., Stanger J.

Embase

Current Treatment Options in Pediatrics. 3 (2) (pp 163-174), 2017. Date of Publication: 01 Jun 2017.

[Review]

AN: 616099948

The purpose of this article is to review the approach to pediatric patients with scrotal pathology and the associated causes including inguinal hernia, cryptorchidism, hydrocele, varicocele, testicular torsion, trauma, and tumors. The reader will understand the diagnostic and treatment options and need for urgent surgical referral. Research has demonstrated the safety and efficacy of laparoscopic hernia repair in pediatric patients. Studies have failed to clarify the optimal timing of neonatal hernia repair and indications for bilateral exploration. Current clinical practice guidelines for cryptorchidism recommend surgical referral by 6 months of age and discourage the use of ultrasound. Most scrotal complaints are managed with low morbidity and preservation of fertility. Scrotal complaints of pain, swelling, bulging, and masses are common in pediatric patients. Management should be done in conjunction with a pediatric surgeon or urologist. Research is required to facilitate practice guidelines for scrotal pathology and address the practice variation that still exists.

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Status

Embase

Institution

(Emr, Stanger) SUNY Upstate Medical University, 721 Irving Ave, Suite 401, Syracuse, NY
13210, United States

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408.

The clinical analysis of small supernumerary marker chromosomes in 17 children with mos 45,X/46,X,+mar karyotype.

Wang H., Wang T., Yang N., He Y., Chen L., Hong L., Shao X., Zhu H., Li H.

Embase

Oncology Letters. 13 (6) (pp 4385-4389), 2017. Date of Publication: June 2017.

[Article]

AN: 616496595

Small supernumerary marker chromosome (sSMC) is a type of structurally abnormal chromosome. In order to identify the origin, morphology and other characteristics of sSMCs in children with mos 45,X/46,X,+mar karyotype, 17 patients (16 females and 1 male) were analyzed. All patients underwent general physical examination, gonadal imaging and molecular cytogenetic analyses, including Giemsa banding, dual-color fluorescence in situ hybridization and detection of the sex-determining region Y gene by polymerase chain reaction. Cytogenetic analyses indicated sSMCs in 14/17 cases were derived from the X chromosome, of which 8 individuals presented with ring-shaped sSMCs and 6 with centric minute-shaped sSMCs. The remaining 3 cases were derived from the Y chromosome, and all presented with minute-shaped sSMCs. All female patients exhibited short stature, gonadal dysgenesis and other typical features of Turner syndrome. The male patient exhibited short stature, hypospadias and bilateral cryptorchidism. In conclusion, the majority of the sSMCs in patients with a mos 45,X/46,X,+mar karyotype were derived from the sex chromosomes. The molecular cytogenetic features of sSMCs may provide useful information for genetic counseling, prenatal diagnosis and individualized treatment.

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Status

Embase

Institution

(Wang, Yang, He, Chen, Hong, Shao, Zhu) Department of Clinical Laboratory, Children's Hospital of Soochow University, Suzhou, Jiangsu 215003, China (Wang, Li, Li) Center for Reproduction and Genetics, Suzhou Hospital Affiliated to Nanjing Medical University, Suzhou, Jiangsu 215002, China

Publisher

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409.

The diagnostic impact of testicular biopsies for intratubular germ cell neoplasia in cryptorchid boys and the subsequent risk of testicular cancer in men with prepubertal surgery for syndromic or non-syndromic cryptorchidism.

Osterballe L., Clasen-Linde E., Cortes D., Engholm G., Hertzum-Larsen R., Reinhardt S., Thorup J.

Embase

Journal of Pediatric Surgery. 52 (4) (pp 587-592), 2017. Date of Publication: 01 Apr 2017.

[Article]

AN: 613255147

Introduction Cryptorchidism is a risk factor for testicular cancer in adult life. It remains unclear how prepubertal surgery for cryptorchidism impacts later development of adult testicular cancer. The aim of study was to investigate tools to identify the cryptorchid boys who later develop testicular cancer. Methods The study cohort consisted of 1403 men operated prepubertally/pubertally for undescended testis between 1971 and 2003. At surgery testicular biopsies were taken from the cryptorchid testes. The boys were followed for occurrence of testicular cancer. The testicular cancer risk was compared to the risk in the Danish Population. Testicular biopsies from the boys who developed testicular cancer during follow-up underwent

histological examination with specific diagnostic immunohistochemical markers for germ cell neoplasia. Results The cohort was followed for 33,627 person years at risk. We identified 16 cases with testicular cancer in adulthood. The standardized incidence ratio was 2.66 (95% CI: 1.52-4.32). At time of primary surgery in prepubertal/pubertal age Intratubular Germ Cell Neoplasia (ITGCN) was diagnosed in 5 cases and the boys were unilaterally orchiectomized. At follow-up new immunohistochemical staining indicated ITGCN in two of the 16 cancer cases at reevaluation of the original biopsies from time of prepubertal/pubertal surgery. One had syndromic cryptorchid and developed seminoma, and another showed nonsyndromic cryptorchidism and developed embryonic teratocarcinoma. Totally, ITGCN was diagnosed in 0.5% (7/1403) of prepubertal cryptorchid boys, whereof 57% (4/7) in syndromic-cryptorchidism. Discussion ITGCN is predominantly observed prepubertally in boys with syndromic-cryptorchidism. In nonsyndromic cryptorchidism testicular cancer develops postpubertally, generally not based on dormant germ cells of ITGCN caused by an early fetal maldevelopment. Levels of Evidence LEVEL I.

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Embase

Institution

(Osterballe, Reinhardt, Thorup) The Department of Paediatric Surgery, Copenhagen University Hospital Rigshospitalet, Copenhagen DK-2100, Denmark (Clasen-Linde) The Department of Pathology, Copenhagen University Hospital Rigshospitalet, Copenhagen DK-2100, Denmark (Cortes) Section of Endocrinology, Department of Paediatrics, Copenhagen University Hospital, Hvidovre, Copenhagen, Denmark

(Cortes, Thorup) Faculty of Health and Medical Sciences, University of Copenhagen, Denmark

(Engholm, Hertzum-Larsen) Department of Documentation & Quality, Danish Cancer Society, Copenhagen, Denmark

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W.B. Saunders

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410.

To 'Pex or Not to 'Pex: What to Do for the Contralateral Testis When a Nubbin Is Discovered.

Kehoe J.E., Christman M.S.

Embase

Current Urology Reports. 18 (2) (no pagination), 2017. Article Number: 9. Date of Publication: 01 Feb 2017.

[Review]

AN: 614471487

Purpose of Review: Testicular remnants or nubbins are commonly found in the evaluation and treatment of cryptorchidism. While much debate focuses on the management of the nubbin itself, there is also great uncertainty and variation in the management of the contralateral descended testis. Herein, we review the relevant literature informing the decision to perform a contralateral orchiopexy. Recent Findings: Although there is very little recent literature directly addressing the question, some studies have better characterized differences in practice, the risk of intravaginal torsion in the contralateral testis and potential consequences in the selection of technique.

Summary: The etiology of a vanishing testis remains obscure, but appears more likely to be the result of a prenatal extravaginal torsion. While indeterminate, the risk of contralateral torsion of a descended testis appears to concentrate around the neonatal period with no substantially increased risk in later years. Contralateral orchiopexy, although a low-risk procedure, likely benefits very few and may carry an as yet poorly described risk to the contralateral testicle depending on the technique of fixation.

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Institution

(Kehoe, Christman) Department of Urology, Naval Medical Center San Diego, 34800 Bob Wilson Drive, San Diego, CA 92134, United States

Publisher

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411.

Seasonal trends in the prevalence of hypospadias: Aetiological implications.

Mamoulakis C., Avgenakis G., Gkatzoudi C., Duijker G., Zisis I.E., Heretis I., Antypas S., Sofikitis N., Spandidos D.A., Tsatsakis A.M., Tzonou A.

Embase

Experimental and Therapeutic Medicine. 13 (6) (pp 2960-2968), 2017. Date of Publication: June 2017.

[Article]

AN: 616123249

The aim of the present study was to examine the seasonality of hypospadias in Greece in an attempt to elucidate the aetiology. All boys born between 1991-1998, who underwent hypospadias repair at 'Aghia Sophia' Children's Hospital, Athens (n=542) were analysed. All Greek live-born males during the same period (population at risk; m=421,175) served as the controls. Seasonality by month of birth was evaluated with specific statistical tools. Meteorological parameters were also analysed. All tests yielded significant results, suggesting a simple harmonic prevalence pattern (highest/lowest: autumn, peak in October/spring, trough in April). Therefore, the first trimester of hypospadiac gestations coincides more frequently with winter. Meteorological parameters varied seasonally (maximal sunlight; air temperature in summer/minimal in winter, maximal rainfall in winter/minimal in summer) and were strongly associated pairwise.

Hypospadiac birth prevalence follows a simple harmonic seasonal pattern and is associated with that of cryptorchidism in Greece. The coincidence of the first or third trimester of a potentially genetically influenced gestation with winter could lead to the phenotypic expression of hypospadias or cryptorchidism, respectively. The potential role of a cyclic-varied androgen-production stimulator, such as human chorionic gonadotrophin may be speculated. The seasonality of a common environmental factor acting directly/indirectly may contribute to these patterns, and possibly to the common pathogenesis of these congenital malformations.

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Status

Embase

Institution

(Mamoulakis, Avgenakis, Zisis, Heretis) Department of Urology, University General Hospital of Heraklion, University of Crete Medical School, Heraklion 71003, Greece (Gkatzoudi, Duijker) Clinic of Social and Family Medicine, Faculty of Medicine, University of Crete, Heraklion 71003, Greece

(Antypas) First Pediatric Surgery Clinic, Aghia Sophia Children's Hospital, Athens 11527, Greece

(Sofikitis) Department of Urology, University of Ioannina, Medical School, Ioannina 45110, Greece

(Spandidos) Laboratory of Clinical Virology, Medical School, Faculty of Medicine, University of Crete, Heraklion 71003, Greece

(Tsatsakis) Department of Forensic Sciences and Toxicology, Faculty of Medicine, University of Crete, Heraklion 71003, Greece

(Tzonou) Department of Hygiene, Epidemiology and Medical Statistics, University of Athens, Medical School, Athens 11527, Greece

Publisher

Spandidos Publications (10 Vriaxidos Street, Athens 116 10, Greece. E-mail: subscriptions@spandidos-publications.com)

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412.

Age-specific foreskin development before adolescence in boys.

Yu S.H., Yu H.S., Kim S.-O.

Embase

Current Pediatric Research. 21 (1) (pp 136-139), 2017. Date of Publication: 2017.

[Article]

AN: 615634031

Purpose: We examined the external genitalia of boys to estimate the age-specific prevalence of foreskin development before adolescence.

Material(s) and Method(s): A total of 189 boys aged 0 to 13 years were enrolled in this study. The boys were categorized into four groups according to their age (group 1-4). The foreskin condition was classified as type I (normal prepuce), type II (adhesion of prepuce), type III (partial phimosis) and type IV (phimosis). Other abnormalities of the genitalia were also recorded. All the examinations were performed by the same urologist.

Result(s): The incidence of type I foreskin was 46.6% in group 1 (age 0-1 year), 50.6% in group 2 (2-5 years), 77.3% in group 3 (6-9 years) and 46.2% in group 4 (10-13 years). The incidence of type IV foreskin was 20.7% in group 1, 19.3% in group 2, 4.5% in group 3 and 53.8% in group 4.

Of the genital abnormalities, cryptorchidism was the most common (n=96), followed by hydrocele (n=61).

Conclusion(s): Physiologic phimosis showed a tendency to decrease with age up to 10 years.

Most of the boys with phimosis did not require treatment.

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Status

Embase

Institution

(Yu, Yu, Kim) Department of Urology, Chonnam National University Medical School, Gwangju,

South Korea

Publisher

Scientific Publishers of India (E-mail: qayyum@del3.vsnl.net.in)

Year of Publication

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413.

Inguinoscrotal pathology.

Guerra L., Leonard M.

Embase

Canadian Urological Association Journal. 11 (1-2 Supplement 1) (pp S41-S46), 2017. Date of

Publication: January-February 2017.

[Review]

AN: 615268512

Infants, children, and adolescents with inguinoscrotal pathology comprise a significant proportion of emergency department and outpatient visits. Visits to the emergency department primarily comprise individuals presenting with scrotal pain due to testicular torsion or torsion of the testicular appendages. At such time, immediate urological consultation is sought. Outpatient visits comprise those individuals with undescended testes, hydroceles, and varicoceles. Rare, but important problems, such as pediatric testicular tumours, may also present in the office setting. Many of these outpatient visits are to primary care physicians, who should have an appreciation of the timing and need for referral. The purpose of this review is to familiarize the general urologist and primary care physician with these varied pathologies and give insight into their

assessment and management. Some of these same conditions are seen in adult patients, but there are some significant differences in their management in the pediatric group. In addition, the utility of imaging studies, such as ultrasound, are discussed within each pathological entity. It is hoped that this overview will assist our general urology and primary care colleagues in patient management for diverse inguinoscrotal pathologies.

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Status

Embase

Institution

(Guerra, Leonard) Department of Surgery, Division of Pediatric Urology, Children's Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON, Canada

Publisher

Canadian Urological Association (E-mail: josephine.sciortino@cua.org)

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414.

Cryptorchidism: A practical review for all community healthcare providers.

Braga L.H., Lorenzo A.J.

Embase

Canadian Urological Association Journal. 11 (1-2 Supplement 1) (pp S26-S32), 2017. Date of Publication: January-February 2017.

[Review]

AN: 615268505

Cryptorchidism is one of the most common congenital anomalies of the male genitalia, occurring in 1% of boys by the age of one year. Even though the etiology of cryptorchidism is multifactorial, management has evolved with the clear recognition that hormonal treatment is not effective and surgery between 6-18 months of age leads to better testicular outcomes. Diagnostic laparoscopy is considered the standard approach for management of non-palpable testes, and can be combined with one or two-stage orchidopexy, with up to 80-90% success rates. This review discusses the natural history of retractile testicles, indications for hormonal treatment and

orchidectomy, ultrasound's role as a diagnostic tool, risks of infertility and testicular cancer, and surgical techniques for inguinal and intra-abdominal testes.

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Status

Embase

Institution

(Braga) Department of Surgery, Division of Urology, McMaster University, Hamilton, ON, Canada

(Lorenzo) Division of Urology, Hospital for Sick Children, Toronto, ON, Canada

Publisher

Canadian Urological Association (E-mail: josephine.sciortino@cua.org)

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2017

415.

Current Practice for Cryptorchidism: Survey of Pediatric Urologists.

Griffin D.L., Cambareri G., Kaplan G.

Embase

Urology Practice. 4 (3) (pp 245-250), 2017. Date of Publication: May 2017.

[Article]

AN: 615088896

Introduction We evaluated current practice patterns in the management and screening of cryptorchidism among pediatric urologists and compared them to the 2014 AUA (American Urological Association) guidelines on cryptorchidism. Methods A 14-question survey was disseminated to members of the SPU (Society for Pediatric Urology) to assess their current practice patterns. Results There were 187 responses (38.4% response rate) with a notable bimodal distribution in respondent practice years, with 39.3% in practice for 10 years or less and 41.4% in practice for more than 20 years. Despite guideline recommendations against the use of ultrasound, the majority of respondents will use it in cases of obesity or bilateral nonpalpable testes (greater than 50%). In the evaluation for bilateral nonpalpable testes most respondents (greater than 80%) perform an endocrine workup. Nevertheless, 55.1% will proceed with surgical exploration even if the workup indicates absence of testicular tissue. Subgroup analysis revealed those in practice for 10 years or less vs greater than 20 years were more likely to perform 2-stage

Fowler-Stephens in cases of short vessels (80.8% vs 58.1%) and to perform transscrotal orchiopexy in cases amenable to that approach (79.5% vs 52%). If examination under anesthesia reveals the testicle in the scrotum, 46.5% still perform orchiopexy, citing concerns the testicle will ascend or parental concerns about the diagnosis. Conclusions Based on the responses some discordance exists between practice patterns and guideline recommendations. Although surgery may be avoided with endocrine evaluation for suspected anorchia, many respondents will still perform exploration. The evaluation, management and surgical approach in cryptorchidism may be influenced by years in practice.

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Status

Embase

Institution

(Griffin) Department of Urology, Naval Medical Center San Diego, San Diego, California, United States (Cambareri) Department of Surgery, Division of Urology, University of California, San Diego, California, United States

(Kaplan) Department of Urology, Rady Children's Hospital-San Diego, San Diego, California, United States

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416.

Efficiency of combined diffusion weighted imaging and conventional MRI in detection of clinically nonpalpable undescended testes.

Ali S.A., Mansour M.G.

Embase

Egyptian Journal of Radiology and Nuclear Medicine. 48 (1) (pp 251-256), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 613557926

Purpose The goal of the study was to highlight the added value of combined DWI and conventional MRI in detecting clinically nonpalpable undescended testes. Patients and methods
Prospective study included 60 males referred for MRI evaluation of clinically diagnosed 66 nonpalpable undescended testes. MRI studies were performed using 1.5-T MRI machine and included axial and coronal spin-echo T1WIs, axial T2WIs, axial and coronal fat suppressed spin-echo T2WIs, and axial DWIs using three sets of b value (50, 400, and 800 s/mm²). All images were transferred to an independent workstation and evaluated by two radiologists for the presence or absence and location of the undescended testes. The findings were compared to laparoscopy results, and then, sensitivity, specificity, and accuracy were calculated for both conventional and combined (DWI and conventional) MRI. Results According to laparoscopic findings, sensitivity, specificity and accuracy of conventional MRI were 73.91%, 100%, and 80% and 69.57%, 100%, and 76.67% for radiologists 1 and 2 respectively, and of combined MRI were 86.9%, 100%, and 90% and 82.61%, 100%, and 86.67% for radiologists 1 and 2 respectively.
Conclusion Adding DWIs to conventional MRI improves the sensitivity and accuracy of detecting clinically nonpalpable undescended testes.

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Status

Embase

Institution

(Ali, Mansour) Radiodiagnosis Department, Ain Shams University Cairo, Egypt

Publisher

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Year of Publication

2017

417.

Early life risk factors for testicular cancer: a case-cohort study based on the Copenhagen School Health Records Register.

Piltoft J.S., Larsen S.B., Dalton S.O., Johansen C., Baker J.L., Cederkvist L., Andersen I.

Embase

Acta Oncologica. 56 (2) (pp 220-224), 2017. Date of Publication: 01 Feb 2017.

[Article]

AN: 614071581

Purpose: One established risk factors for testicular cancer is cryptorchidism. However, it remains unclear whether cryptorchidism is a risk factor in itself or whether the two conditions share common causes in early life (estrogen hypothesis), such as birth weight and birth order. The objective of this study is to utilize data from the Copenhagen School Health Records Register (CSHRR) to evaluate cryptorchidism, birth weight and birth order as risk factors for testicular cancer.

Method(s): The study population consisted of 408 cases of testicular cancer identified by a government issued identification number linkage of the entire CSHRR with the Danish Cancer Registry and a random subsample of 4819 males from the CSHRR. The study design was case-cohort and the period of follow-up between 2 April 1968 and 31 December 2003.

Result(s): Cryptorchidism was significantly associated with testicular cancer in crude analyses [hazard ratio (HR) = 3.60, 95% CI 2.79-4.65]. Birth weight was inversely associated with testicular cancer and no clear association with birth order was observed. The positive association between cryptorchidism and testicular cancer was only slightly attenuated controlling for birth weight and birth order and stratified on birth cohort (HR = 3.46, 95% CI 2.67-4.48).

Conclusion(s): This study confirmed the robustness of the association between cryptorchidism and testicular cancer even after adjustment for birth weight and birth order. Furthermore, the study showed an inverse association between birth weight and testicular cancer.

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PMC Identifier

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Status

Embase

Author NameID

Larsen, Signe Benzon; ORCID: <http://orcid.org/0000-0001-9522-7678> Baker, Jennifer L.;

ORCID: <http://orcid.org/0000-0002-9649-6615>

Andersen, Ingelise; ORCID: <http://orcid.org/0000-0002-0076-265X>

Institution

(Piltoft, Larsen, Dalton, Johansen, Cederkvist) The Unit of Survivorship, The Danish Cancer Society Research Center, Copenhagen, Denmark (Johansen) Oncology Clinic, Finsen Centre, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark

(Baker) Institute of Preventive Medicine, Bispebjerg and Frederiksberg Hospital, The Capital Region, Copenhagen, Denmark

(Andersen) University of Copenhagen, Institute of Public Health, Section of Social Medicine, CSS, Copenhagen K, Denmark

Publisher

Taylor and Francis Ltd (E-mail: healthcare.enquiries@informa.com)

Year of Publication

2017

418.

A preliminary study of shear wave elastography for the evaluation of unilateral palpable undescended testes.

Ucar A.K., Alis D., Samanci C., Aslan M., Habibi H.A., Dikici A.S., Namdar Y., Gultekin M.H., Onal B., Adaletli I.

Embase

European Journal of Radiology. 86 (pp 248-251), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 613448934

Objectives We sought to compare unilateral palpable undescended testes and contralateral descended testes using shear wave elastography (SWE) to show potential quantitative differences in elasticity patterns, which might reflect the histologic features. **Methods** Approval for this prospective study was obtained from the local ethics committee. A total of 29 patients (mean age, 7.52 years; range, 1-18 years) with unilateral palpable undescended testes and contralateral descended testes were examined by greyscale ultrasonography and SWE between February 2015 and April 2016. The volume and the elasticity of each testicle were the main factors evaluated. **Results** There was no difference between undescended testes and contralateral descended testes in terms of volume. However, a significant difference was evident in SWE-derived quantitative data. **Conclusions** SWE seems to be a useful sonographic technique to predict histologic features of the undescended testicle, which might replace testicular biopsy in modern management of the undescended testis.

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PMC Identifier

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Embase

Institution

(Ucar, Alis, Samanci, Aslan, Habibi, Dikici, Namdar, Adaletli) Istanbul University, Cerrahpasa Faculty of Medicine, Department of Radiology, KMPasa, Istanbul 34098, Turkey (Gultekin, Onal) Istanbul University, Cerrahpasa Faculty of Medicine, Department of Urology, KMPasa, Istanbul 34098, Turkey

Publisher

Elsevier Ireland Ltd

Year of Publication

2017

419.

High single scrotal-incision orchidopexy as the standard technique in infants aged 6-24 months. Mohey A., Gharib T.M., Omar R.G., Sebaey A., Elmohamady B.N., Kandeel W.

Embase

Arab Journal of Urology. 15 (1) (pp 78-81), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 614374368

Objective To prospectively investigate the effectiveness of high single scrotal-incision orchidopexy (HSSIO) for palpable undescended testis (PUDT) in infants aged 6-24 months. **Patients and methods** From March 2012 to July 2014, 46 age range-restricted (6-24 months) infants with 57 PUDT underwent HSSIO after obtaining written consent from their parents. The exclusion criteria were ectopic, retractile testes and recurrent cases. All infants were examined before surgery in the outpatient department and after anaesthesia induction immediately before surgery. All infants had general anaesthesia with a caudal block. The operative time, intraoperative and postoperative complications, and follow-up of the infants at 0.5, 3 and 6 months were recorded and analysed. **Results** The mean (SD; range) operative time was 23.45 (3.28; 18-29) min. A hernia sac was found in 39 (68.4%) UDTs. For postoperative complications, only one infant developed a scrotal haematoma that was managed conservatively. The procedure was successful in 56/57 PUDT (98%). An auxiliary procedure was needed in one case, to obtain more length of the cord by extension of the incision to the external ring. **Conclusion** HSSIO is a safe and feasible technique, with many benefits, and as such should be considered as the standard technique for orchidopexy in infants aged 6-24 months.

Copyright © 2017 Arab Association of Urology

Status

Embase

Institution

(Mohey, Gharib, Omar, Sebaey, Elmohamady, Kandeel) Department of Urology, Benha University Hospital, Faculty of Medicine, Benha University, Benha, Egypt

Publisher

Arab Association of Urology (E-mail: araburo@yahoo.com)

Year of Publication

2017

420.

Mutational analysis of HOXA10 gene in Chinese patients with cryptorchidism.

Cheng Z., Wang M., Xu C., Pei Y., Liu J.C., Huang H., He D., Lu P.

Embase

Andrologia. 49 (1) (no pagination), 2017. Article Number: e12592. Date of Publication: 01 Feb 2017.

[Article]

AN: 614078820

Cryptorchidism is one of the most common congenital anomalies and affects 2-4% of full-term new born boys. Its aetiology is poorly understood at present. HOXA10 plays a pivotal role in regulation of testicular descent. Male mice mutant for Hoxa10 exhibit unilateral or bilateral cryptorchidism as a result of impaired development of the gubernaculum. In this study, we performed mutation analysis of HOXA10 gene in a cohort of 98 cryptorchid patients. And we found a mutation (N27K) in a boy with unilateral cryptorchidism. The mutation was not detected in 106 healthy controls. Both in silico analyses and functional studies showed that the mutation affected the function of HOXA10. The results demonstrated that mutation in HOXA10 gene contributes to the pathogenesis of cryptorchidism, but may not be a common cause.

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PMC Identifier

27108669 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27108669>]

Status

Embase

Institution

(Cheng, Pei, Liu, Huang) School of Basic Medical Sciences, Chongqing Medical University, Chongqing, China (Wang) Department of Obstetrics and Gynecology, Second Affiliated Hospital, Chongqing Medical University, Chongqing, China

(Xu) Key Laboratory of Molecular Biophysics of the Ministry of Education, College of Life Science and Technology, Center for Human Genome Research and Cardio-X Institute, Huazhong University of Science and Technology, Wuhan, China

(He, Lu) Department of Pediatric Research Institute, Children's Hospital of Chongqing Medical University, Ministry of Education Key Laboratory of Child Development and Disorders, Chongqing, China

Publisher

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Year of Publication

2017

421.

Cryptorchidism: experience and reason.

Attalla K., Arnone E., Williot P., Greenfield S.P.

Embase

The Canadian journal of urology. 24 (4) (pp 8941-8945), 2017. Date of Publication: 01 Aug 2017.

[Article]

AN: 621895601

INTRODUCTION: To characterize our contemporary clinical experience with cryptorchidism.

MATERIALS AND METHODS: The records of boys referred for cryptorchidism were reviewed from 2001 to 2011. Data regarding the incidence of retractile testes, testicular ascent, surgical approach and outcomes were tabulated. Follow up was both early (< 12 weeks) and late (> 12 weeks).

RESULTS: A total of 1885 patients, or 2593 testes, were identified. Eight hundred and forty-one children (45%) or 1204 testes (46%) were retractile on initial exam--57% bilateral; 187 testes (7%) later 'ascended' on re-examination and underwent surgery--15% bilateral; 1340 (85%) testes were palpable in the inguinal canal and underwent inguinal orchidopexy--98% were successful; 69 (4%) of initially palpable testes were found to be atrophic and removed; 167 (11%) testes were

non-palpable and underwent laparoscopy-46 were atrophic and removed; 31 were vanishing; 33 were brought down using an inguinal approach at the same sitting with 97% success; 47 underwent staged Fowler-Stephens orchidopexy (FSO) and 10 underwent non-staged FSO, with 82% and 78% success respectively. All second stages were performed open.

CONCLUSIONS: Almost half of children referred for cryptorchidism had retractile testes. Surgery for later ascent was required in 16% of testes judged to be retractile at a median age of 8 years, emphasizing the need for repeat examination. High success rates with inguinal orchidopexy were achieved, even in non-palpable testes. Testes requiring FSO were uncommonly encountered- approximately 5 testes/year or 4% of testes undergoing surgery- and success was achieved in approximately 80%.

PMC Identifier

28832317 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28832317>]

Institution

(Attalla) Department of Pediatric Urology, Women & Children's Hospital of Buffalo, Buffalo, New York, USA

Year of Publication

2017

422.

GnRHa Treatment of Cryptorchid Boys Affects Genes Involved in Hormonal Control of the HPG Axis and Fertility.

Hadziselimovic F., Gegenschatz-Schmid K., Verkauskas G., Demougin P., Bilius V., Dasevicius D., Stadler M.B.

Embase

Sexual development : genetics, molecular biology, evolution, endocrinology, embryology, and pathology of sex determination and differentiation. 11 (3) (pp 126-136), 2017. Date of Publication: 2017.

[Article]

AN: 621606661

The gonadotropin-releasing hormone agonist (GnRHa; Buserelin) rescues fertility during adulthood in the majority of high infertility risk cryptorchid boys presenting with defective mini-puberty. However, the molecular events governing this effect are not understood. We report the

outcome of an RNA profiling analysis of testicular biopsies from 4 operated patients who were treated with GnRHa for 6 months versus 3 operated controls who were not treated. GnRHa induces a significant transcriptional response, including protein-coding genes involved in pituitary development, the hypothalamic-pituitary-gonadal axis, and testosterone synthesis. Furthermore, we observed an increased abundance of long noncoding RNAs (lncRNAs) participating in epigenetic processes, including AIRN, FENDRR, XIST, and HOTAIR. These data are consistent with the hypothesis that hypogonadotropic hypogonadism in boys with altered mini-puberty is the consequence of a profoundly altered gene expression program involving protein-coding genes and lncRNAs. Our results point to molecular mechanisms that underlie the ability of GnRHa to rescue fertility.

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PMC Identifier

28505621 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28505621>]

Institution

(Hadziselimovic) Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, Liestal, Switzerland

Year of Publication

2017

423.

Noonan Syndrome: An Underestimated Cause of Severe to Profound Sensorineural Hearing Impairment. Which Clues to Suspect the Diagnosis?.

Ziegler A., Loundon N., Jonard L., Cave H., Baujat G., Gherbi S., Couloigner V., Marlin S.

Embase

Otology & neurotology : official publication of the American Otological Society, American Neurotology Society [and] European Academy of Otology and Neurotology. 38 (8) (pp 1081-1084), 2017. Date of Publication: 01 Sep 2017.

[Review]

AN: 621474557

OBJECTIVE: To highlight Noonan syndrome as a clinically recognizable cause of severe to profound sensorineural hearing impairment. STUDY DESIGN: New clinical cases and review.

SETTING: Patients evaluated for etiological diagnosis by a medical geneticist in a reference center for hearing impairment.

PATIENTS: Five patients presenting with confirmed Noonan syndrome and profound sensorineural hearing impairment.

INTERVENTIONS: Diagnostic and review of the literature.

RESULTS: Five patients presented with profound sensorineural hearing impairment and molecularly confirmed Noonan syndrome. Sensorineural hearing impairment has been progressive for three patients. Cardiac echography identified pulmonary stenosis in two patients and was normal for the three other patients. Short stature was found in two patients. Mild intellectual disability was found in one patient. Inconspicuous clinical features as facial dysmorphism, cryptorchidism, or easy bruising were of peculiar interest to reach the diagnosis of Noonan syndrome.

CONCLUSION: Profound sensorineural hearing impairment can be the main feature of Noonan syndrome. Associated features are highly variable; thus, detailed medical history and careful physical examination are mandatory to consider the diagnosis in case of a sensorineural hearing impairment.

PMC Identifier

28742629 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28742629>]

Institution

(Ziegler) *APHP, Hopital Necker Enfants Malades, Centre de reference des surdités génétiques, Service de Genetique Medicale +APHP, Hopital Necker Enfants Malades, Service d'otorhinolaryngologie ++APHP, Hopital Necker Enfants Malades, Centre de reference des surdités génétiques, Laboratoire de Genetique moleculaire APHP, Hopital Robert Debre APHP, Hopital Necker-Enfants malades, Departement de Genetique, Paris, France

Year of Publication

2017

424.

Variants in congenital hypogonadotrophic hypogonadism genes identified in an Indonesian cohort of 46,XY under-virilised boys.

Ayers K.L., Bouty A., Robevska G., van den Bergen J.A., Juniarto A.Z., Listyasari N.A., Sinclair A.H., Faradz S.M.

Embase

Human genomics. 11 (1) (pp 1), 2017. Date of Publication: 16 Feb 2017.

[Article]

AN: 620722713

BACKGROUND: Congenital hypogonadotrophic hypogonadism (CHH) and Kallmann syndrome (KS) are caused by disruption to the hypothalamic-pituitary-gonadal (H-P-G) axis. In particular, reduced production, secretion or action of gonadotrophin-releasing hormone (GnRH) is often responsible. Various genes, many of which play a role in the development and function of the GnRH neurons, have been implicated in these disorders. Clinically, CHH and KS are heterogeneous; however, in 46,XY patients, they can be characterised by under-virilisation phenotypes such as cryptorchidism and micropenis or delayed puberty. In rare cases, hypospadias may also be present. **RESULTS:** Here, we describe genetic mutational analysis of CHH genes in Indonesian 46,XY disorder of sex development patients with under-virilisation. We present 11 male patients with varying degrees of under-virilisation who have rare variants in known CHH genes. Interestingly, many of these patients had hypospadias.

CONCLUSIONS: We postulate that variants in CHH genes, in particular PROKR2, PROK2, WDR11 and FGFR1 with CHD7, may contribute to under-virilisation phenotypes including hypospadias in Indonesia.

PMC Identifier

28209183 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28209183>]

Institution

(Ayers, Bouty, Robevska, van den Bergen, Sinclair) Murdoch Childrens Research Institute, Melbourne, Victoria, Australia (Ayers, Sinclair) Department of Paediatrics, University of Melbourne, Melbourne, Victoria, Australia

(Bouty) The Royal Children's Hospital, Melbourne, Victoria, Australia

(Juniarto, Listyasari, Faradz) Division of Human Genetics, Centre for Biomedical Research, Faculty of Medicine, Diponegoro University (FMDU), JL. Prof. H. Soedarto, SH, Tembalang, Semarang, 50275, Central Java, Indonesia

Year of Publication

2017

Epigenetic and risk factors of testicular germ cell tumors: a brief review.

Landero-Huerta D.A., Viguera-Villasenor R.M., Yokoyama-Rebollar E., Arechaga-Ocampo E., Rojas-Castaneda J.C., Jimenez-Trejo F., Chavez-Saldana M.

Embase

Frontiers in bioscience (Landmark edition). 22 (pp 1073-1098), 2017. Date of Publication: 01 Mar 2017.

[Review]

AN: 620094569

Testicular germ cell cancer (TGCT) is the most common malignancy among young adult males, which has become important due to its increased incidence and mortality in the population worldwide. The etiology is multifactorial. Recent studies have shown some associations between the development of isolated TGCT and certain risk factors, such as exposure to endocrine disruptors, cryptorchidism, and family history of cancer, in order to identify the key pieces in carcinogenesis. Some of the most important findings in recent years is the association of different genes, such as c-KIT/KITLG, expression of the miR-371-373 cluster and protein expression as c-KIT and POU5F1 in the development of this neoplasia, and the identification of new molecular markers as TGFBR3 gene, identifying aberrant methylation patterns in promoter regions of several genes, expression of miR-1297 which regulates PTEN and protein expression as DMTR1. In the future, a multidisciplinary research strategy could provide valuable new insights into the etiology of TGCTs, which support clinical diagnosis of TGCT in the next years to increase survival in this kind of patients.

PMC Identifier

28199193 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28199193>]

Institution

(Landero-Huerta, Viguera-Villasenor, Rojas-Castaneda, Jimenez-Trejo) Laboratory of Reproductive Biology, National Institute of Pediatrics, Mexico City, Mexico (Yokoyama-Rebollar)

Department of Human Genetics, National Institute of Pediatrics, Mexico City, Mexico

(Arechaga-Ocampo) Department of Natural Sciences and Engineering, Autonomous Metropolitan University Cuajimalpa Campus, Mexico City, Mexico

(Chavez-Saldana) Laboratory of Reproductive Biology, National Institute of Pediatrics, Mexico City, Mexico, mdoloreschs@gmail.com

Year of Publication

2017

426.

Can laparoscopy be part of a paediatric surgery outreach service?.

Peeraully R., Hill R., Colliver D., Williams A., Motiwale S., Davies B.

Embase

Annals of the Royal College of Surgeons of England. 99 (5) (pp 355-357), 2017. Date of Publication: 01 May 2017.

[Article]

AN: 617822773

INTRODUCTION The aim of this study was to assess the outreach laparoscopic service delivered by four paediatric surgeons to a district general hospital (DGH). **METHODS** A retrospective review was carried out of all laparoscopic procedures performed in a single DGH between January 2004 and November 2014 by the four paediatric surgeons providing the outreach service. All operations were identified from the electronic theatre system and archived correspondence. Demographic and clinical details were obtained from contemporaneous records. **RESULTS** Over the 11-year study period, 1,339 operations were performed as part of the outreach paediatric surgery service, with 128 patients (9.6%) undergoing laparoscopy. The indications for laparoscopic surgery were impalpable unilateral or bilateral undescended testes (UDT) (n=79, 62%) or request for insertion of a feeding gastrostomy (n=49, 38%). All but six UDT cases (96%) were performed as day surgery and the median length of stay for gastrostomy patients was 3 days (interquartile range: 2-3 days). There were three UDT cases with surgical complications and one had complications related to the anaesthesia. One gastrostomy case required transfer to our tertiary centre for management of postoperative urinary retention and urethral injury. **CONCLUSIONS** Elective laparoscopic procedures in young children can be provided safely as components of an outreach paediatric surgery service in a DGH setting as part of an increasing volume of operations performed by specialist paediatric surgeons. This enables children to have a high quality service as close to their home as possible.

PMC Identifier

28462639 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28462639>]

Institution

(Peeraully, Hill, Colliver, Williams, Motiwale, Davies) Nottingham University Hospitals NHS Trust, UK

Year of Publication

2017

427.

Prenatal Anogenital Distance Is Shorter in Fetuses With Hypospadias.

Gilboa Y., Perlman S., Kivilevitch Z., Messing B., Achiron R.

Embase

Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine. 36 (1) (pp 175-182), 2017. Date of Publication: 01 Jan 2017.

[Article]

AN: 617651739

OBJECTIVES: Recent research provides evidence that anogenital distance may serve as a novel metric to assess reproductive potential in men. In children, a shorter anogenital distance was linked with cryptorchidism, hypospadias, and micropenis. Scarce data exist in the literature regarding anogenital distance measurement in the fetus. The aim of our study was to assess whether intrauterine measurement of fetal anogenital distance could assist in the differential diagnosis of male genital anomalies. **METHODS:** Anogenital distance was prospectively measured in all cases referred for suspected isolated abnormal male genitalia. Final diagnoses, confirmed by a pediatric urologist, were compared with anogenital distance prenatal measurements.

RESULTS: Fifty-two cases were referred for evaluation because of suspected male external genital malformation during a 12-month period. Cases with normal-appearing genitalia, associated major malformations, and early severe fetal growth restriction were excluded from the study. Postnatal examination revealed 14 cases of hypospadias in varying severity and 8 cases of a buried penis. All fetuses with hypospadias had an anogenital distance measurement below the fifth percentile. Statistical analysis revealed a significant difference between the normal mean anogenital distance for gestational age versus those with hypospadias (mean \pm -SD, 16.90 \pm -4.08 and 11.68 \pm -3.31 mm, respectively; P =.001). No significant difference was found between the normal mean anogenital distance for gestational age versus those with a buried penis (18.85 \pm -2.76 and 19.46 \pm -3.41 mm; P =.700).

CONCLUSIONS: Fetuses with hypospadias have a statistically significant shorter anogenital distance compared with the general population. Therefore, anogenital distance may serve as a complementary objective sonographic parameter in the prenatal assessment and counseling of male external genital anomalies.

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PMC Identifier

27925677 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27925677>]

Institution

(Gilboa, Perlman, Kivilevitch, Messing, Achiron) Prenatal Diagnostic Unit, Department of Obstetrics and Gynecology, Haim Sheba Medical Center, Tel Hashomer, Israel, affiliated with the Sackler Faculty of Medicine, Tel Aviv University, Tel-Aviv, Israel

Year of Publication

2017

428.

Undescended testes: Diagnosis and timely treatment in Australia (1995-2014).

Vikraman J., Donath S., Hutson Ao J.M.

Embase

Australian family physician. 46 (3) (pp 152-158), 2017. Date of Publication: 01 Mar 2017.

[Article]

AN: 617205922

BACKGROUND: Routine primary care checks in infants and prepubertal boys aim for early detection and intervention of undescended testes (UDT). Congenital and acquired UDT cause infertility, and congenital UDT also increases testicular cancer risk. We examined 20 years of Australian orchidopexy data (1995-2014) to explore the national orchidopexy operation rates over time. METHODS: Orchidopexy and population data were collected from the Australian Bureau of Statistics (ABS) for 1995-2014, and census data for each age group were also collected. Poisson regressions were used to analyse the data.

RESULTS: For patients aged 5-14 years over the past 20 years, possibly indicating that acquired UDT is not being diagnosed and treated in some boys, risking infertility in adulthood.

PMC Identifier

28260279 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28260279>]

Year of Publication

2017

429.

Relationship between Undescended Testis Position and Prevalence of Testicular Appendices, Epididymal Anomalies, and Patency of Processus Vaginalis.

Favorito LA; Riberio Julio-Junior H; Sampaio FJ.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

BioMed Research International. 2017:5926370, 2017.

[Journal Article]

UI: 29445742

Objectives: To assess the incidence of testicular appendices (TAs), epididymal anomalies (EAs), and processus vaginalis (PV) patency in patients with undescended testis (UT) according to testicular position and to compare them with human fetuses.

Methods: We studied 85 patients (108 testes) with cryptorchidism and compared the features with those of 15 fetuses (30 testes) with scrotal testes. We analyzed the relationships among the testis and epididymis, patency of PV, and the presence of TAs. We used the Chi-square test for statistical analysis ($p < 0.05$).

Results: In 108 UT, 72 (66.66%) had PV patent, 67 (62.03%) had TAs, and 39 (36.12%) had EAs. Of the 108 UT, 14 were abdominal (12.96%; 14 had PV patency, 9 TAs, and 7 EAs); 81 were inguinal (75%; 52 had PV patency, 45 TAs, and 31 EAs), and 13 were suprascrotal (12.03%; 6 had PV patency, 13 TAs, and 1 EAs). The patency of PV was more frequently associated with EAs ($p = 0.00364$). The EAs had a higher prevalence in UT compared with fetuses ($p = 0.0005$).

Conclusions: Undescended testis has a higher risk of anatomical anomalies and the testes situated in abdomen and inguinal canal have a higher risk of presenting patency of PV and EAs.

Version ID

1

Status

MEDLINE

Author NameID

Favorito, Luciano A; ORCID: <https://orcid.org/0000-0003-1562-6068>

Authors Full Name

Favorito, Luciano A; Riberio Julio-Junior, Helce; Sampaio, Francisco J.

Institution

Favorito, Luciano A. Urogenital Research Unit, State University of Rio de Janeiro, Rio de Janeiro, RJ, Brazil. Riberio Julio-Junior, Helce. Urogenital Research Unit, State University of Rio de Janeiro, Rio de Janeiro, RJ, Brazil.

Sampaio, Francisco J. Urogenital Research Unit, State University of Rio de Janeiro, Rio de Janeiro, RJ, Brazil.

Comments

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Year of Publication

2017

430.

Paediatric Urology and the Dilemma of Low-quality Evidence for the Management of Common Urological Conditions (Vesicoureteral Reflux, Lower Urinary Tract Dysfunction, Undescended Testis) in Children.

Radmayr C; Tekgul S.

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European Urology Focus. 3(2-3):308-309, 2017 04.

[Journal Article]

UI: 28753880

We have to strive for a more prospective scientific approach to every specific urological condition in children, with contributions from all parties involved and an emphasis on proper outcome research.

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Version ID

1

Status

MEDLINE

Authors Full Name

Radmayr, Christian; Tekgul, Serdar.

Institution

Radmayr, Christian. Paediatric Urology, Medical University Innsbruck, Innsbruck, Austria.
Tekgul, Serdar. Division of Paediatric Urology, Department of Urology, Faculty of Medicine,
Hacettepe University, Ankara, Turkey. Electronic address: tekgul-k@tr.net.

Year of Publication

2017

431.

Undescended testes: Diagnostic Algorithm and Treatment.

Haid B; Rein P; Oswald J.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

European Urology Focus. 3(2-3):155-157, 2017 04.

[Journal Article]

UI: 28753808

Undescended testis persistently present at the 6th month of life in a term born boy should be treated, with the aim of bringing them to a scrotal position until the 12th month of life. Acquired undescended testes has to be looked for actively and treated at the earliest possible after diagnosis, that might be deferred due to a lack of attention and less access to a regular physical exam in older boys as opposed to infants.

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1

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MEDLINE

Authors Full Name

Haid, Bernhard; Rein, Patrick; Oswald, Josef.

Institution

Haid, Bernhard. Department for Pediatric Urology, Ordensklinikum Linz, Hospital of the Sisters of Charity, Austria. Electronic address: Bernhard.Haid@ordensklinikum.at. Rein, Patrick.

Department for Pediatric Urology, Ordensklinikum Linz, Hospital of the Sisters of Charity, Austria.

Oswald, Josef. Department for Pediatric Urology, Ordensklinikum Linz, Hospital of the Sisters of Charity, Austria.

Year of Publication

2017

432.

Efficacy of single-stage and two-stage Fowler-Stephens laparoscopic orchidopexy in the treatment of intraabdominal high testis.

Wang CY; Wang Y; Chen XH; Wei XY; Chen F; Zhong M.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Asian Journal of Surgery. 40(6):490-494, 2017 Nov.

[Comparative Study. Journal Article]

UI: 28410943

BACKGROUND/OBJECTIVE: To compare the curative effect of single-stage and two-stage Fowler-Stephens (F-S) laparoscopic orchidopexy for intraabdominal high testis and explore the appropriate surgical approach.

METHODS: We performed a prospective analysis of the clinical data of 28 patients who underwent F-S laparoscopic orchidopexy for intraabdominal high testis in our department from May 2012 to April 2015, including 15 cases of the single-stage F-S operation and 13 cases of the two-stage F-S operation. By comparing the two groups preoperative and postoperative (6 months) clinical data of testicular position, testicular volume, and sex hormone levels [testosterone (T), follicle stimulating hormone (FSH), and estradiol (E2)], we analyzed the difference in efficacy between the two procedures.

RESULTS: Twenty-eight patients completed laparoscopic surgery, no case was converted, and no testis was excised. All patients were followed up for 9-25 months after the operation, with an average follow-up of 16.2 months. The postoperative testicular volume of the single-stage and two-stage F-S groups was not significantly reduced ($p>0.05$). In both groups, the postoperative T levels were significantly increased compared to the preoperative levels ($p<0.05$), while the FSH and E2 levels were significantly decreased ($p<0.05$). The differences in testicular volume and T, FSH, and E2 levels between the two surgical procedures were not significant ($p>0.05$). In the single-stage F-S group, the testes were located in the scrotum in 13 cases and retracted to the

lower groin in two cases. In the two-stage F-S group, the testes were located in the scrotum in 12 cases and retracted to the lower groin in one case. The difference in postoperative testicular position between the two groups was not significant ($p>0.05$).

CONCLUSION: In the case of testis with good collateral circulation, single-stage F-S laparoscopic orchidopexy had the same safety and efficacy as the two-stage F-S procedure. Surgical options should be based on comprehensive consideration of intraoperative testicular location, testicular ischemia test, and collateral circumstances surrounding the testes. Under the appropriate conditions, we propose single-stage F-S laparoscopic orchidopexy be preferred. It may be appropriate to avoid unnecessary application of the two-stage procedure that has a higher cost and causes more pain for patients.

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Version ID

1

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Authors Full Name

Wang, Chang-Yuan; Wang, Yang; Chen, Xiao-Hua; Wei, Xiao-Yu; Chen, Feng; Zhong, Min.

Institution

Wang, Chang-Yuan. Department of Pediatric Surgery, Fujian Medical University Union Hospital, Fuzhou City, China. Wang, Yang. Department of Pediatric Surgery, Fujian Medical University Union Hospital, Fuzhou City, China. Electronic address: 18650331562@163.com.

Chen, Xiao-Hua. Department of Pediatric Surgery, Fujian Medical University Union Hospital, Fuzhou City, China.

Wei, Xiao-Yu. Department of Pediatric Surgery, Fujian Medical University Union Hospital, Fuzhou City, China.

Chen, Feng. Department of Pediatric Surgery, Fujian Medical University Union Hospital, Fuzhou City, China.

Zhong, Min. Department of Pediatric Surgery, Fujian Medical University Union Hospital, Fuzhou City, China.

Year of Publication

2017

Feasibility and Efficacy of a Urologic Profession Campaign on Cryptorchidism Using Internet and Social Media.

Borgmann H; Kliesch S; Roth S; Roth M; Degener S.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
Urologia Internationalis. 98(4):478-482, 2017.

[Journal Article]

UI: 27806367

INTRODUCTION: We performed a professional campaign in Germany intending to establish the urologic profession as a competent and helpful point of contact for patients with cryptorchidism. The aim of this study was to assess the feasibility of this campaign and to quantify the efficacy of using Internet vs. social media.

MATERIALS AND METHODS: The strategic design of the campaign comprised a strategy meeting, creation of a landing page, and targeted advertisements on Google in the form of Adwords and on Facebook in the form of sidebar ads and sponsored posts. Outcome measurements were number of impressions, homepage sessions, and downloads of an information brochure.

RESULTS: The campaign generated 2,511,923 impressions, 7,369 homepage sessions and 1,086 downloads of information brochures using a total investment budget of 7,500. Use of Google Adwords was more efficient on outcome measurements than Facebook. A subanalysis of Facebook advertisements showed that sidebar ads and sponsored posts were equally efficient.

CONCLUSIONS: New media are an effective platform for a profession campaign. Google Adwords is a more effective and cost-efficient platform than Facebook for a targeted campaign.

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Version ID

1

Status

MEDLINE

Authors Full Name

Borgmann, Hendrik; Kliesch, Sabine; Roth, Stephan; Roth, Mael; Degener, Stephan.

Institution

Borgmann, Hendrik. Department of Urology, University Hospital Mainz, Mainz, Germany.

Year of Publication

2017

434.

Is inguinal orchidopexy still a current procedure in the treatment of intraabdominal testis in the era of laparoscopic surgery?.

Arena S; Impellizzeri P; Perrone P; Scalfari G; Centorrino A; Turiaco N; Parisi S; Antonuccio P; Romeo C.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Pediatric Surgery. 52(4):650-652, 2017 Apr.

[Journal Article]

UI: 27629799

PURPOSE: To report our experience in surgical management of nonpalpable intraabdominal testis (NPIT) by inguinal orchidopexy without division of the spermatic vessels.

METHODS: We reviewed the records of NPIT patients who underwent orchidopexy between 2012 and 2015. All patients were evaluated ultrasonographically. When the testis was not detected ultrasonographically, a laparoscopic exploration was performed. If the testis was found on laparoscopy, surgery was resumed through an inguinal incision. A follow-up was performed at 1week, 1, 3 and 6months.

RESULTS: Twenty-one NPIT patients were treated, mean age 21.0+/-11.7months. Ultrasound identified 15 cases of NPIT (71%); diagnostic laparoscopy was performed in 6 (29%). All patients underwent an inguinal orchidopexy. At 1week, four testes were in a high scrotal position. At 6months follow-up, one testis was in a high scrotal position and one retracted up to the external inguinal ring. No atrophy was recorded.

CONCLUSIONS: Despite several attempts to find a surgical technique without any significant complications, all described procedures failed to meet the target. In our experience, inguinal orchidopexy is a safe, reliable and successful surgical procedure for the management of NPIT. It should be preferred to a technique requiring vascular division, burdened with a higher incidence of atrophy.

TYPE OF STUDY: Treatment study.

LEVELS OF EVIDENCE: Case series with no comparison group.

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Version ID

1

Status

MEDLINE

Authors Full Name

Arena, Salvatore; Impellizzeri, Pietro; Perrone, Patrizia; Scalfari, Gianfranco; Centorrino, Antonio; Turiaco, Nunzio; Parisi, Saveria; Antonuccio, Pietro; Romeo, Carmelo.

Institution

Arena, Salvatore. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Electronic address: salarena@unime.it. Impellizzeri, Pietro. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Perrone, Patrizia. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Scalfari, Gianfranco. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Centorrino, Antonio. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Turiaco, Nunzio. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Parisi, Saveria. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Antonuccio, Pietro. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy. Romeo, Carmelo. Department of human pathology in adult and developmental age "Gaetano Barresi", Unit of Pediatric Surgery, University of Messina, Italy.

Year of Publication

2017

435.

PITUITARY STALK INTERRUPTION SYNDROME: REPORT OF TWO CASES AND LITERATURE REVIEW.

Lichiardopol C; Albulescu DM.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Acta Endocrinologica. 13(1):96-105, 2017 Jan-Mar.

[Journal Article]

UI: 31149155

Pituitary stalk interruption syndrome (PSIS) consisting of the triad: ectopic posterior pituitary (EPP), thin or absent pituitary stalk and anterior pituitary hypoplasia is a rare pituitary malformation with variable degrees of pituitary insufficiency, from isolated growth hormone deficiency to TSH, gonadotropin and ACTH deficiency which may occur in time, with normo, hyper or hypoprolactinemia and central diabetes insipidus in up to 10% of cases. Also, extrapituitary malformations have been described in some cases. Genetic defects were identified only in 5% of cases. MRI findings are considered predictive for the endocrine phenotype. We aim to describe two cases with PSIS without central diabetes insipidus, anosmia and extrapituitary malformations, except for minor head dysmorphic features. The first case was referred at the age of 4 years for short stature (-4SDS for height, bone age 2 years), diagnosed with severe GH deficiency and developed central hypothyroidism and hypoprolactinemia during five-years follow-up. The second case, a 26 year old male with birth asphyxia, cryptorchidism, poor growth in childhood and adolescence (-3 to -4 height SDS), absent puberty and normal adult height (-1.18 SDS; bone age 15.5 years and active growth plates) had GH, TSH, ACTH deficiency and low normal PRL levels. Increasing medical awareness on PSIS clinical and endocrine heterogeneity may help a more early and accurate diagnosis. Corroboration of neuroimaging and endocrine data will improve our knowledge and understanding and will create premises for molecular diagnosis, genetic counseling and a better patients' management.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Lichiardopol, C; Albulescu, D M.

Institution

Lichiardopol, C. University of Medicine and Pharmacy Craiova, Dept. of Endocrinology, Craiova, Romania. Lichiardopol, C. University of Medicine and Pharmacy Craiova, Dept. of Medical Imagery, Craiova, Romania.

Albulescu, D M. Emergency Clinical Hospital, Dept. of Endocrinology, Craiova, Romania.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6525749>

Year of Publication

2017

436.

Prune belly syndrome: A report of 15 cases from Sudan.

Kheir AEM; Ali EMA; Medani SA; Maaty HS.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Sudanese Journal of Paediatrics. 17(1):42-48, 2017.

[Journal Article]

UI: 29213170

Prune belly syndrome is a rare congenital malformation of unknown aetiology, composed of a triad of deficient abdominal wall muscle, cryptorchidism and urinary tract anomalies. The majority of patients have associated pulmonary, skeletal, cardiac, and gastrointestinal defects. This was a prospective, case finding study that was conducted in the main paediatric hospitals in Khartoum state, during the period December 2015 to September 2016. A total of 15 patients with prune belly syndrome were collected. Patients' characteristics were noted including socio-demographic data, laboratory and radiological investigations and any medical or surgical intervention. There were 12 males and 3 females with a male to female ratio of 4:1. Most of the patients (80%) had hydronephrosis and hydroureter. The study revealed that 60% of the patients had associated anomalies, there were 4 (26.6%) with cardiac defects, 3 (20%) with orthopaedic defects one patient with small bowel volvulus and one patient with cleft lip. 6 (40%) patients received medical intervention and 8 (53%) patients underwent surgical procedures. At the last follow up visit, 2 (13.4%) patients had normal renal function tests, 8 (53.3%) ended with chronic kidney disease, and 5 died with a mortality rate of 33.3%. Prune belly syndrome is a rare entity with wide variability in severity and clinical manifestations. The mortality in prune belly syndrome remains high despite medical and surgical interventions.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Kheir, Abdelmoneim E M; Ali, Eltigani M A; Medani, Safaa A; Maaty, Huda S.

Institution

Kheir, Abdelmoneim E M. Department of Paediatrics and Child Health, Faculty of Medicine, University of Khartoum and Soba University Hospital, Sudan. Kheir, Abdelmoneim E M. Corresponding author.

Ali, Eltigani M A. Department of Paediatrics and Child Health, Faculty of Medicine, University of Khartoum and Soba University Hospital, Sudan.

Medani, Safaa A. Department of Pediatrics and Child Health, Faculty of Medicine, Al Neelain University, Khartoum, Sudan.

Maaty, Huda S. Department of Paediatrics, Soba university hospital, Khartoum, Sudan.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5621858>

Year of Publication

2017

437.

On the descent of the epididymo-testicular unit, cryptorchidism, and prevention of infertility.

[Review]

Hadziselimovic F.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Basic & Clinical Andrology. 27:21, 2017.

[Journal Article. Review]

UI: 29163975

This comprehensive review provides in-depth coverage of progress made in understanding the molecular mechanisms underlying cryptorchidism, a frequent pathology first described in about 1786 by John Hunter. The first part focuses on the physiology, embryology, and histology of epididymo-testicular descent. In the last 20 years epididymo-testicular descent has become the victim of schematic drawings with an unjustified rejection of valid histological data. This part also includes discussion on the roles of gonadotropin-releasing hormone, fibroblast growth factors, Mullerian inhibiting substance, androgens, inhibin B, and insulin-like 3 in epididymo-testicular descent. The second part addresses the etiology and histology of cryptorchidism as well as the importance of mini-puberty for normal fertility development. A critical view is presented on current clinical guidelines that recommend early orchidopexy alone as the best possible treatment.

Finally, by combining classical physiological information and the output of cutting-edge genomics data into a complete picture the importance of hormonal treatment in preventing cryptorchidism-induced infertility is underscored.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Hadziselimovic, Faruk.

Institution

Hadziselimovic, Faruk. Cryptorchidism Research Institute, Kindermedizinisches Zentrum Liestal, Liestal, Switzerland. Hadziselimovic, Faruk. Pediatrics at the University of Basel and Director of Cryptorchidism Research Institute, Kindermedizinisches Zentrum, Bahnhofplatz 11, 4410 Liestal, Switzerland.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5686796>

Other Abstract

Publisher: Cette revue complete traite en profondeur les progres realises dans la comprehension des mecanismes moleculaires a la base de la cryptorchidie, une pathologie frequente decrite pour la premiere fois aux environs de 1786 par John Hunter. La premiere partie est. centree sur la physiologie, l'embryologie et l'histologie de la descente epididymo-testiculaire. Durant les 20 dernieres annees, la descente epididymo-testiculaire est. devenue la victime de dessins schematiques associes a un rejet injustifie de donnees histologiques valides. Cette partie discute aussi les roles qu'ont dans la descente epididymo-testiculaire l'hormone liberant les gonadotrophines, les facteurs de croissance fibroblastiques, l'hormone antimullerienne, les androgenes, l'inhibine B, et l'insuline-like 3. La seconde partie aborde l'etiologie et l'histologie, ainsi que l'importance de la minipuberte pour un developpement normal de la fertilité. Un regard critique est. porte sur les recommandations cliniques actuelles qui conseillent la seule orchidopexie precoce comme le meilleur traitement possible. Finalement, en combinant les informations issues de la physiologie classique et la production des donnees genomiques les plus en pointe dans un tableau complet, l'importance du traitement hormonal dans la prevention de l'infertilite induite par la cryptorchidie est soulignee.; Language: French

Year of Publication

2017

438.

Early Experience with Laparoscopic Management of Nonpalpable Undescended Testes.

Ekwunife OH; Modekwe VI; Ugwu JO; Ugwunne CA.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Nigerian Journal of Surgery. 23(2):115-118, 2017 Jul-Dec.

[Journal Article]

UI: 29089736

BACKGROUND: Nonpalpable undescended testes (NPT) constitute 20%-30% of undescended testes, and its management has been a challenge both in diagnosis and treatment. Worldwide, laparoscopy is the current gold standard of management. In Nigeria, the management of NPT has largely been by open surgery with consequent high morbidity. In Nigeria, the trend is changing from a largely open management with its attendant high morbidity, to laparoscopic management which is the current worldwide gold standard of care.

AIM: This study aims to classify the laparoscopic features of NPT and determine the outcome of managed cases in our center.

METHODOLOGY: Prospective data were collected from consecutive patients who had laparoscopy for NPT at the Paediatric Surgical Unit of Nnamdi Azikiwe University Teaching Hospital, Nnewi, Nigeria from June 2014 to July 2016.

RESULTS: A total of 15 patients with 23 testes were treated. There were eight patients with bilateral NPT; four had left and the remaining three right NPT. The age ranged from 1.2 to 29 years with a median of 5 years. Eleven out of the 22 internal inguinal rings were open. The position of the testes was canalicular (2), peeping (2), low abdominal (6), high abdominal (6), blind-ended vas (1), absent vas and vessels (5). No further intervention was needed for the six agenetic/atrophic testes. Standard open orchiopexy was done for the two canalicular testes. Eight testes were brought down by one stage laparoscopic orchiopexy while four were brought down by staged laparoscopic Fowler-Stephens procedure. Laparoscopic orchiectomy was done in two patients (a grossly dysmorphic testes [nubbin] and a high abdominal testis in a 29-year-old). Orchiopexy was successful in 11 out of 15 fixed testes. Of the unsuccessful ones, three testes were atrophic (volume less than what it was initially) while two were high scrotal (one testes has both complications). There was no conversion to open abdominal surgery. All patients were discharged within 24 h of surgery.

CONCLUSION: Laparoscopy provides for a better management of NPT by combining diagnosis and intervention in the same sitting with a good success rate and minimal postoperative morbidity.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Ekwunife, Okechukwu Hyginus; Modekwe, Victor Ifeanyichukwu; Ugwu, Jideofor Okechukwu; Ugwunne, Chuka Abunike.

Institution

Ekwunife, Okechukwu Hyginus. Department of Surgery, Nnamdi Azikiwe University Teaching Hospital, PMB 5025, Nnewi, Anambra State, Nigeria. Modekwe, Victor Ifeanyichukwu.

Department of Surgery, Nnamdi Azikiwe University Teaching Hospital, PMB 5025, Nnewi, Anambra State, Nigeria.

Ugwu, Jideofor Okechukwu. Department of Surgery, Nnamdi Azikiwe University Teaching Hospital, PMB 5025, Nnewi, Anambra State, Nigeria.

Ugwunne, Chuka Abunike. Department of Surgery, Nnamdi Azikiwe University Teaching Hospital, PMB 5025, Nnewi, Anambra State, Nigeria.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5649426>

Year of Publication

2017

439.

A case of an infant with congenital combined pituitary hormone deficiency and normalized liver histology of infantile cholestasis after hormone replacement therapy.

Wada K; Kobayashi H; Moriyama A; Haneda Y; Mushimoto Y; Hasegawa Y; Onigata K; Kumori K; Ishikawa N; Maruyama R; Sogo T; Murphy L; Taketani T.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Clinical Pediatric Endocrinology. 26(4):251-257, 2017.

[Journal Article]

UI: 29026274

Congenital combined pituitary hormone deficiency (CPHD) may present with cholestasis in the neonate or during early infancy. However, its precise mechanism is unknown. A 3-mo-old boy presented with cryptorchidism and hypoplastic scrotum after birth. Neonatal jaundice was noted but temporarily improved with phototherapy. Jaundice recurred at 2 mo of age. Elevated direct bilirubin (D-Bil) and liver dysfunction were found but cholangiography showed no signs of biliary atresia (BA). Liver biopsy findings showed giant cell formation of hepatocytes with hypoplastic bile ducts. Subsequent magnetic resonance imaging (MRI) of the head revealed a hypoplastic pituitary gland with an ectopic posterior lobe, and the patient was diagnosed with congenital CPHD based on decreased secretion of cortisol and GH by the pituitary anterior lobe load test. D-Bil levels promptly improved after hydrocortisone (HDC) replacement. We subsequently began replacement with levothyroxine (L-T4) and GH, and liver histology showed normal interlobular bile ducts at 8 mo old. This is the first case report of proven histological improvement after hormone replacement therapy. This suggested that pituitary-mediated hormones, especially cortisol, might be involved in the development of the bile ducts.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Wada, Keisuke; Kobayashi, Hironori; Moriyama, Aisa; Haneda, Yasuhiro; Mushimoto, Yuichi; Hasegawa, Yuki; Onigata, Kazumichi; Kumori, Koji; Ishikawa, Noriyoshi; Maruyama, Riruke; Sogo, Tsuyoshi; Murphy, Lynne; Taketani, Takeshi.

Institution

Wada, Keisuke. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan. Kobayashi, Hironori. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

Moriyama, Aisa. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

Haneda, Yasuhiro. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

Mushimoto, Yuichi. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

Hasegawa, Yuki. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

Onigata, Kazumichi. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

Kumori, Koji. Department of Digestive and General Surgery, School of Medicine, Shimane University, Shimane, Japan.

Ishikawa, Noriyoshi. Department of Pathology, Shimane University Hospital, Shimane, Japan.

Maruyama, Riruke. Department of Pathology, Shimane University Hospital, Shimane, Japan.

Sogo, Tsuyoshi. Department of Pediatric Hepatology and Gastroenterology, Saiseikai Yokohama City Tobu Hospital, Kanagawa, Japan.

Murphy, Lynne. Department of Medical English Education, School of Medicine, Shimane University, Shimane, Japan.

Taketani, Takeshi. Department of Pediatrics, School of Medicine, Shimane University, Shimane, Japan.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5627226>

Year of Publication

2017

440.

An atypical case of Noonan syndrome with KRAS mutation diagnosed by targeted exome sequencing.

Kim J; Cho SY; Yang A; Jang JH; Choi Y; Lee JE; Jin DK.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Annals of Pediatric Endocrinology & Metabolism. 22(3):203-207, 2017 Sep.

[Journal Article]

UI: 29025208

Noonan syndrome (NS) is a genetic disorder caused by autosomal dominant inheritance and is characterized by a distinctive facial appearance, short stature, chest deformity, and congenital heart disease. In individuals with NS, germline mutations have been identified in several genes involved in the RAS/mitogen-activated protein kinase signal transduction pathway. Because of its clinical and genetic heterogeneity, the conventional diagnostic protocol with Sanger sequencing requires a multistep approach. Therefore, molecular genetic diagnosis using targeted exome

sequencing (TES) is considered a less expensive and faster method, particularly for patients who do not fulfill the clinical diagnostic criteria of NS. In this case, the patient showed short stature, dysmorphic facial features suggestive of NS, feeding intolerance, cryptorchidism, and intellectual disability in early childhood. At the age of 16, the patient still showed extreme short stature with delayed puberty and characteristic facial features suggestive of NS. Although the patient had no cardiac problems or chest wall deformities, which are commonly present in NS and are major concerns for patients and clinicians, the patient showed several other characteristic clinical features of NS. Considering the possibility of a genetic disorder, including NS, a molecular genetic study with TES was performed. With TES analysis, we detected a pathogenic variant of c.458A > T in KRAS in this patient with atypical NS phenotype and provided appropriate clinical management and genetic counseling. The application of TES enables accurate molecular diagnosis of patients with nonspecific or atypical features in genetic diseases with several responsible genes, such as NS.

Version ID

1

Status

PubMed-not-MEDLINE

Author NameID

Cho, Sung Yoon; ORCID: <https://orcid.org/0000-0003-2913-059X>

Authors Full Name

Kim, Jinsup; Cho, Sung Yoon; Yang, Aram; Jang, Ja-Hyun; Choi, Youngbin; Lee, Ji-Eun; Jin, Dong-Kyu.

Institution

Kim, Jinsup. Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea. Cho, Sung Yoon. Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea.

Yang, Aram. Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea.

Jang, Ja-Hyun. Green Cross Laboratories, Green Cross Genome, Yongin, Korea.

Choi, Youngbin. Department of Pediatrics, Inha University Hospital, Inha University Graduate School of Medicine, Incheon, Korea.

Lee, Ji-Eun. Department of Pediatrics, Inha University Hospital, Inha University Graduate School of Medicine, Incheon, Korea.

Jin, Dong-Kyu. Department of Pediatrics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5642084>

Year of Publication

2017

441.

Focal Dermal Hypoplasia (Goltz Syndrome): A Cross-sectional Study from Eastern India.

Ghosh SK; Dutta A; Sarkar S; Nag SS; Biswas SK; Mandal P.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Indian Journal of Dermatology. 62(5):498-504, 2017 Sep-Oct.

[Journal Article]

UI: 28979012

INTRODUCTION: Focal dermal hypoplasia (Goltz syndrome), is an extremely rare genetic disorder characterized by distinct skin manifestations and a wide range of abnormalities involving the ocular, dental, skeletal, urinary, gastrointestinal, cardiovascular, and central nervous systems. The objective of the present series is to emphasize the different typical as well as unusual features of this rare syndrome.

METHODOLOGY: This cross-sectional observational study was performed over a period of 8 years in a tertiary care hospital of Eastern India. Consecutive patients with the clinical diagnosis of Goltz syndrome were studied.

RESULTS: A total of 8 patients with Goltz syndrome were evaluated. Out of them, one patient was a boy and the rest were girl. The age ranged from 3 days to 9 years. There was no family history. A characteristic Blaschkoid hypo- and hyper-pigmented skin lesions, congenital nodular fat herniation, and skin atrophy were present in all patients. Congenital cutaneous aplasia was present in 50% of the patients. Facial asymmetry and ear deformity (megalopinna and low-set ears) were seen in 37.5% and 12.5% of patients, respectively. Cutaneous telangiectasia was noticed in 37.5% of patients. Freckle- and lentigines-like pigmentation within the hypopigmented macules was found in 25% of patients. Raspberry-like papillomas around mouth were documented in 6 (75%) patients. Dysplastic nail changes with ridging were seen in 7 (87.5%) patients. Genital abnormality in the form of bilateral undescended testes and microphthalmia with aniridia were found in one patient each. Limb defects were present in all patients. Left-sided renal agenesis was found in one patient. The patient also had multiple cortical cysts of the right kidney.

LIMITATIONS: Genetic testing could not be performed in the present series.

CONCLUSIONS: Our case series showed a few unusual or extremely rare manifestations such as undescended testes, dermal sinus, kyphoscoliosis, aniridia, unilateral kidney agenesis, and renal cortical cysts among others.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Ghosh, Sudip Kumar; Dutta, Abhijit; Sarkar, Sharmila; Nag, Shanka Subhra; Biswas, Surajit Kumar; Mandal, Prabhakar.

Institution

Ghosh, Sudip Kumar. Department of Dermatology, Venereology, and Leprosy R.G.Kar Medical College, Kolkata, West Bengal, India. Dutta, Abhijit. Department of Pediatrics, North Bengal Medical College, West Bengal, India.

Sarkar, Sharmila. Department of Psychiatry, Calcutta National Medical College, Kolkata, West Bengal, India.

Nag, Shanka Subhra. Department of Pediatrics, North Bengal Medical College, West Bengal, India.

Biswas, Surajit Kumar. Department of Dermatology, Venereology, and Leprosy R.G.Kar Medical College, Kolkata, West Bengal, India.

Mandal, Prabhakar. Department of Dermatology, Venereology, and Leprosy R.G.Kar Medical College, Kolkata, West Bengal, India.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5618837>

Year of Publication

2017

442.

Laparoscopy in the Evaluation of Impalpable Testes and Its Short-term Outcomes: A 7 Years' Experience.

Kumar R; Mandal KC; Halder P; Hadiuzzaman M; Mukhopadhyay M; Mukhopadhyay B.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Indian Association of Pediatric Surgeons. 22(4):232-236, 2017 Oct-Dec.

[Journal Article]

UI: 28974876

AIMS: The aim of this study is to report and analyze results of laparoscopy in impalpable testes performed between 2009 and 2016 and its short-term outcomes.

MATERIALS AND METHODS: Demographic data, laterality, laparoscopic findings, operative time, procedure, hospital stay, complications, and follow-up data of 76 patients with 79 impalpable testes from 2009 to 2016 were retrospectively collected and analyzed. Successful outcome was defined as maintenance of intrascrotal position with no atrophy at a follow-up of at least 6 months.

RESULTS: Impalpable testes constituted 24% of undescended testes in our series. Mean age was 3.9 years. Forty-two patients had left-sided, 31 right-sided, and three bilateral impalpable testes. Of the 79 clinically impalpable testes, on laparoscopy, 3 were vanishing testes, 52 were intra-abdominal (6 high-lying and 46 low-lying), 18 canalicular and 6 nubbin testes. Ultimately, 52 underwent laparoscopic orchiopexy: 46 single-staged orchiopexy and 6 two-staged Fowler-Stephens procedure. Mean operating time was 77 min. Complications were few and mostly minor. Eleven patients were lost in follow-up. On a mean follow-up of 23 months, one testis that underwent single-staged laparoscopic orchiopexy atrophied whereas good size and intrascrotal position were maintained in the rest.

CONCLUSIONS: Laparoscopy in impalpable testes was safe, feasible, and effective. Overall outcome was good which was obtained by minimal use of electrocautery, dissection with wide strip of peritoneum and extensive retroperitoneal dissection for mobilization. There is a need for wide reporting of cases from high-volume pediatric surgery centers in India.

Version ID

1

Status

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Authors Full Name

Kumar, Rajarshi; Mandal, Kartik Chandra; Halder, Pankaj; Hadiuzzaman, Md; Mukhopadhyay, Madhumita; Mukhopadhyay, Biswanath.

Institution

Kumar, Rajarshi. Department of General Surgery, Calcutta National Medical College and Hospital, Kolkata, West Bengal, India. Mandal, Kartik Chandra. Department of Paediatric Surgery, Dr. B. C. Roy Post Graduate Institute of Paediatric Sciences, Kolkata, West Bengal, India.

Halder, Pankaj. Department of Paediatric Surgery, Dr. B. C. Roy Post Graduate Institute of Paediatric Sciences, Kolkata, West Bengal, India.

Hadiuzzaman, Md. Department of Paediatric Surgery, Nil Ratan Sircar Medical College and Hospital, Kolkata, West Bengal, India.

Mukhopadhyay, Madhumita. Department of Pathology, Institute of Post Graduate Medical Education and Research, Kolkata, West Bengal, India.

Mukhopadhyay, Biswanath. Apollo Gleneagles Hospital, Kolkata, West Bengal, India.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5615898>

Year of Publication

2017

443.

Chromosome 15 structural abnormalities: effect on IGF1R gene expression and function.
Cannarella R; Mattina T; Condorelli RA; Mongioi LM; Pandini G; La Vignera S; Calogero AE.
OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
Endocrine Connections. 6(7):528-539, 2017 Oct.

[Journal Article]

UI: 28899882

Insulin-like growth factor 1 receptor (IGF1R), mapping on the 15q26.3 chromosome, is required for normal embryonic and postnatal growth. The aim of the present study was to evaluate the IGF1R gene expression and function in three unrelated patients with chromosome 15 structural abnormalities. We report two male patients with the smallest 15q26.3 chromosome duplication described so far, and a female patient with ring chromosome 15 syndrome. Patient one, with a 568 kb pure duplication, had overgrowth, developmental delay, mental and psychomotor retardation, obesity, cryptorchidism, borderline low testis volume, severe oligoasthenoteratozoospermia and gynecomastia. We found a 1.8-fold increase in the IGF1R mRNA and a 1.3-fold increase in the IGF1R protein expression ($P < 0.05$). Patient two, with a 650 kb impure duplication, showed overgrowth, developmental delay, mild mental retardation, precocious puberty, low testicular volume and severe oligoasthenoteratozoospermia. The IGF1R mRNA and protein expression was similar to that of the control. Patient three, with a 46,XX r(15)

(p10q26.2) karyotype, displayed intrauterine growth retardation, developmental delay, mental and psychomotor retardation. We found a <0.5-fold decrease in the IGF1R mRNA expression and an undetectable IGF1R activity. After reviewing the previously 96 published cases of chromosome 15q duplication, we found that neurological disorders, congenital cardiac defects, typical facial traits and gonadal abnormalities are the prominent features in patients with chromosome 15q duplication. Interestingly, patients with 15q deletion syndrome display similar features. We speculate that both the increased and decreased IGF1R gene expression may play a role in the etiology of neurological and gonadal disorders.

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Version ID

1

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PubMed-not-MEDLINE

Authors Full Name

Cannarella, Rossella; Mattina, Teresa; Condorelli, Rosita A; Mongioi, Laura M; Pandini, Giuseppe; La Vignera, Sandro; Calogero, Aldo E.

Institution

Cannarella, Rossella. Department of Clinical and Experimental MedicineUniversity of Catania, Catania, Italy. Mattina, Teresa. GeneticsUniversity of Catania, Catania, Italy.

Condorelli, Rosita A. Department of Clinical and Experimental MedicineUniversity of Catania, Catania, Italy.

Mongioi, Laura M. Department of Clinical and Experimental MedicineUniversity of Catania, Catania, Italy.

Pandini, Giuseppe. Department of Clinical and Experimental MedicineUniversity of Catania, Catania, Italy.

La Vignera, Sandro. Department of Clinical and Experimental MedicineUniversity of Catania, Catania, Italy.

Calogero, Aldo E. Department of Clinical and Experimental MedicineUniversity of Catania, Catania, Italy acaloger@unict.it.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5597972>

Year of Publication

2017

444.

Omphalocele and Gastroschisis in Newborns: Over 16 Years of Experience from a Single Clinic.

Watanabe S; Suzuki T; Hara F; Yasui T; Uga N; Naoe A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Neonatal Surgery. 6(2):27, 2017 Apr-Jun.

[Journal Article]

UI: 28770124

Infants born with potentially life-threatening conditions of omphalocele and gastroschisis may require long-term hospitalization. We aimed to compare the outcomes of these two conditions occurring over a 16-year period (2001-16). It is a retrospective study of 19 newborns undergoing surgery for these two abdominal wall defects (8 patients with omphalocele and 11 cases of gastroschisis). The average birth weights for the newborns with omphaloceles and gastroschisis were 2554.5 g and 2248.6 g respectively. Associated anomalies included trisomy 18, Beckwith-Wiedemann syndrome, congenital heart disease, Meckel's diverticulum, inguinal hernias, renal deformities, limb deformities, cryptorchidism, body stalk anomalies, and closed gastroschisis. The average hospital stay for the newborns with omphaloceles and gastroschisis were 42.6 days 50.2 days respectively. The time to the start of postoperative nutritional supplementation for the newborns with omphaloceles and gastroschisis were 4.3 days for the infants with omphaloceles and 7.3 days for respectively. The survival rates for the newborns with omphaloceles and gastroschisis were similar, 87.5% and 81.8% respectively. Survival rates in omphalocele correlated negatively with associated anomalies. In gastroschisis cases, strict care is necessary when intestinal dilation is observed via fetal sonography.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Watanabe, Shunosuke; Suzuki, Tatuya; Hara, Fujio; Yasui, Toshihiro; Uga, Naoko; Naoe, Atuki.

Institution

Watanabe, Shunosuke. Department of Pediatric Surgery, Fujita Health University, Japan.

Suzuki, Tatuya. Department of Pediatric Surgery, Fujita Health University, Japan.

Hara, Fujio. Department of Pediatric Surgery, Fujita Health University, Japan.

Yasui, Toshihiro. Department of Pediatric Surgery, Fujita Health University, Japan.

Uga, Naoko. Department of Pediatric Surgery, Fujita Health University, Japan.

Naoe, Atuki. Department of Pediatric Surgery, Fujita Health University, Japan.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5538593>

Year of Publication

2017

445.

Congenital urogenital abnormalities in children with congenital hypothyroidism.

Yousefi Chaijan P; Dorreh F; Sharafkhah M; Amiri M; Ebrahimimonfared M; Rafeie M; Safi F.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Medical Journal of the Islamic Republic of Iran. 31:7, 2017.

[Journal Article]

UI: 28638814

Background: Congenital hypothyroidism (CH), as one of the most common congenital endocrine disorders, may be significantly associated with congenital malformations. This study investigates urogenital abnormalities in children with primary CH (PCH). Methods: This case-control study was conducted on 200 children aged three months to 1 year, referred to Amir-Kabir Hospital, Arak, Iran. One hundred children with PCH, as the case group, and 100 healthy children, as the control group, were selected using convenient sampling. For all children, demographic data checklists were filled, and physical examination, abdomen and pelvic ultrasound and other diagnostic measures (if necessary) were performed to evaluate the congenital urogenital abnormalities including anomalies of the penis and urethra, and disorders and anomalies of the scrotal contents.

Results: Among 92 (100%) urogenital anomalies diagnosed, highest frequencies with 37 (40.2%), 26(28.2%) and 9 (9.7%) cases including hypospadias, Cryptorchidism, and hydrocele, respectively. The frequency of urogenital abnormalities among 32 children with PCH, with 52 cases (56.5%) was significantly higher than the frequency of abnormalities among the 21 children in the control group, with 40 cases (43.4%). (OR=2.04; 95%CI: 1.1-3.6; p=0.014). Conclusion: Our study demonstrated that PCH is significantly associated with the congenital urogenital abnormalities. However, due to the lack of evidence in this area, further studies are

recommended to determine the necessity of conducting screening programs for abnormalities of the urogenital system in children with CH at birth.

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1

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PubMed-not-MEDLINE

Authors Full Name

Yousefi Chaijan, Parsa; Dorreh, Fatemeh; Sharafkhah, Mojtaba; Amiri, Mohammad; Ebrahimimonfared, Mohsen; Rafeie, Mohammad; Safi, Fatemeh.

Institution

Yousefi Chaijan, Parsa. Department of Pediatrics Nephrology, AmirKabir Hospital, School of Medicine, Arak University of Medical Sciences, Arak, Iran. Dorreh, Fatemeh. Department of Pediatrics Nephrology, AmirKabir Hospital, School of Medicine, Arak University of Medical Sciences, Arak, Iran.

Sharafkhah, Mojtaba. Students Research Committee, School of Medicine, Arak University of Medical Sciences, Arak, Iran.

Amiri, Mohammad. Department of Emergency Medicine, Valiasr Hospital, School of Medicine, Arak University of Medical Sciences, Arak, Iran.

Ebrahimimonfared, Mohsen. Department of Neurology, Valiasr Hospital, School of Medicine, Arak University of Medical Sciences, Arak, Iran.

Rafeie, Mohammad. Department of Biostatistics and Epidemiology, School of Medicine, Arak University of Medical Sciences, Arak, Iran. rafeie@yahoo.com.

Safi, Fatemeh. Department of Radiology, Valiasr Hospital, School of Medicine, Arak University of Medical Sciences, Arak, Iran.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5473016>

Year of Publication

2017

446.

Maternal Uniparental Disomy 14 (Temple Syndrome) as a Result of a Robertsonian Translocation.

Bertini V; Fogli A; Bruno R; Azzara A; Michelucci A; Mattina T; Bertelloni S; Valetto A.
OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
Molecular syndromology. 8(3):131-138, 2017 May.

[Journal Article]

UI: 28588434

Maternal uniparental disomy of chromosome 14 (upd(14)mat) or Temple syndrome is an imprinting disorder associated with a relatively mild phenotype. The absence of specific congenital malformations makes this condition underdiagnosed in clinical practice. A boy with a de novo robertsonian translocation 45,XY,rob(13;14)(q10;q10) is reported; a CGH/SNP array showed a loss of heterozygosity in 14q11.2q13.1. The final diagnosis of upd(14)mat was made by microsatellite analysis, which showed a combination of heterodisomy and isodisomy for different regions of chromosome 14. Obesity after initial failure to thrive developed, while compulsive eating habits were not present, which was helpful for the clinical differential diagnosis of Prader-Willi syndrome. In addition, the boy presented with many phenotypic features associated with upd(14)mat along with hypoesthesia to pain, previously unreported in this disorder, and bilateral cryptorchidism, also rarely described. These features, as well as other clinical manifestations (i.e., truncal obesity, altered pubertal timing), may suggest a hypothalamic-pituitary involvement. A detailed cytogenetic and molecular characterization of the genomic rearrangement is presented. Early genetic diagnosis permits a specific follow-up of children with upd(14)mat in order to optimize the long-term outcome.

Version ID

1

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Authors Full Name

Bertini, Veronica; Fogli, Antonella; Bruno, Rossella; Azzara, Alessia; Michelucci, Angela; Mattina, Teresa; Bertelloni, Silvano; Valetto, Angelo.

Institution

Bertini, Veronica. Molecular Genetics Unit, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy. Fogli, Antonella. Molecular Genetics Unit, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy.

Bruno, Rossella. Molecular Genetics Unit, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy.

Bruno, Rossella. Department of Surgical, Medical, Molecular Pathology and Critical Area, University of Pisa, Pisa, Italy.

Azzara, Alessia. Molecular Genetics Unit, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy.

Michelucci, Angela. Molecular Genetics Unit, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy.

Mattina, Teresa. Centro di Riferimento per la Prevenzione Diagnosi e Cura delle Malattie Genetiche Rare, Università di Catania, Catania, Italy.

Bertelloni, Silvano. Adolescent Medicine, Pediatric Division, Department of Obstetrics, Gynecology and Pediatrics, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy.

Valetto, Angelo. Molecular Genetics Unit, Azienda Ospedaliero-Universitaria (AOU) Pisana, Pisa, Italy.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5448444>

Year of Publication

2017

447.

Male child with somatic mosaic Osteopathia Striata with Cranial Sclerosis caused by a novel pathogenic AMER1 frameshift mutation.

Hague J; Delon I; Brugger K; Martin H; Sparnon L; Simonic I; Abbs S; Park SM.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

American Journal of Medical Genetics. Part A. 173(7):1931-1935, 2017 Jul.

[Journal Article]

UI: 28497491

Osteopathia striata with cranial sclerosis (OSCS; OMIM #300373) is a rare X-linked dominant condition caused by mutations in the AMER1 gene (also known as WTX or FAM123B). It is a condition which usually affects females in whom the clinical phenotype can be extremely variable. Conversely affected males typically die in utero or during the neonatal period [Perdu et al. (); Clinical Genetics 80: 383-388; Vasiljevic et al. (); Prenatal Diagnosis 35: 302-304]. There have been a small number of reported cases of surviving males, including three patients who are somatic mosaic for the condition [Chenier, Noor, Dupuis, Stavropoulos, & Mendoza-Londono, (); American Journal of Medical Genetics Part A 158A: 2946-2952; Holman et al. (); American

Journal of Medical Genetics Part A 155A: 2397-2408; Joseph, Shoji, & Econs, (); The Journal of Clinical Endocrinology and Metabolism 95: 1506-1507]. We report a case of a male child who has proven somatic mosaicism for OPCS associated with a novel pathogenic frameshift mutation, c.607_611delAGGCC (p.Arg203 fs) in AMER1. We describe the multisystemic clinical features which include macrocephaly with ventriculomegaly and requirement for ventriculoperitoneal shunt, cleft palate, and respiratory difficulties after birth requiring tracheostomy insertion, persistent patent ductus arteriosus, failure to thrive and gastrostomy insertion, growth retardation, ophthalmoplegia, kidney malformation, cryptorchidism, and developmental delay. The use of new technologies with next generation sequencing (NGS) may improve the detection rate of mosaicism in rare conditions.

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Version ID

1

Status

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Author NameID

Hague, Jennifer; ORCID: <http://orcid.org/0000-0002-9779-8009>

Authors Full Name

Hague, Jennifer; Delon, Isabelle; Brugger, Kim; Martin, Howard; Sparnon, Leanne; Simonic, Ingrid; Abbs, Stephen; Park, Soo-Mi.

Institution

Hague, Jennifer. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK. Delon, Isabelle. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Brugger, Kim. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Martin, Howard. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Sparnon, Leanne. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Simonic, Ingrid. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Abbs, Stephen. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Park, Soo-Mi. Department of Clinical Genetics, Cambridge University Hospitals NHS Foundation Trust, Cambridge, UK.

Year of Publication

2017

448.

Pediatric urologists must advocate for improved quality of care in patients with cryptorchidism.

Romao RL.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Canadian Urological Association Journal. 11(1-2Suppl1):S33, 2017 Jan-Feb.

[Journal Article]

UI: 28265314

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Authors Full Name

Romao, Rodrigo L P.

Institution

Romao, Rodrigo L P. Division of Pediatric Urology, Division of Pediatric Surgery, IWK Health
Centre, Dalhousie University, Halifax, NS, Canada.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5332231>

Year of Publication

2017

449.

Pediatric Penile and Glans Anthropometry Nomograms: An Aid in Hypospadias Management.

Puri A; Sikdar S; Prakash R.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Indian Association of Pediatric Surgeons. 22(1):9-12, 2017 Jan-Mar.

[Journal Article]

UI: 28082769

OBJECTIVE: To establish pediatric penile and glans anthropometry nomograms. This may be used as a reference model for penile assessment while managing hypospadias.

PATIENTS AND METHODS: Between October 2012 and September 2013, 263 boys of varying ages (0-16 years) were included in the study. Those with genetic, endocrine disorders, having genital anomaly, undescended testis, neonates, and infants with a nonretractile prepuce, with multiple congenital anomalies and refusal to take part in the study were excluded. Evaluated outcome variables were stretched penile length, glans circumference (GC) at coronal sulcus, glans diameter at coronal sulcus (Gdcl), mid glans diameter, and ventral glans length. Glans ratios were generated by dividing Gdcl by GC. Data were expressed as mean, median, and standard deviation. Correlation between age and variables was evaluated using nonparametric Spearman's rank correlation coefficient.

RESULTS: The patients were divided in six age groups, namely 0-1 (n = 61), 1-3 (n = 37), 3-5 (n = 36), 5-7 (n = 36), 7-12 (n = 45), and >12 years (n = 48). Gdcl was the maximum transverse glans diameter and based on it small glans size varied widely from 8.9 to 35.04 mm for various age groups. Although glans anthropometry showed age-related changes, glans ratio remained relatively constant between 0.49 and 0.53 (mean: 0.5 +/- 0.051, r = 0.29). All the variables except glans ratio showed a significant positive correlation with age (r = 0.954-0.98, P < 0.01).

CONCLUSION: Penile anthropometry nomograms provide a reference model for hypospadias. This may aid in (a) objective preoperative assessment of glans size (b) patient selection for preoperative hormonal stimulation (c) provides a yardstick for postoperative cosmesis.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Puri, Archana; Sikdar, Satyajit; Prakash, Raghu.

Institution

Puri, Archana. Department of Pediatric Surgery, Lady Hardinge Medical College, New Delhi, India. Sikdar, Satyajit. Department of Pediatric Surgery, Lady Hardinge Medical College, New Delhi, India.

Prakash, Raghu. Department of Pediatric Surgery, Lady Hardinge Medical College, New Delhi, India.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5217151>

Year of Publication

2017

450.

Association of Levels of Serum Inhibin B and Follicle-stimulating Hormone with Testicular Vascularity, Volume, and Echotexture in Children with Undescended Testes.

Chinya A; Ratan SK; Aggarwal SK; Garg A; Mishra TK.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Indian Association of Pediatric Surgeons. 22(1):3-8, 2017 Jan-Mar.

[Journal Article]

UI: 28082768

AIMS: The aim of our study was to assess the association between reproductive hormones (inhibin B [inh B], follicle-stimulating hormone [FSH]) with testicular volume, echogenicity, and blood flow (resistive index [RI]) in children with undescended testis (UDT).

SETTINGS AND DESIGN: This was a prospective study of 1-year study duration.

MATERIALS AND METHODS: A total of 33 patients (16 unilateral and 17 bilateral) UDTs aged 5-12 years with palpable UDT were included in the study. Morning fasting blood samples were taken for estimation of serum inh B and FSH as well as inh B/FSH ratio. Testicular ultrasound was done to compute testicular volume, testicular echogenicity, and testicular vascularity in terms of RI.

RESULTS: The mean age of patients enrolled in the study was 8.29 years for unilateral UDT and 7.97 years in bilateral UDT and it was comparable. The study groups were further subdivided into two age-wise subgroups school goers (5-8 years) and prepubertal (9-12 years). The values of inh B, FSH, and inh B/FSH ratios as well as mean testicular volume were comparable between both groups and subgroups. Overall mean testicular volume had a positive correlation with FSH, inh B, and inh B/FSH, but statistical significance was reached only for inh B ($P < 0.001$) in children with both unilateral and bilateral UDT. Apart from five patients with hypoechogenicity within the testis, all remaining testes were of homogenous echotexture with no instances of irregular echogenicity or tumor. Children with $RI > 0.6$ were separately studied. The incidence of high RI (> 0.6) was also

comparable in unilateral or bilateral disease. These subjects had unfavorable biochemical parameters in terms of low inh B levels and high FSH levels.

CONCLUSIONS: Our findings hint to the fact that palpable UDT forms a homogenous group, whether unilateral or bilateral, whereas impalpable testes may form a separate category and need further studies to substantiate this hypothesis.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Chinya, Abhishek; Ratan, Simmi K; Aggarwal, Satish K; Garg, Anju; Mishra, T K.

Institution

Chinya, Abhishek. Department of Pediatric Surgery, Maulana Azad Medical College, New Delhi, India. Ratan, Simmi K. Department of Pediatric Surgery, Maulana Azad Medical College, New Delhi, India.

Aggarwal, Satish K. Department of Pediatric Surgery, Maulana Azad Medical College, New Delhi, India.

Garg, Anju. Department of Radiodiagnosis, Maulana Azad Medical College, New Delhi, India.

Mishra, T K. Department of Biochemistry, Maulana Azad Medical College, New Delhi, India.

PMC Identifier

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Year of Publication

2017

451.

Previously Unreported Biallelic Mutation in DNAJC19: Are Sensorineural Hearing Loss and Basal Ganglia Lesions Additional Features of Dilated Cardiomyopathy and Ataxia (DCMA) Syndrome?.

Ucar SK; Mayr JA; Feichtinger RG; Canda E; Coker M; Wortmann SB.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Jimd Reports. 35:39-45, 2017.

[Journal Article]

UI: 27928778

BACKGROUND: Dilated cardiomyopathy (DCM), non-progressive cerebellar ataxia (A), testicular dysgenesis, growth failure, and 3-methylglutaconic aciduria are the hallmarks of DNAJC19 defect (or DCMA syndrome) due to biallelic mutations in DNAJC19. To date DCMA syndrome has been reported in 19 patients from Canada and in two Finnish siblings. The underlying pathomechanism is unknown; however, DNAJC19 is presumed to be involved in mitochondrial membrane related processes (e.g., protein import and cardiolipin remodeling). Here, we report an additional patient with progressive cerebellar atrophy and white matter changes.

PATIENT AND METHODS: A Turkish boy presented at age 2 months with dilated cardiomyopathy (initially worsening then stabilizing in the second year of life), growth failure, bilateral cryptorchidism, and facial dysmorphism. Mental and motor developmental were, respectively, moderately and severely delayed. Profound intentional tremor and dyskinesia, spasticity (particularly at the lower extremities), and dystonia were observed. Sensorineural hearing loss was also diagnosed. MRI showed bilateral basal ganglia signal alterations. Plasma lactate levels were increased, as was urinary excretion of 3-methylglutaconic acid. He deceased aged 3 years.

RESULTS: Sanger Sequencing of DNAJC19 confirmed the clinical diagnosis of DNAJC19 defect by revealing the previously unreported homozygous stop mutation c.63delC (p.Tyr21*). Investigation of enzymes of mitochondrial energy metabolism revealed decreased activity of cytochrome c oxidase in muscle tissue.

DISCUSSION: Sensorineural hearing loss and bilateral basal ganglia lesions are common symptoms of mitochondrial disorders. This is the first report of an association with DNAJC19 defect.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Ucar, Sema Kalkan; Mayr, Johannes A; Feichtinger, Rene G; Canda, Ebru; Coker, Mahmut; Wortmann, Saskia B.

Institution

Ucar, Sema Kalkan. Division of Metabolic Disease, Ege University Medical Faculty, Department of Pediatrics, Izmir, 35100, Turkey. sema.kalkan.ucar@ege.edu.tr. Mayr, Johannes A. Department of Pediatrics Salzburger Landeskliniken and Paracelsus Medical University Salzburg, Salzburg, Austria.

Feichtinger, Rene G. Department of Pediatrics Salzburger Landeskliniken and Paracelsus Medical University Salzburg, Salzburg, Austria.

Canda, Ebru. Division of Metabolic Disease, Ege University Medical Faculty, Department of Pediatrics, Izmir, 35100, Turkey.

Coker, Mahmut. Division of Metabolic Disease, Ege University Medical Faculty, Department of Pediatrics, Izmir, 35100, Turkey.

Wortmann, Saskia B. Department of Pediatrics Salzburger Landeskliniken and Paracelsus Medical University Salzburg, Salzburg, Austria.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5585102>

Year of Publication

2017

452.

The diagnostic utility of combined diffusion-weighted imaging and conventional magnetic resonance imaging for detection and localization of non palpable undescended testes.

Emad-Eldin S., Abo-Elnagaa N., Hanna S.A., Abdel-Satar A.H.

Embase

Journal of Medical Imaging and Radiation Oncology. 60 (3) (pp 344-351), 2016. Date of Publication: 01 Jun 2016.

[Article]

AN: 609999818

Introduction We aimed to evaluate the diagnostic performance of combined diffusion weighted imaging (DWI) and conventional magnetic resonance imaging (MRI), including fat-suppression T2WI for identification and localization of non palpable undescended testes (UDTs). **Methods** This prospective study included 40 consecutive patients, with 47 non-palpable undescended testes (unilateral in 33 cases and bilateral in seven cases). Their age ranged from 5 months to 18 years, mean = 7.5 +/- 5.9 years. MRI examinations included T1WI, T2WI, fat-suppression T2WI and DWI at b value of 50, 400 and 800 s/mm². All patients underwent laparoscopic exploration. **Results** According to the laparoscopy findings, the final diagnoses of the location of UDTs were: intra-canalicular (n = 18, 38%), low intra-abdominal (n = 6, 13%), high intra-abdominal (n = 5, 11%). Absent or vanishing testes were detected in 18 cases (38%). The diagnostic accuracy, sensitivity, specificity of combined DWI and conventional MRI were 95.7%, 93.5% and 100% respectively. **Conclusion** Combined DWI and MRI showed a greater performance compared to

conventional MRI alone for identification of non-palpable UDTs. Based on our findings, we can obviate the need for diagnostic laparoscopy in patients who had preoperative detection of inguinal testes or nubbins. However, laparoscopy is still needed to confirm an absent rather than undetected non-viable abdominal testes.

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Embase

Author NameID

Hanna, Sameh Az; ORCID: <http://orcid.org/0000-0002-1197-0489>

Institution

(Emad-Eldin, Abo-Elnagaa, Hanna) Diagnostic and Intervention Radiology Department, Cairo University Hospitals, El-Manial, Cairo 11956, Egypt (Abdel-Satar) Pediatric Surgery Department, Cairo University Hospitals, Cairo, Egypt

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453.

Anorectal malformations associated spinal cord anomalies.

Totonelli G., Morini F., Catania V.D., Schingo P.M., Mosiello G., Palma P., Iacobelli B.D., Bagolan P.

Embase

Pediatric Surgery International. 32 (8) (pp 729-735), 2016. Date of Publication: 2016.

[Article]

AN: 622047901

Purpose The present study aims to identify clinical and pathological factors that can predict the risk of spinal cord anomalies (SCA) in patients with anorectal malformations (ARM), the need for neurosurgery, and to define the impact of SCA on the outcome of patients with ARM. Methods A 16-year retrospective analysis of all patients treated at a single tertiary children's Hospital with

diagnosis of ARM. Data were collected to assess the impact of defined clinical characteristics on prevalence of SCA (detected at MRI). Children surgically treated or not for SCA were compared for age, clinical symptoms and type of anomalies at surgery or at last follow-up, respectively. Moreover, patients with intermediate/high ARMs, with or without SCA were compared for neurogenic bladder (NB), constipation, soiling and need for bowel management (BM). Results Two hundred and seventy-five children were treated for ARM in the study period, 142 had spinal MRI that showed SCA in 85. Patients with SCA had significantly higher prevalence of preterm birth ($p < 0.05$), cardiac anomalies ($p = 0.02$), vertebral anomalies ($p = 0.0075$), abnormal sacrum ($p < 0.0001$), and VACTERL association ($p = 0.0233$). Ten patients were surgically treated for SCA. The prevalence of neurological bladder and neuro-motor deficits, of vertebral and genital anomalies, particularly cryptorchidism, was significantly higher in the operated group ($p < 0.01$, for each analysis). In patients with intermediate/high ARMs, no significant difference was observed between those with or without SCA, in terms of prevalence of NB, intestinal function and need for BM. Conclusions In patients with ARM, factors that can predict a higher prevalence of SCA and also determine an increased indication to neurosurgery may be identified. SCA by itself does not seem to affect the functional prognosis of children with intermediate/high ARM. These data may help physicians in stratifying the clinical and diagnostic pathway of patients with ARM.

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Status

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Institution

(Totonelli, Morini, Catania, Iacobelli, Bagolan) Neonatal Surgery Unit, Department of Medical and Surgical Neonatology, Bambino Gesù Children's Hospital, IRCCS, Piazza S. Onofrio, 4, Rome 00165, Italy (Schingo) Department of Radiology, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

(Mosiello) Neuro-Urology Unit, Department of Surgery, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

(Palma) Neurosurgery Unit, Department of Neuroscience and Neurorehabilitation, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

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454.

The epidemiologic evidence linking prenatal and postnatal exposure to endocrine disrupting chemicals with male reproductive disorders: A systematic review and meta-analysis.

Bonde J.P., Flachs E.M., Rimborg S., Glazer C.H., Giwercman A., Ramlau-Hansen C.H., Hougaard K.S., Hoyer B.B., Haervig K.K., Petersen S.B., Rylander L., Specht I.O., Toft G., Brauner E.V.

Embase

Human Reproduction Update. 23 (1) (pp 104-125), 2016. Date of Publication: 2016.

[Article]

AN: 621789462

BACKGROUND: More than 20 years ago, it was hypothesized that exposure to prenatal and early postnatal environmental xenobiotics with the potential to disrupt endogenous hormone signaling might be on the causal path to cryptorchidism, hypospadias, low sperm count and testicular cancer. Several consensus statements and narrative reviews in recent years have divided the scientific community and have elicited a call for systematic transparent reviews. We aimed to fill this gap in knowledge in the field of male reproductive disorders.

OBJECTIVE AND RATIONALE: The aim of this study was to systematically synthesize published data on the risk of cryptorchidism, hypospadias, low sperm counts and testicular cancer following in utero or infant exposure to chemicals that have been included on the European Commission's list of Category 1 endocrine disrupting chemicals defined as having documented adverse effects due to endocrine disruption in at least one intact organism.

SEARCH METHOD(S): A systematic literature search for original peer reviewed papers was performed in the databases PubMed and Embase to identify epidemiological studies reporting associations between the outcomes of interest and exposures documented by biochemical analyses of biospecimens including maternal blood or urine, placenta or fat tissue as well as amnion fluid, cord blood or breast milk; this was followed by meta-analysis of quantitative data.

OUTCOME(S): The literature search resulted in 1314 references among which we identified 33 papers (28 study populations) fulfilling the eligibility criteria. These provided 85 risk estimates of links between persistent organic pollutants and rapidly metabolized compounds (phthalates and Bisphenol A) and male reproductive disorders. The overall odds ratio (OR) across all exposures and outcomes was 1.11 (95% CI 0.91-1.35). When assessing four specific chemical subgroups with sufficient data for meta-analysis for all outcomes, we found that exposure to one of the four compounds, p,p'-DDE, was related to an elevated risk: OR 1.35 (95% CI 1.04-1.74). The data did not indicate that this increased risk was driven by any specific disorder.

WIDER IMPLICATION(S): The current epidemiological evidence is compatible with a small increased risk of male reproductive disorders following prenatal and postnatal exposure to some persistent environmental chemicals classified as endocrine disruptors but the evidence is limited. Future epidemiological studies may change the weight of the evidence in either direction. No evidence of distortion due to publication bias was found, but exposure-response relationships are not evident. There are insufficient data on rapidly metabolized endocrine disruptors and on specific exposure-outcome relations. A particular data gap is evident with respect to delayed effects on semen quality and testicular cancer. Although high quality epidemiological studies are still sparse, future systematic and transparent reviews may provide pieces of evidence contributing to the narrative and weight of the evidence assessments in the field.

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Institution

(Bonde, Flachs, Glazer, Hoyer, Haervig, Petersen, Specht, Brauner) Department of Occupational and Environmental Medicine, Bispebjerg University Hospital, Copenhagen NV DK-2400, Denmark (Bonde) Institute of Public Health, University of Copenhagen, Copenhagen K DK-1016, Denmark

(Rimborg) The Royal Library/ University of Copenhagen Library, Copenhagen N DK-2200, Denmark

(Giwercman) Molecular Reproductive Medicine, Department of Translational Medicine, Lund University, Lund, Sweden

(Ramlaau-Hansen) Institute of Public Health, Aarhus University, Aarhus C DK-8000, Denmark

(Hougaard) National Research Centre for the Working Environment, Copenhagen O DK-2100, Denmark

(Hoyer, Toft) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus N DK-8200, Denmark

(Rylander) Department of Occupational and Environmental Medicine, University of Lund, Lund SE-221 85, Sweden

(Brauner) Research Center for Prevention and Health (RCPH), University of Copenhagen, Glostrup DK-2600, Denmark

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455.

Laparoscopic treatment of nonpalpable testicle. Factors predictive for diminished size.

Bracho-Blanchet E., Unda-Haro S., Ordorica-Flores R., Nieto-Zermeno J., Zalles-Vidal C.,
Fernandez-Portilla E., Davila-Perez R.

Embase

Journal of Pediatric Surgery. 51 (7) (pp 1201-1206), 2016. Date of Publication: July 2016.

[Article]

AN: 2000646914

Purpose: The purposes of this study were to demonstrate the usefulness of laparoscopy in intraabdominal testicle (IAT) and to determine factors associated with diminished size during the final outcome after laparoscopic orchidopexy.

Method(s): This is a retrospective analysis of consecutive patients from 1999 to 2013 with a minimum follow-up of 1 year. Patient and testicular factors were related to diminished size.

Result(s): Sixty one patients, and 92 testicles were included. Median age at operation was 42 months. Initially we found 66 normal sized testes (71.7%), 22 hypotrophic (23.9%) and four atrophic (4.3%). Eighty seven testes were brought down laparoscopically, 50 in one surgical stage and 37 in two stages. Mean follow-up was 40.2 months and the final outcome was success: 73.5% and diminished size: 26.5%. Variables associated with diminished size were hypotrophy during initial evaluation, short spermatic vessels, section of spermatic vessels, two-stage surgery and tension to reach contralateral inguinal ring. Multivariate analysis showed that initial hypotrophy (odds ratio [OR] 4.96, confidence interval 95% [CI] 1.36-18.10) and tension to reach contralateral ring (OR 4.11, 95% CI 1.18-14.34) were associated with diminished size.

Conclusion(s): Laparoscopy is useful in treating IAT. Initial size and tension to reach contralateral ring are factors associated with diminished size.

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Institution

(Bracho-Blanchet, Unda-Haro, Ordorica-Flores, Nieto-Zermeno, Zalles-Vidal, Fernandez-Portilla, Davila-Perez) Departamento de Cirugia General, Hospital Infantil de Mexico Federico Gomez, Dr. Marquez 162 Col. Doctores, Mexico, D.F. 06720, Mexico

Publisher

W.B. Saunders

Year of Publication

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456.

Associated congenital anomalies in infants with isolated gastroschisis: A single-institutional experience.

Corona-Rivera J.R., Nieto-Garcia R., Lopez-Marure E., Cardenas-Ruiz Velasco J.J., Bobadilla-Morales L., Mellin-Sanchez E.L., Aguirre-Guillen R.L., Perez-Ramirez R.O., Zapata-Aldana E., Sandoval-Talamantes A.K., Solis-Ledezma S., Corona-Rivera A., Gomez-Ruiz L.M.

Embase

American Journal of Medical Genetics, Part A. 170 (2) (pp 316-321), 2016. Date of Publication: 01 Feb 2016.

[Article]

AN: 607447577

The aim of our study was to determine the frequency and type of associated congenital anomalies in patients with isolated gastroschisis born at the Dr. Juan I. Menchaca Civil Hospital of Guadalajara (Guadalajara, Mexico), and to explore its possible association with the included outcome variables. One hundred-eight cases with isolated gastroschisis were reviewed from 2009 to 2014. The occurrence of intestinal and extraintestinal associated anomalies (either secondary or primary) was prospectively assessed. The type of gastroschisis, length of hospital stay (LOS), and in-hospital mortality were outcome variables for statistical analysis. Of infants with gastroschisis, 52 (48.1%) had one or more associated anomalies (AA), with increased odds in males (OR=2.3, 95%CI: 1.1-5.0). AA classified, as secondary and primary were present in 34.3 and 5.6% of patients, respectively. Of secondary AA, 25.9% were intestinal anomalies, and 17.6% were extraintestinal. Primary AA were congenital heart disease (n=3), meningomyelocele, and hydrocephaly and amniotic band sequence in one instance, respectively. Multivariate logistic regression showed that secondary AA (both intestinal and extraintestinal) were associated with

complex gastroschisis, prolonged LOS, and in-hospital death, whereas primary AA were not related to a worse outcome. Our results highlight the pathogenic importance of properly investigating and categorizing the presence of others secondary or primary AA when diagnosis of gastroschisis is made.

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(Corona-Rivera, Bobadilla-Morales, Mellin-Sanchez, Aguirre-Guillen, Zapata-Aldana, Sandoval-Talamantes, Solis-Ledezma, Corona-Rivera) Center for Registry and Research in Congenital Anomalies (CRIAC), Service of Genetics and Cytogenetics Unit, Pediatrics Division, Dr. Juan I. Menchaca Civil Hospital of Guadalajara, Guadalajara, Jalisco, Mexico (Corona-Rivera, Bobadilla-Morales, Corona-Rivera) Department of Molecular Biology and Genomics, Dr. Enrique Corona-Rivera Institute of Human Genetics, Health Sciences University Center, University of Guadalajara, Guadalajara, Jalisco, Mexico

(Nieto-Garcia) Service of Cardiology, Dr. Juan I. Menchaca Civil Hospital of Guadalajara, Guadalajara, Jalisco, Mexico

(Lopez-Marure) Service of Radiology, Dr. Juan I. Menchaca Civil Hospital of Guadalajara, Guadalajara, Jalisco, Mexico

(Cardenas-Ruiz Velasco) Service of Pediatric Surgery, Dr. Juan I. Menchaca Civil Hospital of Guadalajara, Guadalajara, Jalisco, Mexico

(Perez-Ramirez, Gomez-Ruiz) Service of Neonatology, Pediatrics Division, Dr. Juan I. Menchaca Civil Hospital of Guadalajara, Guadalajara, Jalisco, Mexico

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2016

Male reproductive disorders and fertility trends: Influences of environment and genetic susceptibility.

Skakkebaek N.E., Rajpert-De Meyts E., Buck Louis G.M., Toppari J., Andersson A.-M., Eisenberg M.L., Jensen T.K., Jorgensen N., Swan S.H., Sapra K.J., Ziebe S., Priskorn L., Juul A.

Embase

Physiological Reviews. 96 (1) (pp 55-97), 2016. Date of Publication: 18 Nov 2015.

[Article]

AN: 606942117

It is predicted that Japan and European Union will soon experience appreciable decreases in their populations due to persistently low total fertility rates (TFR) below replacement level (2.1 child per woman). In the United States, where TFR has also declined, there are ethnic differences. Caucasians have rates below replacement, while TFRs among African-Americans and Hispanics are higher. We review possible links between TFR and trends in a range of male reproductive problems, including testicular cancer, disorders of sex development, cryptorchidism, hypospadias, low testosterone levels, poor semen quality, childlessness, changed sex ratio, and increasing demand for assisted reproductive techniques. We present evidence that several adult male reproductive problems arise in utero and are signs of testicular dysgenesis syndrome (TDS). Although TDS might result from genetic mutations, recent evidence suggests that it most often is related to environmental exposures of the fetal testis. However, environmental factors can also affect the adult endocrine system. Based on our review of genetic and environmental factors, we conclude that environmental exposures arising from modern lifestyle, rather than genetics, are the most important factors in the observed trends. These environmental factors might act either directly or via epigenetic mechanisms. In the latter case, the effects of exposures might have an impact for several generations post-exposure. In conclusion, there is an urgent need to prioritize research in reproductive physiology and pathophysiology, particularly in highly industrialized countries facing decreasing populations. We highlight a number of topics that need attention by researchers in human physiology, pathophysiology, environmental health sciences, and demography.

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(Skakkebaek, Rajpert-De Meyts, Buck Louis, Toppari, Andersson, Eisenberg, Jensen, Jorgensen, Swan, Sapra, Ziebe, Priskorn, Juul) Department of Growth and Reproduction and EDMaRC, Rigshospitalet, University of Copenhagen, Copenhagen, Denmark (Skakkebaek, Rajpert-De

Meyts, Buck Louis, Toppari, Andersson, Eisenberg, Jensen, Jorgensen, Swan, Sapra, Ziebe, Priskorn, Juul) Division of Epidemiology, Statistics and Prevention Research, Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health, Bethesda, MD, United States

(Skakkebaek, Rajpert-De Meyts, Buck Louis, Toppari, Andersson, Eisenberg, Jensen, Jorgensen, Swan, Sapra, Ziebe, Priskorn, Juul) Department of Physiology and Pediatrics, University of Turku, Turku University Hospital, Turku, Finland

(Skakkebaek, Rajpert-De Meyts, Buck Louis, Toppari, Andersson, Eisenberg, Jensen, Jorgensen, Swan, Sapra, Ziebe, Priskorn, Juul) Stanford University, Stanford, CA, United States

(Skakkebaek, Rajpert-De Meyts, Buck Louis, Toppari, Andersson, Eisenberg, Jensen, Jorgensen, Swan, Sapra, Ziebe, Priskorn, Juul) Icahn School of Medicine at Mount Sinai, New York, NY, United States

(Skakkebaek, Rajpert-De Meyts, Buck Louis, Toppari, Andersson, Eisenberg, Jensen, Jorgensen, Swan, Sapra, Ziebe, Priskorn, Juul) The Fertility Clinic, Rigshospitalet, Copenhagen, Denmark

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2016

458.

Congenital inguinal hernia, hydrocoele and undescended testis.

Khoo A.K., Cleeve S.J.

Embase

Surgery (United Kingdom). 34 (5) (pp 226-231), 2016. Date of Publication: 01 May 2016.

[Article]

AN: 610176221

Congenital inguinal hernias (CIH), hydrocoeles and undescended testes (UDT) are common groin conditions in neonates, infants and children that are encountered by general practitioners, paediatricians, general surgeons and paediatric surgeons. CIH, hydrocoeles and UDT share a common embryological origin. Clinical differentiation between the three conditions can be challenging particularly as they may exist in isolation or combination in the same patient.

Accurate clinical distinction is imperative as the management and outcome is different for each condition. Surgery and outcomes for these conditions is discussed.

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(Khoo, Cleeve) Royal London Hospital, London, United Kingdom

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459.

Testicular problems in children.

Godbole P.P.

Embase

Paediatrics and Child Health (United Kingdom). 26 (6) (pp 246-251), 2016. Date of Publication: 01 Jun 2016.

[Review]

AN: 609614731

Testicular problems in children can be either congenital or acquired. These problems are often difficult to diagnose and carry significant sequelae if untreated. Early surgical consultation is often needed for correction of the problem. This article reviews the pathophysiology of the most common paediatric testicular abnormalities with emphasis on the diagnostic modalities employed and discusses the current treatment choices.

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Institution

(Godbole) Sheffield Children's Foundation Trust, Sheffield, United Kingdom

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460.

New Mutations Associated with Rasopathies in a Central European Population and Genotype-Phenotype Correlations.

Cizmarova M., Hlinkova K., Bertok S., Kotnik P., Duba H.C., Bertalan R., Polockova K., Kostalova L., Pribilincova Z., Hlavata A., Kovacs L., Ilencikova D.

Embase

Annals of Human Genetics. 80 (1) (pp 50-62), 2016. Date of Publication: 01 Jan 2016.

[Article]

AN: 607220991

We performed the genetic analysis of Rasopathy syndromes in patients from Central European by direct sequencing followed by next generation sequencing of genes associated with Rasopathies. All 51 patients harboured the typical features of Rasopathy syndromes. Thirty-five mutations were identified in the examined patients (22 in PTPN11, two in SOS1, one in RIT1, one in SHOC2, two in HRAS, three in BRAF, two in MAP2K1 and two in the NF1 gene). Two of them (p.Gly392Glu in the BRAF gene and p.Gln164Lys in the MAP2K1 gene) were novel with a potentially pathogenic effect on the structure of these proteins. Statistically significant differences in the presence of pulmonary stenosis (63.64% vs. 23.81%, $P = 0.013897$) and cryptorchidism (76.47% vs. 30%, $P = 0.040224$) were identified as the result of comparison of the prevalence of phenotypic features in patients with the phenotype of Noonan syndrome and mutation in the PTPN11 gene, with the prevalence of the same features in patients without PTPN11 mutation. Cryptorchidism as a statistically significant feature in our patients with PTPN11 mutation was not reported as significant in other European countries (Germany, Italy and Greece). The majority of mutations were clustered in exons 3 (45.45%), 8 (22.73%), and 13 (22.73%) of the PTPN11 gene.

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Institution

(Cizmarova, Hlinkova, Kostalova, Pribilincova, Hlavata, Kovacs, Ilencikova) 2nd Department of Pediatrics, University Children's Hospital, Bratislava, Slovakia (Bertok, Kotnik) Department of Endocrinology, Diabetes and Metabolic Diseases, University Children's Hospital, Ljubljana, Slovenia

(Duba, Ilencikova) Department of Human Genetics, Women's and Children's Clinic, Linz, Austria (Bertalan) Department of Pediatric Endocrinology, University Teaching Hospital, Veszprem, Hungary

(Polockova) Department of Pediatric Endocrinology, Hospital with Policlinic, Karvina, Czech Republic

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461.

A Systematic Review of Children's Environmental Health in Brazil.

Froes Asmus C.I.R., Camara V.M., Landrigan P.J., Claudio L.

Embase

Annals of Global Health. 82 (1) (pp 132-148), 2016. Date of Publication: 01 Jan 2016.

[Review]

AN: 612634372

In the region of the Americas, approximately 100,000 children under the age of 5 years die each year due to environmental hazards. Brazil, due to its large size and wide range of environmental challenges, presents numerous hazards to children's health. The aim of this study was to systematically review the scientific literature that describes children's exposures to environmental pollutants in Brazil and their effects on Brazilian children's health. A systematic review of the scientific literature was performed without language restrictions and time of publication (years). The literature search was conducted in the following key resources: PubMed (MEDLINE), Scopus and Web of Science with the MeSH Terms: Environmental exposure AND Brazil (filters: Human, Child [birth to 18 years] and Affiliation Author). The Virtual Health Library was also employed to access the databases Scielo and Lilacs. The search strategy was [DeCS Terms]: Child OR

adolescent AND Environmental exposure AND Brazil. Health effects in children associated with exposure to environmental pollutants in Brazil were reported in 74 studies, during the period between 1995 and 2015. The most frequently cited effect was hospital admission for respiratory causes including wheezing, asthma, and pneumonia among children living in areas with high concentrations of air pollutants. A broad spectrum of other health effects possibly linked to pollutants also was found such as prematurity, low birth weight, congenital abnormality (cryptorchidism, hypospadias, micropenis), poor performance in tests of psychomotor and mental development, and behavioral problems. Exposure to pesticides in utero and postnatally was associated with a high risk for leukemia in children <2 years old. These results show that there is a need in Brazil for stricter monitoring of pollutant emissions and for health surveillance programs especially among vulnerable populations such as pregnant women and young children.

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Institution

(Froes Asmus, Camara) Public Health Institute, School of Medicine, Federal University of Rio de Janeiro, Rio de Janeiro, Brazil (Froes Asmus, Landrigan, Claudio) Preventive Medicine Department, Icahn School of Medicine of Mount Sinai, New York, NY, United States (Landrigan) Arnhold Global Health Institute, Icahn School of Medicine of Mount Sinai, New York, NY, United States

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462.

Prenatal paracetamol exposure is associated with shorter anogenital distance in male infants.

Fisher B.G., Thankamony A., Hughes I.A., Ong K.K., Dunger D.B., Acerini C.L.

Embase

Human Reproduction. 31 (11) (pp 2642-2650), 2016. Date of Publication: 01 Nov 2016.

[Article]

AN: 613366925

STUDY QUESTION What is the relationship between maternal paracetamol intake during the masculinisation programming window (MPW, 8-14 weeks of gestation) and male infant anogenital distance (AGD), a biomarker for androgen action during the MPW? **SUMMARY ANSWER**

Intrauterine paracetamol exposure during 8-14 weeks of gestation is associated with shorter AGD from birth to 24 months of age. **WHAT IS ALREADY KNOWN** The increasing prevalence of male reproductive disorders may reflect environmental influences on foetal testicular development during the MPW. Animal and human xenograft studies have demonstrated that paracetamol reduces foetal testicular testosterone production, consistent with reported epidemiological associations between prenatal paracetamol exposure and cryptorchidism. **STUDY DESIGN, SIZE, DURATION** Prospective cohort study (Cambridge Baby Growth Study), with recruitment of pregnant women at ~12 post-menstrual weeks of gestation from a single UK maternity unit between 2001 and 2009, and 24 months of infant follow-up. Of 2229 recruited women, 1640 continued with the infancy study after delivery, of whom 676 delivered male infants and completed a medicine consumption questionnaire. **PARTICIPANTS/MATERIALS, SETTING, METHOD** Mothers self-reported medicine consumption during pregnancy by a questionnaire administered during the perinatal period. Infant AGD (measured from 2006 onwards), penile length and testicular descent were assessed at 0, 3, 12, 18 and 24 months of age, and age-specific Z scores were calculated. Associations between paracetamol intake during three gestational periods (<8 weeks, 8-14 weeks and >14 weeks) and these outcomes were tested by linear mixed models. Two hundred and twenty-five (33%) of six hundred and eighty-one male infants were exposed to paracetamol during pregnancy, of whom sixty-eight were reported to be exposed during 8-14 weeks. AGD measurements were available for 434 male infants. **MAIN RESULTS AND THE ROLE OF CHANCE** Paracetamol exposure during 8-14 weeks of gestation, but not any other period, was associated with shorter AGD (by 0.27 SD, 95% CI 0.06-0.48, $P = 0.014$) from birth to 24 months of age. This reduction was independent of body size. Paracetamol exposure was not related to penile length or testicular descent. **LIMITATIONS, REASONS FOR CAUTION** Confounding by other drugs or endocrine-disrupting chemicals cannot be discounted. The cohort was not fully representative of pregnant women in the UK, particularly in terms of maternal ethnicity and smoking prevalence. There is likely to have been misclassification of paracetamol exposure due to recall error. **WIDER IMPLICATIONS OF THE FINDINGS** Our observational findings support experimental evidence that intrauterine paracetamol exposure during the MPW may adversely affect male reproductive development. **STUDY**

FUNDING/COMPETING INTERESTS This work was supported by a European Union Framework V programme, the World Cancer Research Fund International, the Medical Research Council (UK), the Newlife Foundation for Disabled Children, the Evelyn Trust, the Mothercare Group

Foundation, Mead Johnson Nutrition, and the National Institute for Health Research Cambridge Comprehensive Biomedical Research Centre. The authors declare no conflict of interest. Copyright © 2016 The Author 2016. Published by Oxford University Press on behalf of the European Society of Human Reproduction and Embryology.

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Institution

(Fisher, Thankamony, Hughes, Ong, Dunger, Acerini) Department of Paediatrics, University of Cambridge, Addenbrooke's Hospital, Box 116, Level 8, Hills Road, Cambridge CB2 0QQ, United Kingdom (Ong) MRC Epidemiology Unit, University of Cambridge, Institute of Metabolic Science, Box 285, Cambridge Biomedical Campus, Cambridge CB2 0QQ, United Kingdom

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463.

Surgical pleth index in children younger than 24 months of age: A randomized double-blinded trial.

Harju J., Kalliomaki M.-L., Leppikangas H., Kiviharju M., Yli-Hankala A.

Embase

British Journal of Anaesthesia. 117 (3) (pp 358-364), 2016. Date of Publication: 01 Sep 2016.

[Article]

AN: 613253507

Background The surgical pleth index (SPI) is a measurement of intraoperative nociception. Evidence of its usability in children is limited. Given that the autonomic nervous system is still developing during the first years of life, the performance of the SPI on small children cannot be concluded from studies carried out in older age groups. Methods Thirty children aged <2 yr, planned for elective open inguinal hernia repair or open correction of undescended testicle, were recruited. The children were randomized into two groups; the saline group received ultrasound-

guided saline injection in the ilioinguinal and iliohypogastric nerve region before surgery and ropivacaine after surgery, whereas the block group received the injections in the opposite order. The SPI was recorded blinded and was analysed at the time points of intubation, incision, and when signs of inadequate anti-nociception were observed. Results There was a significant increase in the SPI after intubation ($P=0.019$) and after incision in the saline group ($P=0.048$), but not at the time of surgical incision in the block group ($P=0.177$). An increase in the SPI was also seen at times of clinically apparent inadequate anti-nociception ($P=0.008$). The between-patient variability of the SPI was large. Conclusions The SPI is reactive in small children after intubation and after surgical stimuli, but the reactivity of the SPI is rather small, and there is marked inter-individual variability in reactions. The reactivity is blunted by the use of ilioinguinal and iliohypogastric nerve block.

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Institution

(Harju, Kalliomaki, Leppikangas, Kiviharju, Yli-Hankala) Department of Anaesthesia, Tampere University Hospital, PL2000, Tampere 33521, Finland (Yli-Hankala) Medical School, University of Tampere, Tampere, Finland

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464.

Maternal first trimester serum levels of free-beta human chorionic gonadotrophin and male genital anomalies.

Schneuer F.J., Bower C., Holland A.J.A., Tasevski V., Jamieson S.E., Barker A., Lee L., Majzoub J.A., Nassar N.

Embase

Human Reproduction. 31 (8) (pp 1895-1903), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 611633714

Study Question Are maternal first trimester levels of serum free-beta hCG associated with the development of hypospadias or undescended testis (UDT) in boys? **Summary Answer** Overall, first trimester maternal levels of serum free-beta hCG are not associated with hypospadias or UDT. However, elevated levels were found in severe phenotypes (proximal hypospadias and bilateral UDT) suggesting an altered pathway of hormonal release in early pregnancy. **What is Known Already** Human chorionic gonadotrophin peaks in first trimester of pregnancy stimulating fetal testosterone production, which is key to normal male genital development. Endocrine-disrupting insults early in pregnancy have been associated with increased risk of common genital anomalies in males such as hypospadias and UDT. One plausible etiological pathway is altered release of hCG. **Study Design, Size, Duration** We conducted a record-linkage study of two separate populations of women attending first trimester aneuploidy screening in two Australian states, New South Wales (NSW) and Western Australia (WA), in 2006-2009 and 2001-2003, respectively. **Participants/Materials, Setting, Methods** Included were women who gave birth to a singleton live born male infant. There were 12 099 boys from NSW and 10 518 from WA included, of whom 90 and 77 had hypospadias; and 107 and 109 UDT, respectively. Serum levels of free-beta hCG were ascertained from laboratory databases and combined with relevant birth outcomes and congenital anomalies via record linkage of laboratory, birth, congenital anomalies and hospital data. Median and quartile levels of gestational age specific free-beta hCG multiple of the median (MoM) were compared between affected and unaffected boys. Logistic regression was used to evaluate the association between levels of free-beta hCG MoM and hypospadias or UDT, stratified by suspected placental dysfunction and co-existing anomalies. Where relevant, pooled analysis was conducted. **Main Results and the Role of Chance** There was no difference in median hCG levels amongst women with an infant with hypospadias (NSW = 0.88 MoM, P = 0.83; WA = 0.84 MoM, P = 0.76) or UDT (NSW = 0.89 MoM, P = 0.54; WA = 0.95 MoM, P = 0.95), compared with women with an unaffected boy (NSW = 0.92 MoM; WA = 0.88 MoM). Low (<25th centile) or high (>75th centile) hCG levels were not associated with hypospadias or UDT, nor when stratifying by suspected placental dysfunction and co-existing anomalies. However, there was a tendency towards high levels for severe types, although confidence intervals were wide. When combining NSW and WA results, high hCG MoM levels (>75th centile) were associated with increased risk of proximal hypospadias (odds ratio (OR) 4.34; 95% CI: 1.08-17.4) and bilateral UDT (OR 2.86; 95% CI: 1.02-8.03). **LIMITATIONS, REASONS FOR CAUTION** There were only small numbers of proximal hypospadias and bilateral UDT in both cohorts and although we conducted pooled analyses, results reported on these should be interpreted with caution. Gestational age by ultrasound may have been inaccurately estimated in small and large

for gestational age fetuses affecting hCG MoM calculation in those pregnancies. Despite the reliability of our datasets in identifying adverse pregnancy outcomes, we did not have pathology information to confirm tissue lesions in the placenta and therefore our composite outcome should be considered as a proxy for placental dysfunction. Wider Implications of the Findings This is one of the largest population-based studies examining the association between maternal first trimester serum levels of free-beta hCG and genital anomalies-hypospadias and UDT; and the first to compare specific phenotypes by severity. Overall, our findings does not support the hypothesis that alteration in maternal hCG levels is associated with the development of male genital anomalies; however, high hCG free-beta levels found in severe types suggest different underlying etiology involving higher production and secretion of hCG. These findings require further exploration and replication. STUDY FUNDING/COMPETING INTEREST(S) This work was funded by the National Health and Medical Research Council (NHMRC) grant APP1047263. N.N. is supported by a NHMRC Career Development Fellowship APP1067066. C.B. was supported by a NHMRC Principal Research Fellowship #634341. The funding agencies had no role in the design, analysis, interpretation or reporting of the findings. There are no competing interests. TRIAL REGISTRATION NUMBER Not applicable.

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Institution

(Schneuer, Nassar) Menzies Centre for Health Policy, School of Public Health, University of Sydney, Level 6 the Hub, Charles Perkins Centre D17, Sydney, NSW 2006, Australia (Bower, Jamieson) Telethon Kids Institute, University of Western Australia, Crawley, WA 6009, Australia (Holland) Discipline of Paediatrics and Child Health, Children's Hospital at Westmead, Sydney Medical School, University of Sydney, Sydney, NSW 2145, Australia

(Tasevski) Pathology North, NSW Health Pathology, Royal North Shore Hospital, St Leonards, NSW 2065, Australia

(Barker) Department of Paediatric Surgery, Princess Margaret Hospital for Children, Subiaco, WA 6008, Australia

(Lee) Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, David Geffen School of Medicine at UCLA, Los Angeles, CA 90095, United States

(Majzoub) Division of Endocrinology, Boston Children's Hospital, Harvard Medical School, CLSB 16028, 300 Longwood Avenue, Boston, MA 02115, United States

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465.

Outcome Analysis of Redo Orchiopexy: Scrotal vs Inguinal.

Lopes R.I., Naoum N.K., Chua M.E., Canil T., dos Santos J., Farhat W.A.

Embase

Journal of Urology. 196 (3) (pp 869-874), 2016. Date of Publication: September 2016.

[Article]

AN: 611362810

Purpose Redo orchiopexy after previous surgery is technically challenging and requires skills and care to ensure preservation of cord structures. We report our experience with redo orchiopexy in children. Materials and Methods We retrospectively reviewed patients who had undergone redo orchiopexy between January 2004 and May 2015. Variables evaluated included primary procedure, type of redo procedure, operative time, shift of surgical route, operative and postoperative complications, and testicular location at last followup. Results A total of 3,384 orchiopexies were performed during the study period, with 61 children (1.8%) requiring redo orchiopexy. Mean +/- SD patient age at redo orchiopexy was 6.4 +/- 3.6 years (range 1.5 to 17.1) and average followup was 24.9 months (2.1 to 99.6). The primary surgical procedure preceding redo surgery was inguinal orchiopexy in 45.9% of the patients, scrotal orchiopexy in 13.1% and laparoscopy in 13.1%, and 27.9% of patients were status post inguinal surgery (hernia/hydrocele repair). Redo surgery was performed by inguinal approach in 33 patients, while 28 children underwent a scrotal approach. There was no statistical difference in intraoperative and postoperative complication rates for the 2 approaches ($p = 0.52$ and $p = 0.26$, respectively). However, there was a statistically significant difference in overall operative time between approaches ($p = 0.003$) with scrotal orchiopexy being significantly shorter (53.1 minutes) compared to inguinal orchiopexy (84.6). Conclusions Scrotal and inguinal orchiopexy appear to be viable in managing secondarily ascending testes, with the scrotal approach offering some advantage in terms of length of procedure.

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(Lopes, Naoum, Chua, Canil, dos Santos, Farhat) Division of Urology, Department of Surgery, Hospital for Sick Children, University of Toronto, Toronto, Ontario, Canada

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466.

Long-Term Outcome of Low Scrotal Approach Orchiopexy without Ligation of the Processus Vaginalis.

Hyuga T., Kawai S., Nakamura S., Kubo T., Nakai H.

Embase

Journal of Urology. 196 (2) (pp 542-547), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 611157065

Purpose We performed low scrotal approach orchiopexy in patients with prescrotal cryptorchidism. The processus vaginalis was not ligated if it was not widely patent. We retrospectively evaluated the long-term outcomes of low scrotal approach orchiopexy without processus vaginalis ligation. **Materials and Methods** A total of 137 patients (227 testes) were diagnosed with prescrotal cryptorchidism between October 2009 and April 2014. All patients underwent low scrotal approach orchiopexy. Mean age at surgery was 34.9 months. The processus vaginalis was deemed to be not widely patent when a sound could not be passed into the abdominal cavity through the internal inguinal ring, and the processus vaginalis was not ligated in such cases. **Results** Intraoperative findings revealed that the processus vaginalis was widely patent in 10 testes and was not widely patent in 217. A widely patent processus vaginalis was closed via scrotal approach in 5 testes, while an inguinal approach was necessary in 5. Median followup was 44 months (range 20 to 73). Postoperative complications included reascending testis in 1 case where an inguinal approach was necessary. No patient manifested testicular atrophy or inguinal hernia. **Conclusions** Low scrotal approach orchiopexy is a useful and

safe procedure for treating patients with prescrotal cryptorchidism. Ligation is unnecessary when the processus vaginalis is not widely patent.

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Institution

(Hyuga, Kawai, Nakamura, Kubo, Nakai) Department of Pediatric Urology, Jichi Medical University, Children's Medical Center Tochigi, Tochigi, Japan

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467.

Prevalence of KISS1 Receptor mutations in a series of 603 patients with normosmic congenital hypogonadotrophic hypogonadism and characterization of novel mutations: A single-centre study.

Francou B., Paul C., Amazit L., Cartes A., Bouvattier C., Albarel F., Maiter D., Chanson P., Trabado S., Brailly-Tabard S., Brue T., Guiochon-Mantel A., Young J., Bouligand J.

Embase

Human Reproduction. 31 (6) (pp 1363-1374), 2016. Date of Publication: 15 Jun 2016.

[Article]

AN: 610736321

STUDY QUESTION What is the exact prevalence of Kisspeptin Receptor (KISS1R) mutations in the population of patients with normosmic congenital hypogonadotrophic hypogonadism (nCHH) by comparison with other genes, involved in gonadotrophin-releasing hormone (GnRH) release or action? SUMMARY ANSWER KISS1R mutants are responsible for the nCHH phenotype in only a small minority of cases and were less prevalent than GnRH Receptor (GNRHR) mutations.

WHAT IS KNOWN ALREADY The respective prevalence of each of the genetic causes of nCHH is unclear. Large series of patients are very rare and suffer from heterogeneity of the population of CHH studied. STUDY DESIGN, SIZE, DURATION Patients with nCHH were consecutively

enrolled in a single French referral centre and were gradually tested for KISS1R between January 2006 and April 2015. PARTICIPANTS/MATERIALS, SETTING, METHODS A total of 603 patients with nCHH (399 men and 204 women) were diagnosed at the Bicetre Hospital and underwent KISS1R analysis. The GNRHR, tachykinin receptor 3 (TACR3), gonadotrophin-releasing hormone 1 (GNRH1), tachykinin 3 (TAC3) and KISS1 genes were also sequenced. Functional characterization of KISS1R mutations included a study of signal transduction using a reporter gene (serum response element-luciferase (SRE-Luc) involved in the mitogen-activated protein (MAP) kinase pathway. MAIN RESULTS AND THE ROLE OF CHANCE We detected 15 KISS1R variants (10 novel), in 12 of the 603 patients (2.0%, 95% CI [0.9-3.1]. KISS1R mutations were less prevalent than GNRHR (4.7%) and TACR3 (2.6%) mutations but more prevalent than GNRH1 (1.5%), TAC3 (1.0%) and KISS1 (0%) mutations. KISS1R mutants were present in the biallelic state in 8 of the 12 patients concerned. Among 5 men with biallelic KISS1R mutations, 4 had either micropenis or cryptorchidism. In vitro analysis of the 5 new variants present in the biallelic state (C95W, Y103*, C115W, P176R and A287E) showed a loss of function. LIMITATIONS, REASONS FOR CAUTION The prevalence of TACR3, GNRH1, TAC3 and KISS1 mutations was calculated from a smaller number of nCHH patients than KISS1R and GNRHR. This should prompt caution concerning the reported prevalence of mutations in these four genes. WIDER IMPLICATIONS OF THE FINDINGS We show that KISS1R mutants are responsible for the nCHH phenotype in only a small minority of cases. Together, the genes analysed here were mutated in fewer than 15% of patients, suggesting a role of other genes in nCHH. The presence of cryptorchidism and/or micropenis in the majority of men with biallelic KISS1R mutations strongly suggests that this gene is essential for prenatal GnRH secretion. STUDY FUNDING, COMPETING INTEREST(S) This work was supported in part by grants from Paris-Sud University (Bonus Qualite Recherche, and Attractivite grants) to J.B., French Ministry of Health, Hospital Clinical Research Program on Rare Diseases. Assistance Publique Hopitaux de Paris, Programme Hospitalier de Recherche Clinique (PHRC # P081212 HYPOPROTEO) to J.Y. C.P. was supported by student fellowships 'Annee Recherche' from Agence Regionale de Sante Provence Alpes Cotes d'Azur. The authors have nothing to disclose.

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Institution

(Francou, Amazit, Bouvattier, Chanson, Trabado, Brailly-Tabard, Guiochon-Mantel, Young, Bouligand) Univ Paris-Sud, Assistance Publique-Hopitaux de Paris, Hopital Bicetre, Le Kremlin

Bicetre, France (Francou, Paul, Amazit, Chanson, Trabado, Brailly-Tabard, Guiochon-Mantel, Young, Bouligand) INSERM Unite Mixte de Recherche 1185, Assistance Publique-Hopitaux de Paris, Hopital Bicetre, Le Kremlin Bicetre F-94276, France

(Francou, Trabado, Brailly-Tabard, Guiochon-Mantel, Bouligand) Service de Genetique Moleculaire, Pharmacogenetique, et Hormonologie, Assistance Publique-Hopitaux de Paris, Hopital Bicetre, Le Kremlin Bicetre, France

(Cartes, Chanson, Young) Service d'Endocrinologie et des Maladies de la Reproduction, Centre de Reference des Maladies Endocriniennes Rares de la Croissance, Assistance Publique-Hopitaux de Paris, Hopital Bicetre, Le Kremlin Bicetre, France

(Bouvattier) Service d'Endocrinologie Pediatrique, Centre de Reference des Maladies du Developpement Sexuel, Assistance Publique-Hopitaux de Paris, Hopital Bicetre, Le Kremlin Bicetre, France

(Albarel, Brue) Service d'Endocrinologie Hopital de la Conception, Marseille, France

(Maiter) Departement d'Endocrinologie, Metabolisme et Nutrition, Universite Catholique de Louvain, Cliniques Universitaires, Saint-Luc, Belgium

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468.

Proximally Directed Double-looped Epididymis and Vas Deferens Simulating Vas Duplication: The Importance of Precise Definition of Abnormal Ductal Anatomy in Cryptorchidism.

Hester A.G., Kogan S.J.

Embase

Urology. 97 (pp 184-187), 2016. Date of Publication: 01 Nov 2016.

[Article]

AN: 613220222

Objective Epididymal and vasal abnormalities are frequently recognized with undescended testes. The most common defect identified is the extended or elongated epididymis, an anomaly in which the epididymal tail extends distally beyond the testis before looping back upon itself, then following its normal course. Although the impact on fertility has not been established, the

necessity of recognizing these vaso-epididymal abnormalities surgically is obvious as these frequently simulate a blind-ending spermatic cord leading to inadvertent excision or leaving the unseen testis more proximally in the abdomen. With this in mind, we describe a more complex vaso-epididymal structural abnormality with proximal extension of the epididymis and vas simulating duplication of these structures. Methods The varied surgical findings of a proximally directed double-looped abnormal epididymis and vas deferens associated with cryptorchid testes were identified in 15 children. In all instances, the epididymis extended proximally up the spermatic cord for at least 4 cm, simulating reduplication of the vas deferens and leading to bizarre ductal anatomy and confusing anatomical findings. Results In our practice, a number of abnormalities of the vaso-epididymal structures have been defined, but we isolated a number of situations in which identification of a proximally directed double-looped epididymis was noted during surgical exploration for cryptorchidism. Our review demonstrated several situations such as this where misidentification could have resulted in inadvertent transection of the vas deferens. Conclusion The need for recognition of these unusual ductal abnormalities associated with cryptorchid testes is re-emphasized by these extreme examples to prevent surgical mishaps at orchidopexy.

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Author NameID

Hester, Austin G.; ORCID: <http://orcid.org/0000-0003-1495-6487>

Institution

(Hester, Kogan) Department of Urology, Wake Forest University, Winston-Salem, NC, United States

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Volume alteration of undescended testes: Before and after orchiopexy.

Tseng C.-S., Huang K.-H., Pu Y.-S., Chiang I.-N.

Embase

Urological Science. 27 (3) (pp 161-165), 2016. Date of Publication: 01 Sep 2016.

[Article]

AN: 612946631

Objectives We used ultrasound to investigate the volume of undescended testes before and after orchiopexy, and compared these data with normally descended testes. **Materials and Methods** We retrospectively reviewed boys in the age range of 0-18 years who had undergone unilateral or bilateral orchiopexy due to undescended testes (International Classification of Diseases-Ninth Revision, ICD-9 752.51) in National Taiwan University Hospital, Taipei, Taiwan between January 2010 and December 2013. A total of 116 boys received preoperative testicular ultrasound evaluation, and 75 of them received regular ultrasound during a mean follow-up period of 2.5 years. The volume of the testes was calculated by applying Hansen formula [testicular volume = length (L) x width (W)² x 0.52] and compared with a cohort of 92 boys constructed for normative values of testicular volume from The Netherlands. **Results** The mean volume of the 145 undescended testes among 118 boys was 0.238 mL. The volume of the undescended testes was significantly smaller ($p < 0.001$) than the mean normative value of 0.418 mL. The volume of postorchiopexy undescended testes (0.356 mL) revealed a growing trend in the mean 2.5-year follow-up with a significance increase of size ($p = 0.001$), but has not yet reached the normal testicular size (0.604 mL). **Conclusion** The preorchiopexy volumes of undescended testes are significantly smaller than normative values. The follow-up postorchiopexy volumes of undescended testes actually increased in size, although they were still smaller than normative values. These Taiwanese testicular growth curves should become reference values in pediatric clinical practice when evaluating testicular development. **Keywords:** cryptorchidism, orchiopexy, testicular volume, treatment outcome, undescended testis

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Institution

(Tseng, Huang, Pu, Chiang) Department of Urology, National Taiwan University Hospital, National Taiwan University College of Medicine, Taipei, Taiwan (Republic of China)

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470.

Placental weight and male genital anomalies: A nationwide danish cohort study.

Arendt L.H., Ramlau-Hansen C.H., Wilcox A.J., Henriksen T.B., Olsen J., Lindhard M.S.

Embase

American Journal of Epidemiology. 183 (12) (pp 1122-1128), 2016. Date of Publication: 15 Jun 2016.

[Article]

AN: 612907346

The most consistently reported risk indicators for the male genital anomalies cryptorchidism and hypospadias are prematurity and low birth weight. Placental dysfunction has been hypothesized as a possible underlying cause, and an association between placental weight at birth and hypospadias has been indicated. In a population-based cohort of 388,422 Danish singleton boys born alive (1997-2008), we studied the association between placental weight and cryptorchidism and hypospadias. Missing data were handled with multiple imputation, and we estimated hazard ratios by means of Cox regression models. During follow-up, 1,713 boys were diagnosed with hypospadias and 6,878 with cryptorchidism (3,624 underwent corrective surgery). We observed an association between low placental weight and risk of both genital anomalies. Boys with a placental weight in the lowest decile (<10%) had higher risks of both cryptorchidism (hazard ratio = 1.52, 95% confidence interval: 1.31, 1.76) and hypospadias (hazard ratio = 1.97, 95% confidence interval: 1.59, 2.45) than boys in the reference decile (50.0-59.9%). In conclusion, we found higher risks of both genital malformations in boys born with a low placental weight. The relationship seemed stronger for hypospadias than for cryptorchidism. Taken together, our data support a role for placental dysfunction in the etiology of these anomalies.

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Institution

(Arendt, Ramlau-Hansen, Wilcox, Henriksen, Olsen, Lindhard) Section for Epidemiology,
Department of Public Health, Faculty of Health, Aarhus University, Bartholins Alle 2, Aarhus C
8000, Denmark

Publisher

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471.

Fetal growth restriction but not preterm birth is a risk factor for severe hypospadias.

Hashimoto Y., Kawai M., Nagai S., Matsukura T., Niwa F., Hasegawa T., Heike T.

Embase

Pediatrics International. 58 (7) (pp 573-577), 2016. Date of Publication: 01 Jul 2016.

[Article]

AN: 611437845

Background: Hypospadias has multifactorial causes and occurs at a high frequency among very low-birthweight infants. Placental insufficiency is hypothesized to be one cause of hypospadias; that is, decreased human chorionic gonadotropin (hCG) secretion caused by placental insufficiency is suspected to result in abnormal male external genitalia, but there is little direct evidence to support this. The aim of this study was therefore to identify the features of hypospadias and to clarify the male genital abnormalities caused by fetal growth restriction (FGR).

Method(s): We reviewed the clinical data of boys who underwent hypospadias repair between 2005 and 2011 at Kyoto University Hospital.

Result(s): Twenty boys were included in this study. Fifteen (75%) of the subjects were preterm or low-birthweight infants. Thirteen (65%) had FGR, 60% of whom had severe hypospadias regardless of gestational age. In addition, 92% of the FGR infants also had other genital anomalies, such as cryptorchidism, bifid scrotum, or micropenis. In contrast, only 14% and 43% of the non-FGR infants had severe hypospadias or genital anomalies other than hypospadias, respectively. Placental histopathology was available in eight FGR infants, in seven of whom it was suggestive of blood flow deficiency such as infarction and single umbilical artery.

Conclusion(s): Infants with FGR have a high incidence of hypospadias. FGR caused by placental dysfunction, but not low birthweight, is a risk factor for severe hypospadias associated with multiple genital anomalies.

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Institution

(Hashimoto) Department of Pediatric Endocrinology and Metabolism, Children's Medical Center, Osaka City General Hospital, Osaka, Japan (Hashimoto, Kawai, Nagai, Matsukura, Niwa, Hasegawa, Heike) Department of Pediatrics, Graduate School of Medicine, Kyoto University, Kyoto, Japan

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472.

Anogenital distance as a marker of androgen exposure in humans.

Thankamony A., Pasterski V., Ong K.K., Acerini C.L., Hughes I.A.

Embase

Andrology. 4 (4) (pp 616-625), 2016. Date of Publication: 01 Jul 2016.

[Review]

AN: 611401518

Abnormal foetal testis development has been proposed to underlie common disorders of the male reproductive system such as cryptorchidism, hypospadias, reduced semen quality and testicular germ cell tumour, which are regarded as components of a 'testicular dysgenesis syndrome'. The increasing trends and geographical variation in their incidence have been suggested to result from in utero exposure to environmental chemicals acting as endocrine disruptors. In rodents, the anogenital distance (AGD), measured from the anus to the base of genital tubercle, is a sensitive biomarker of androgen exposure during a critical embryonic window of testis development. In

humans, several epidemiological studies have shown alterations in AGD associated with prenatal exposure to several chemicals with potential endocrine disrupting activity. However, the link between AGD and androgen exposure in humans is not well-defined. This review focuses on the current evidence for such a relationship. As in rodents, a clear gender difference is detected during foetal development of the AGD in humans which is maintained thereafter. Reduced AGD in association with clinically relevant outcomes of potential environmental exposures, such as cryptorchidism or hypospadias, is in keeping with AGD as a marker of foetal testicular function. Furthermore, AGD may reflect variations in prenatal androgen exposure in healthy children as shorter AGD at birth is associated with reduced masculine play behaviour in preschool boys. Several studies provide evidence linking shorter AGD with lower fertility, semen quality and testosterone levels in selected groups of adults attending andrology clinics. Overall, the observational data in humans are consistent with experimental studies in animals and support the use of AGD as a biomarker of foetal androgen exposure. Future studies evaluating AGD in relation to reproductive hormones in both infants and adults, and to gene polymorphisms, will help to further delineate the effect of prenatal and postnatal androgen exposures on AGD.

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Institution

(Thankamony, Pasterski, Acerini, Hughes) Department of Paediatrics, Addenbrooke's Hospital, University of Cambridge, Cambridge, United Kingdom (Thankamony) Department of Growth and Reproduction, Rigshospitalet, University of Copenhagen, Denmark

(Pasterski) Department of Psychology, University of Cambridge, Cambridge, United Kingdom

(Ong) MRC Epidemiology Unit, Institute of Metabolic Science, University of Cambridge, Cambridge, United Kingdom

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Polymorphism rs2274911 of GPRC6A as a novel risk factor for testis failure.

De Toni L., Di Nisio A., Speltra E., Rocca M.S., Ghezzi M., Zuccarello D., Turiaco N., Ferlin A., Foresta C.

Embase

Journal of Clinical Endocrinology and Metabolism. 101 (3) (pp 953-961), 2016. Date of Publication: March 2016.

[Article]

AN: 609266697

Context: The G protein-coupled receptor GPRC6A is an emerging effector with multiple endocrine roles, including stimulation of T production from the testis. Recently, two men with an inactivating mutation (F464Y) of GPRC6A have been identified, and they showed primary testicular failure and deranged spermatogenesis. Furthermore, one of them also reported cryptorchidism at birth. In addition, a polymorphism (rs2274911, Pro91Ser) in GPRC6A is associated with prostate cancer, a typical androgen-sensitive cancer.

Objective(s): To study the possible association between rs2274911 polymorphism and male fertility and/or cryptorchidism. Design, Patients, Settings: A total of 611 subjects, including 343 infertile patients, 197 normozoospermic controls, and 71 cryptorchid newborns, were retrospectively selected.

Method(s): Sequencing analysis for rs2274911 polymorphism and F464Y mutation, and serum levels of FSH, LH, and T were assessed. In vitro functional studies for rs2274911 and F464Y were also performed.

Result(s): Homozygous subjects for the risk allele A of rs2274911 had a 4.60-fold increased risk of oligozoospermia and 3.52-fold increased risk of cryptorchidism. A significant trend for increased levels of LH in the GA and AA genotypes, compared with GG homozygotes, was detected in men with azoospermia/cryozoospermia (P for trend .027), further supporting an association with primary testicular failure. The mutation F464Y was found in one cryptorchid child (one in 71; 1.41%). Functional studies showed that the A allele of rs2274911 and the F464Y substitution were associated with lower exposition of the receptor on the cell membrane and a reduced downstream phosphorylation of ERK1/2 with respect to wild type.

Conclusion(s): Our results suggest that GPRC6A inactivation or sub-function contributes to reduced exposure to androgens, leading to cryptorchidism during fetal life and/or low sperm production in adulthood.

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PMC Identifier

26735260 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=26735260>]

Status

Embase

Institution

(De Toni, Di Nisio, Speltra, Rocca, Ghezzi, Ferlin, Foresta) Department of Medicine, Unit of Andrology, Reproductive Medicine, University of Padova, Via Giustiniani, 2, Padova 35128, Italy
(Zuccarello) Clinical Genetics Unit, Department of Woman and Child Health, University of Padova, Padova 35128, Italy

(Turiaco) Department of Paediatric, Gynaecological, Microbiological, and Biomedical Sciences, University of Messina, Messina 98124, Italy

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Endocrine Society (E-mail: jnl.info@oup.co.uk)

Year of Publication

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474.

Undescended testes: An update.

Holland A.J.A., Nassar N., Schneuer F.J.

Embase

Current Opinion in Pediatrics. 28 (3) (pp 388-394), 2016. Date of Publication: 01 Jun 2016.

[Review]

AN: 610317968

Purpose of review This article summarizes the latest evidence on the risk factors, management and outcomes of undescended testes (UDTs). Recent findings UDTs remain common, with increasing evidence that acquired UDT or the ascending testis syndrome should be considered part of the spectrum of this disease. Prompt diagnosis and early referral for surgical evaluation and treatment would seem most likely to result in an optimal functional and cosmetic outcome. Hormonal treatment, rather than orchidopexy, remains popular in some centers, despite a lack of good evidence to support its efficacy, although it may have an important adjunct role in optimizing fertility. Although often performed, ultrasound does not generally assist in the diagnosis and management of UDT, with enhanced education of primary care physicians more likely to facilitate early referral. The testis, rather than quiescent, appears biologically active in the male infant, with increasing evidence of an adverse impact on future spermatogenesis and fertility in men with a UDT. **Summary** Male infants with a UDT should be diagnosed and referred early for surgical

evaluation. It seems likely that the optimal timing for surgery should be before the boy's first birthday. There remains a need for high-quality, long-term outcomes data to guide optimal management.

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Status

In-Process

Institution

(Holland) Douglas Cohen Department of Paediatric Surgery, Discipline of Paediatrics and Child Health, Children's Hospital at Westmead, University of Sydney, Sydney, NSW, Australia
(Nassar, Schneuer) Menzies Centre for Health Policy, School of Public Health, University of Sydney, Sydney, NSW, Australia

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

Year of Publication

2016

475.

Familial forms of disorders of sex development may be common if infertility is considered a comorbidity.

Brauner R., Picard-Dieval F., Lottmann H., Rouget S., Bignon-Topalovic J., Bashamboo A., McElreavey K.

Embase

BMC Pediatrics. 16 (1) (no pagination), 2016. Article Number: 195. Date of Publication: 29 Nov 2016.

[Article]

AN: 613438649

Background: Families with 46,XY Disorders of Sex Development (DSD) have been reported, but they are considered to be exceptionally rare, with the exception of the familial forms of disorders affecting androgen synthesis or action. The families of some patients with anorchia may include individuals with 46,XY gonadal dysgenesis. We therefore analysed a large series of patients with

46,XY DSD or anorchia for the occurrence in their family of one of these phenotypes and/or ovarian insufficiency and/or infertility and/or cryptorchidism.

Method(s): A retrospective study chart review was performed for 114 patients with 46,XY DSD and 26 patients with 46,XY bilateral anorchia examined at a single institution over a 33 year period.

Result(s): Of the 140 patients, 25 probands with DSD belonged to 21 families and 7 with anorchia belonged to 7 families. Familial forms represent 22% (25/114) of the 46,XY DSD and 27% (7/26) of the anorchia cases. No case had disorders affecting androgen synthesis or action or 5 alpha-reductase deficiency. The presenting symptom was genital ambiguity (n = 12), hypospadias (n = 11) or discordance between 46,XY karyotyping performed in utero to exclude trisomy and female external genitalia (n = 2) or anorchia (n = 7). Other familial affected individuals presented with DSD and/or premature menopause (4 families) or male infertility (4 families) and/or cryptorchidism. In four families mutations were identified in the genes SRY, NR5A1, GATA4 and FOG2/ZFPM2. Surgery discovered dysgerminoma or gonadoblastoma in two cases with gonadal dysgenesis.

Conclusion(s): This study reveals a surprisingly high frequency of familial forms of 46,XY DSD and anorchia when premature menopause or male factor infertility are included. It also demonstrates the variability of the expression of the phenotype within the families. It highlights the need to the physician to take a full family history including fertility status. This could be important to identify familial cases, understand modes of transmission of the phenotype and eventually understand the genetic factors that are involved.

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Embase

Institution

(Brauner, Picard-Dieval, Rouget) Fondation Ophtalmologique Adolphe de Rothschild and
Universite Paris Descartes, Paris, France (Lottmann) Assistance Publique-Hopitaux de Paris,
Hopital Necker-Enfants Malades, Service de chirurgie viscerale pediatrique, Paris, France
(Bignon-Topalovic, Bashamboo, McElreavey) Human Developmental Genetics, Institut Pasteur,
Paris, France

Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2016

476.

Management of undescended testes: European Association of Urology/European Society for Paediatric Urology Guidelines.

Radmayr C., Dogan H.S., Hoebeke P., Kocvara R., Nijman R., Stein R., Undre S., Tekgul S.

Embase

Journal of Pediatric Urology. 12 (6) (pp 335-343), 2016. Date of Publication: 01 Dec 2016.

[Review]

AN: 613309287

Context Undescended testis is the most common endocrinological disease in the male newborn period. Incidence varies between 1.0% and 4.6% in full-term neonates, with rates as high as 45% in preterm neonates. Failure or delay of treatment can result in reduced fertility and/or increased testicular cancer risk in adulthood. Objective To provide recommendations for the diagnosis and treatment of boys with undescended testes which reduce the risk of impaired fertility and testicular cancer in adulthood. Evidence acquisition Embase and Pubmed were searched for all relevant publications, from 1990 to 2015 limited to English language. Data were narratively synthesized in light of methodological and clinical heterogeneity. The risk of bias of each included study was assessed. Evidence synthesis There is consensus that early treatment, by 18 months at the latest, for undescended testes is mandatory to avoid possible sequelae regarding fertility potential and cancer risk. The current standard therapy is orchidopexy, while hormonal therapy is still under debate. However, in some individuals the successful scrotal placement of previously undescended testes may not prevent potential negative long-term outcomes regarding fertility and testicular malignancy. Conclusions There is good evidence for early placement of undescended testes in the scrotal position to prevent potential impairment of fertility and reduce the risk of testicular malignancy. No consensus exists on the various forms of hormonal treatment, which are assessed on an individual basis.

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PMC Identifier

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Status

Embase

Institution

(Radmayr) Paediatric Urology, Medical University of Innsbruck, Innsbruck, Austria (Dogan, Tekgul) Hacettepe University, Faculty of Medicine, Department of Urology, Division of Paediatric Urology, Ankara, Turkey

(Hoebeke) Department of Urology, Ghent University Hospital, Gent, Belgium

(Kocvara) Department of Urology, General Teaching Hospital in Praha, Charles University 1st Faculty of Medicine, Praha, Czechia

(Nijman) Department of Urology, Division of Paediatric Urology, University of Groningen, Groningen, Netherlands

(Stein) Division of Paediatric Urology, Department of Urology, Mainz University Medical Centre, Johannes Gutenberg University, Mainz, Germany

(Undre) Department of Paediatric Urology, Great Ormond Street Hospital for Sick Children, London, United Kingdom

Publisher

Elsevier Ltd

Year of Publication

2016

477.

Maternal and fetal risk factors for bladder exstrophy: A nationwide Swedish case-control study. Reinfeldt Engberg G., Mantel A., Fossum M., Nordenskjold A.

Embase

Journal of Pediatric Urology. 12 (5) (pp 304.e1-304.e7), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 613135647

Introduction Bladder exstrophy is a rare, congenital, complex malformation where the underlying cause is largely unknown. Both environmental and genetic mechanisms are thought to be involved. There are divergent results concerning the prevalence, birth descriptive data, and potential maternal risk factors for bladder exstrophy. Few previous studies have reflected nationwide populations, population registers, or spanned a longer period of time. Objective To describe and assess bladder exstrophy and the potential maternal risk factors, for a time period of four decades, by conducting a nationwide register study of bladder exstrophy in Sweden.

Methods A matched-design, case-control, linkage-analysis study nested within the entire pool of

live births in Sweden between 1973 and 2011 was performed. Cases with bladder exstrophy were identified using nationwide population-based birth and health registers. Inclusion criteria were people born in Sweden with the classification of bladder exstrophy according to the ICD coding system. Cases were matched with five controls per patient, based on birth year and sex. Prevalence was assessed and birth descriptive data were compiled. Potential maternal risk factors were obtained from medical birth registers of cases and assessed using conditional and multivariate logistic regression models to obtain odds ratios as a measure of the relative risk. Classification of the diagnosis in the registers constituted a possible limitation for determining the correct study population, which demanded strict validation and inclusion criteria. All data were collected prospectively, thereby avoiding potential recall bias. Results The prevalence was calculated to be approximately 3 per 100,000 live births, with a male-to-female ratio of 1.14:1. In 92.5% of the cases, bladder exstrophy was an isolated malformation without associated major malformations. However, 41% had had surgery for congenital inguinal hernia and 11% of the male subjects had been operated on for cryptorchidism. A significantly higher proportion of cases had a birth weight <1500 g compared with controls, but other characteristics were comparable with controls. High maternal age was the only significant potential associated maternal risk factor. Conclusions One hundred and twenty children born with bladder exstrophy in Sweden during the last four decades were identified; this resulted in prevalence in Sweden of 3 per 100,000. The prevalence was stable over time and the sex ratio was equal. Birth characteristics were comparable to controls, and bladder exstrophy generally occurred as an isolated malformation without major associated malformations. Advanced maternal age was the only significant potential maternal risk factor.

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PMC Identifier

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Status

Embase

Institution

(Reinfeldt Engberg, Fossum, Nordenskjold) Department of Women's and Children's Health and Center for Molecular Medicine, Karolinska Institutet, Stockholm, Sweden (Reinfeldt Engberg, Fossum, Nordenskjold) Department of Pediatric Surgery, Section of Urology, Astrid Lindgren Children's Hospital, Karolinska University Hospital, Stockholm, Sweden (Mantel) Clinical Epidemiology Unit, Department of Medicine, Solna (MedS), K2, Karolinska Institutet, Stockholm, Sweden

Publisher

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478.

Undescended testis? How best to teach the physical examination.

Zundel S., Blumenstock G., Herrmann-Werner A., Trueck M., Schmidt A., Wiechers S.

Embase

Journal of Pediatric Urology. 12 (6) (pp 406.e1-406.e6), 2016. Date of Publication: 01 Dec 2016.

[Article]

AN: 613089370

Background Undescended testis in boys is common. Guidelines recommend surgical treatment between the ages of 6 months and 2 years; nevertheless, orchidopexy is frequently performed at later ages. One reason is the belated diagnosis due to a perceived difficulty in the physical examination (PE) and correct localization of the testis. **Objectives** We aimed to find an effective method for teaching the physical examination of the testis in a child. **Study design** An interdisciplinary team developed teaching sessions, including an educational video and a simulator. Medical students (n = 133) were randomized into three groups: self-study only, video, and video and simulator. The sessions were carried out and quantitative feedback was collected from the teachers and students. The learning achievements of the different groups were assessed with an objective structured clinical examination (OSCE). The differences in mean OSCE results between all three groups were tested using one-way analysis of variance (ANOVA). For multiple pairwise comparisons, a closed testing procedure was performed using unpaired t-tests. **Results** The self-study only group acquired the poorest results in the OSCE, with a mean score of 5.1 out of 10. The video-only-group reached a mean of 6.7, and the video-and-simulator group performed best with a mean score of 8.5. The differences between all three groups were found to be statistically significant, with $P = 0.007$. The attached figure illustrates this data. If analyzed in pairs, this difference was particularly apparent between the groups self-study only vs video and simulator, with $P = 0.002$. Qualitative feedback revealed doubtful effectiveness for educational videos, but positive reactions to training on a simulator. **Discussion** The poor results of the self-study-only group were in accordance with the literature, where textbook learning was found not to increase OSCE results. The effectiveness of video tutorials remains doubtful; studies focusing on this teaching method are divergent and the present students' feedback supports this data. The effective teaching with the simulator has been proven for other

skills (i.e. ultrasound skills). The analyzed cohort for this study was small, and the study should be repeated at different institutions and with larger numbers of students to assure generalizability.

Conclusions Low-fidelity pediatric simulators with palpable testis are available and are able to improve examining skills in medical students. We hope the presented study inspires medical educators in their teaching of the PE of the pediatric testis.[Figure presented]

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Embase

Institution

(Zundel, Trueck, Schmidt) Department of Pediatric Surgery, University Hospital of Tuebingen, Tuebingen, Germany (Blumenstock) Department of Medical Biometry, University of Tuebingen, Tubingen, Germany

(Herrmann-Werner) Department of Psychosomatic Medicine and Medical Faculty, University Hospital of Tuebingen, Multidisciplinary Skills Lab "DocLab", Tubingen, Germany

(Wiechers) Department of Pediatric Hematology, Oncology and General Pediatrics, University Hospital of Tuebingen, Tuebingen, Germany

Publisher

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2016

479.

Timing of orchiopexy for undescended testis in Israel: A quality of care study.

Hidas G., Chaim J.B., Udassin R., Graeb M., Gofrit O.N., Zisk-Rony R.Y., Pode D., Duvdevani M. M., Yutkin V., Neheman A., Fruman A., Arbel D., Kopuler V., Armon Y., Landau E.H.

Embase

Israel Medical Association Journal. 18 (11) (pp 697-700), 2016. Date of Publication: November 2016.

[Article]

AN: 613282679

Background: Strong evidence suggests that in order to prevent irreversible testicular damage surgical correction (orchiopexy) for undescended testis (UDT) should be performed before the age of 1 year.

Objective(s): To evaluate whether orchiopexy is delayed in our medical system, and if so, to explore the pattern of referral for orchiopexy as a possible contributing factor in such delays.

Method(s): We conducted a retrospective chart review of all children who underwent orchiopexy for UDT between 2003 and 2013 in our institution. We collected data on the age at surgery and the child's health insurance plan. We also surveyed pediatricians from around the country regarding their pattern of UDT patient referral to a pediatric urologist or surgeon for surgical correction.

Result(s): A total of 813 children underwent orchiopexy in our institute during the study period. The median age at surgery was 1.49 years (range 0.5-13). Only 11% of the children underwent surgery under the age of 1 year, and 53% between the ages of 1 and 2 years. These findings were consistent throughout the years, with no difference between the four health insurance plans. Sixty-three pediatricians who participated in the survey reported that they referred children to surgery at a median age of 1 year (range 0.5-3 years).

Conclusion(s): Our results demonstrate delayed orchiopexy in our medical system. There is a need to improve awareness for early specialist consultation in order to facilitate earlier surgery and better care.

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PMC Identifier

28466623 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28466623>]

Status

Embase

Institution

(Hidas, Chaim, Graeb, Neheman, Landau) Pediatric Urology Unit, Hadassah-Hebrew University Medical Center, Jerusalem, Israel (Gofrit, Pode, Duvdevani M., Yutkin) Department of Urology, Hadassah-Hebrew University Medical Center, Jerusalem, Israel

(Udassin, Duvdevani M., Fruman, Arbel, Kopuler, Armon) Department of Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel

(Zisk-Rony) School of Nursing, Hadassah-Hebrew University Medical Center, Jerusalem, Israel

Publisher

Israel Medical Association (2 Twin Towers, 11th Floor, 35 Jabotinsky Street, PO Box 3566, Ramat Gan 52135, Israel)

Year of Publication

2016

480.

Prader-Willi syndrome in neonates: Twenty cases and review of the literature in Southern China.
Wang P., Zhou W., Yuan W., Huang L., Zhao N., Chen X.

Embase

BMC Pediatrics. 16 (1) (no pagination), 2016. Article Number: 124. Date of Publication: 09 Aug 2016.

[Article]

AN: 611586183

Background: Prader-Willi syndrome is a rare genetic abnormality that can be challenging to diagnose early, but for which early interventions improve prognosis.

Method(s): To improve understanding of Prader-Willi syndrome in neonates in Asia, we retrospectively analyzed the clinical records of 20 affected newborns diagnosed in the Department of Neonatology, Guangzhou Women and Children's Medical Center, Guangzhou, China from January 2007 to December 2014 and performed a review of the relevant literature.

Result(s): Fourteen boys and six girls presented with hypotonia, poor responsiveness, feeding difficulty, and infrequent, weak crying. Different from western patients, the 20 Asian patients exhibited at least five of the following typical features: prominent forehead, narrow face, almond-shaped eyes, small mouth, downturned mouth, thin upper lip, and micromandible. All 14 boys had a small scrotum, including nine with cryptorchidism. Diagnoses were made with microarray comparative genomic hybridization. All 20 infants required feeding tubes. Fifteen received swallowing training immediately after admission; the period of continuous tube feeding for these patients ranged from 8 to 22 days (mean, 14 +/- 5.3 days). For the five patients who did not receive swallowing training, the period of continuous tube feeding ranged from 15 to 35 days (mean, 18 +/- 4.3 days). Comprehensive care measures included: giving parents detailed health education and basic information about this disease, teaching skills to promote feeding and prevent suffocation, increasing children's passive activity, providing nutrition management for normal development, and preventing excessive or inadequate nutrient intake.

Conclusion(s): Neonates with Prader-Willi syndrome in Asia have hypotonia, poor responsiveness, feeding difficulty, infrequent and weak crying, genital hypoplasia, and characteristic facial features. Recognition of the syndrome in neonates with confirmation by genetic testing is essential, because early diagnosis allows early intervention. Treatment

measures including swallowing training can improve prognosis, prevent growth retardation and obesity, and elevate quality of life in individuals with Prader-Willi syndrome.

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27506196 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27506196>]

Status

Embase

Institution

(Wang) The First Affiliated Hospital of Jinan University, No 613, Huangpuda Road, Guangzhou, Guangdong 510623, China (Wang, Zhou, Yuan, Huang, Zhao, Chen) Department of Neonatology, Guangzhou Women and Children's Medical Center, No 9, Jinsui Road, Guangzhou, Guangdong 510623, China

Publisher

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Year of Publication

2016

481.

International Consultation on Urological Diseases: Congenital Anomalies of the Genitalia in Adolescence.

Higuchi T., Holmdahl G., Kaefer M., Koyle M., Wood H., Woodhouse C., Wood D.

Embase

Urology. 94 (pp 288-310), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 612748615

Objective To provide a comprehensive overview of genital anomalies encountered among adolescents, including late effects of problems addressed earlier in childhood. Materials and Methods The major congenital genital anomalies encountered in pediatric urology were identified. They include hypospadias, exstrophy-epispadias, cloacal malformations, disorders of sexual development, undescended testes, and some acquired penile anomalies seen in adolescence (priapism, adolescent varicocele). Recommendations of the International Consultation on Urological Diseases are provided on various aspects of these conditions, such as postpubertal

cosmesis and function, fertility implications, and long-term nephrological considerations (when relevant). Results Specific recommendations for care, including strength of clinical recommendation, are provided in this paper. Whereas the basis of this paper is to discuss specific management recommendations as they relate to several heterogeneous conditions, general recommendations include patient-centered discussions regarding operative treatment be deferred until the patient is able to articulate goals and participate in shared decision-making and utilization of multidisciplinary teams for conditions where multiple organ systems may be involved. Conclusion Congenital abnormalities of the genitalia are common and widely heterogeneous. Late effects and concerns often emerge after puberty, and patients should be followed throughout their adult lives to address such concerns.

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Status

Embase

Institution

(Higuchi) University of Colorado, Aurora, CO, United States (Holmdahl) Department of Pediatric Surgery/Urology, The Queen Silvia Children's Hospital, Gothenburg, Sweden

(Kaefer) Riley Hospital for Children, Indianapolis, IN, United States

(Koyle) Hospital for Sick Children, Toronto, Ontario, Canada

(Wood) Cleveland Clinic, Cleveland, OH, United States

(Woodhouse) University College, London, United Kingdom

(Wood) University College London Hospitals, London, United Kingdom

Publisher

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Year of Publication

2016

482.

Laparoscopic orchiopexy: Short-term outcomes. Experience of a single centre.

Pederiva F., Guida E., Codrich D., Scarpa M.G., Olenik D., Schleef J.

Embase

Minerva Pediatrica. 68 (4) (pp 278-281), 2016. Date of Publication: August 2016.

[Article]

AN: 610835943

Background: Increased infertility and smaller volume accompany undescended testis. Timing of orchiopexy is still a matter of debate. We evaluated the growth of non-palpable testes after laparoscopic orchiopexy according to age at surgery, intraoperative findings and type of procedure.

METHOD(S): Forty-one boys undergoing laparoscopy for nonpalpable testes were retrospectively reviewed and divided into two groups, ≤ 18 months and < 18 months, according to their age at surgery.

RESULT(S): At follow-up, 14 testes in the younger group had normal size, while 3 atrophied either after single (2) or two stage procedure (1). Similarly, in older boys 11 testes grew normally, while 5 atrophied after both procedures.

CONCLUSION(S): Most of the non-palpable testes grew normally after laparoscopic orchiopexy and the postoperative volume seemed independent from the surgical strategy. Both techniques led to a few cases of testicular hypotrophy. In our experience, the age at surgery did not affect the outcome in terms of testicular growth.

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Institution

(Pederiva, Guida, Codrich, Scarpa, Olenik, Schleef) Pediatric Surgery, Institute for Maternal and Child Health, IRCCS Burlo Garofolo, Via dell'Istria 65/1, Trieste 34137, Italy

Publisher

Edizioni Minerva Medica (E-mail: subscriptions.dept@minervamedica.it)

Year of Publication

2016

483.

Congenital hypogonadotropic hypogonadism: Implications of absent mini-puberty.

Dwyer A.A., Jayasena C.N., Quinton R.

Embase

Minerva Endocrinologica. 41 (2) (pp 188-195), 2016. Date of Publication: June 2016.

[Review]

AN: 612971382

The phenomenon known as "mini-puberty" refers to activation of the neonatal hypothalamo-pituitary axis causing serum concentrations of gonadotrophins and testosterone (T) to approach adult male levels. This early neonatal period is a key proliferative window for testicular germ cells and immature Sertoli cells. Although failure to spontaneously initiate (adolescent) puberty is the most evident consequence of a defective gonadotropin-releasing hormone (GnRH) neurosecretory network, absent mini-puberty is also likely to have a major impact on the reproductive phenotype of men with congenital hypogonadotropic hypogonadism (CHH). Furthermore, the phase of male mini-puberty represents a key window-ofopportunity to identify congenital GnRH deficiency (either isolated CHH, or as part of combined pituitary hormone deficiency) in childhood. Among male neonates exhibiting "red flag" indicators for CHH (i.e. maldescended testes with or without cryptorchidism) a single serum sample (between 4-8 weeks of life) can pinpoint congenital GnRH deficiency far more rapidly and with much greater accuracy than dynamic tests performed in later childhood or adolescence. Potential consequences for missing absent mini-puberty in a male neonate include the lack of monitoring of pubertal progression/lack of progression, and the missed opportunity for early therapeutic intervention. This article will review our current understanding of the mechanisms and clinical consequences of mini-puberty. Furthermore, evidence for the optimal clinical management of patients with absent mini-puberty will be discussed.

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Status

Embase

Institution

(Dwyer) Endocrinology, Diabetes and Metabolism Service, Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland (Dwyer) University of Lausanne, Institute of Higher Education and Research in Healthcare, Lausanne, Switzerland

(Jayasena) Section of Investigative Medicine, Imperial College London, Faculty of Medicine, Hammersmith Hospital, London, United Kingdom

(Quinton) Endocrine Research Group, Institute of Genetic Medicine, Newcastle University, Newcastleupon-Tyne, United Kingdom

(Quinton) Endocrine Unit, Royal Victoria Infirmary, Newcastle-upon-Tyne Hospitals, Newcastle-upon-Tyne NE1 4LP, United Kingdom

Publisher

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Year of Publication

2016

484.

Congenital bilateral Anorchia: A study of 5 cases in Jordan.

Bustanji H., Khawaja N., Ajlouni K.

Embase

Jordan Medical Journal. 50 (1) (pp 39-47), 2016. Date of Publication: 2016.

[Article]

AN: 609433141

We report the clinical and hormonal findings in 5 cases of bilateral anorchism. Five male patients aged 3-5 years presented with suspected cryptorchidism. Physical examination, hormonal, imaging, chromosomal, and molecular analyses of these cases were performed. Ultrasonography of the pelvis and magnetic resonance of the abdomen were performed and failed to show any true testicular tissue or showed only atrophied suspicious testicular tissue. Chromosomal analysis revealed a normal male karyotype and molecular analysis did not reveal mutations or polymorphisms in the SRY gene. The basal FSH and LH levels were increased, and there were increase in response to gonadotropinreleasing hormone test, testosterone levels failed to increase after hCG administration. Lastly, surgical exploration confirmed the absence of testicular structure in three of them. Diagnostically, the very low anti Mullerian hormone level combined with the lack of testosterone response to hCG are the hormonal hallmarks of bilateral congenital anorchia.

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Status

Embase

Institution

(Bustanji, Khawaja, Ajlouni) National Center for Diabetes Endocrinology and Genetics, P.O. Box 13165, Amman 11942, Jordan

Publisher

University of Jordan (E-mail: imansour@ju.edu.jo)

Year of Publication

2016

485.

Complications of inguinal herniotomy are comparable in term and premature infants.

Hughes K., Horwood J.F., Clements C., Leyland D., Corbett H.J.

Embase

Hernia. 20 (4) (pp 565-569), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 607647548

Purpose: Inguinal hernias are common, and prevalence is highest in premature males.

Recognised complications include recurrence, iatrogenic cryptorchidism and testicular atrophy.

We reviewed complication rates following inguinal herniotomy (IH), comparing premature (gestation <36 weeks) and term infants.

Method(s): A retrospective case note review of infants aged 0-12 months undergoing IH between January 2006 and December 2010. Data collected included demographics, side of hernia, incarceration, complications, duration of follow-up and need for further surgery. Comparison was made using unpaired student t test and Fishers exact test.

Result(s): Four hundred and eight patients underwent IH (365 male, 42 female, 1 complete androgen insensitivity); 197 were premature (prem), mean weight 3.81 kg (1.02-9.4); 211 were term (gestation \geq 36 weeks), mean weight 5.85 kg (2.4-11.7), $p = 0.0001$ versus prem. Total herniotomies performed = 472 (131 Left, 213 Right, 64 bilateral); 89 hernias were incarcerated at presentation (60 prem vs. 29 term, $p = 0.0001$). Bowel resection was required in six patients (5 prem vs. 1 term, $p = 0.1109$), and 14 had simultaneous orchidopexy (12 prem vs. 2 term, $p = 0.0049$). Early post-operative complication rate 2.8 % (8 prem vs. 5 term; $p = 0.4037$). Two hundred and forty patients attended follow-up; 58.8 % (125 prem, 115 term). This group accounted for 279 IH (264 male, 15 female). Mean follow-up = 5 months (0.5-36). Complication rates: recurrence 2.7 %, metachronous hernia 7.5 %, iatrogenic cryptorchidism 3.8 % and testicular atrophy 0.7 %.

Conclusion(s): No significant difference was found in complication rates between premature and term infants despite significantly more premature infants presenting with incarcerated hernias.

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Embase

Institution

(Hughes, Horwood, Clements, Leyland, Corbett) Department of Paediatric Surgery, Alder Hey Children's Hospital NHS Foundation Trust, Liverpool L12 2AP, United Kingdom

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486.

Disparities in the context of opportunities for cancer prevention in early life.

Masseti G.M., Thomas C.C., Ragan K.R.

Embase

Pediatrics. 138 (Supplement 1) (pp S65-S77), 2016. Date of Publication: November 2016.

[Article]

AN: 613505269

Persistent health disparities are a major contributor to disproportionate burden of cancer for some populations. Health disparities in cancer incidence and mortality may reflect differences in exposures to risk factors early in life. Understanding the distribution of exposures to early life risk and protective factors for cancer across different populations can shed light on opportunities to promote health equity at earlier developmental stages. Disparities may differentially influence risk for cancer during early life and create opportunities to promote health equity. Potential risk and protective factors for cancer in early life reveal patterns of disparities in their exposure. These disparities in exposures can manifest in downstream disparities in risk for cancer. These risk and protective factors include adverse childhood experiences; maternal alcohol consumption in

pregnancy; childhood obesity; high or low birth weight; benzene exposure; use of assisted reproductive technologies; pesticide and insecticide exposure; isolated cryptorchidism; early pubertal timing; exposure to radiation; exposure to tobacco in utero and in early life; allergies, asthma, and atopy; and early exposure to infection. Disparities on the basis of racial and ethnic minority status, economic disadvantage, disability status, sex, geography, and nation of origin can occur in these risk and protective factors. Vulnerable populations experience disproportionately greater exposure to risk factors in early life. Addressing disparities in risk factors in early life can advance opportunities for prevention, promote health equity, and possibly reduce risk for subsequent development of cancer.

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Embase

Institution

(Thomas, Ragan) Division of Cancer Prevention and Control, 4770 Buford Hwy NE, MS K76, Atlanta, GA 30341, United States (Masseti) Division of Cancer Prevention and Control, Centers for Disease Control and Prevention, Atlanta, United States

Publisher

American Academy of Pediatrics (141 Northwest Point Blvd, P.O. Box 927, Elk Grove Village IL 60007-1098, United States)

Year of Publication

2016

487.

Pediatric inguinal and scrotal surgery - Practice patterns in U.S. academic centers.

Chan Y.Y., Durbin-Johnson B., Kurzrock E.A.

Embase

Journal of Pediatric Surgery. 51 (11) (pp 1786-1790), 2016. Date of Publication: 01 Nov 2016.

[Article]

AN: 612796427

Purpose Both pediatric urologists and pediatric surgeons perform hernia repairs, hydrocelectomies and orchiopexies. We hypothesized that surgeons perform more incarcerated and female hernia repairs while urologists perform more orchiopexies and hydrocelectomies.

Methods The Vizient-AAMC Faculty Practice Solutions Center database was queried from January 2009 to December 2014 to identify patients 10 years or younger who underwent the above procedures performed by pediatric specialists. Age, gender, race, insurance, geographic region and surgeon volume were examined. Results In the study 55,893 surgeries were identified: 26,073 primary hernia repairs, 462 recurrent hernia repairs, 3399 laparoscopic hernia repairs, 9414 hydrocele repairs and 16,545 orchiopexies. Pediatric surgeons performed 89% of primary hernia repairs with an annual median surgeon volume of 4 cases/year. Pediatric urologists performed 62% of hydrocelectomies and 83% of orchiopexies with annual median surgeon volumes of 6 and 24, respectively. Pediatric surgeons performed all procedures in younger patients and performed more female and incarcerated hernia repairs.

Conclusions Pediatric surgeons operate on younger patients and treat more patients with inguinal hernias while pediatric urologists care for more boys with undescended testes and hydroceles. This knowledge of referral patterns and care between specialties with overlapping expertise will allow improvements in training and access. Levels of evidence Cost Effectiveness Study, Level of Evidence III.

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Institution

(Chan, Kurzrock) Department of Urology, University of California Davis Children's Hospital, Sacramento, CA, United States (Durbin-Johnson) Division of Biostatistics, University of California Davis, Davis, CA, United States

Publisher

W.B. Saunders

Year of Publication

2016

First trimester phthalate exposure and male newborn genital anomalies.

Sathyanarayana S., Grady R., Barrett E.S., Redmon B., Nguyen R.H.N., Barthold J.S., Bush N.R., Swan S.H.

Embase

Environmental Research. 151 (pp 777-782), 2016. Date of Publication: 01 Nov 2016.

[Article]

AN: 612690975

Background Anti-androgenic phthalates are environmental chemicals that affect male genital development in rodents leading to genitourinary birth defects. We examined whether first trimester phthalate exposure may exert similar effects in humans leading to an increased incidence of newborn male genital anomalies in a multi-center cohort study. **Methods** We recruited first trimester pregnant women within The Infant Development and the Environment Study (TIDES) from 2010 to 2012 from four study centers and limited analyses to all mother/male infant dyads who had complete urinary phthalate and birth exam data (N=371). We used multivariate logistic regression to determine the odds of having a genital anomaly in relation to phthalate exposure. **Results** Hydrocele was the primary abnormality observed in the cohort (N=30) followed by undescended testes (N=5) and hypospadias (N=3). We observed a statistically significant 2.5 fold increased risk (95% CI 1.1, 5.9) of having any anomaly and 3.0 fold increased risk (95% CI 1.2, 7.6) of isolated hydrocele in relation to a one log unit increase in the sum of di-ethylhexyl phthalate (DEHP) metabolites. **Conclusions** First trimester urinary DEHP metabolite concentrations were associated with increased odds of any newborn genital anomaly, and this association was primarily driven by isolated hydrocele which made up the majority of anomalies in newborn males. The association with hydrocele has not been previously reported and suggests that it may be an endpoint affected by prenatal phthalate exposures in the first trimester of development. Future human studies should include hydrocele assessment in order to confirm findings.

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Embase

Institution

(Sathyanarayana, Grady) Seattle Children's Research Institute, Seattle, WA, United States

(Sathyanarayana) Department of Environmental and Occupational Health Sciences, University of Washington, Seattle, WA, United States

(Sathyanarayana) Department of Pediatrics, University of Washington School of Medicine, Seattle, WA, United States

(Barrett) Department of Obstetrics and Gynecology, University of Rochester School of Medicine and Dentistry, Rochester, NY, United States

(Redmon) Department of Endocrinology, University of Minnesota Medical School, Minneapolis, MN, United States

(Nguyen) Division of Epidemiology & Community Health, University of Minnesota, Minneapolis, MN, United States

(Barthold) Division of Urology, Department of Surgery, Nemours/Alfred I DuPont Hospital for Children, DE, United States

(Bush) Departments of Psychiatry and Pediatrics, University of California, San Francisco, United States

(Swan) Department of Preventive Medicine, Icahn School of Medicine at Mount Sinai, New York City, NY, United States

Publisher

Academic Press Inc. (E-mail: apjcs@harcourt.com)

Year of Publication

2016

489.

A 22-year retrospective study: educational update and new referral pattern of age at orchidopexy.

Wei Y., Wu S.-D., Wang Y.-C., Lin T., He D.-W., Li X.-L., Liu J.-H., Liu X., Hua Y., Lu P., Zhang D.-Y., Wen S., Wei G.-H.

Embase

BJU International. 118 (6) (pp 987-993), 2016. Date of Publication: 01 Dec 2016.

[Article]

AN: 611875459

Objectives: To determine the current age at orchidopexy in China and whether changing targets have altered practice, as research suggesting progressive deterioration in an undescended testis (UDT) has led to the reduction in the target age for orchidopexy to 6-12 months but it is still unknown whether changing targets have altered practice.

Patients and Methods: The demographics of orchidopexies performed in the Children's Hospital of Chongqing Medical University between 1993 and 2014 were reviewed. A survey of the general public's awareness of UDT and survey of primary healthcare practitioners' current opinion on age at orchidopexy and referral patterns were performed.

Result(s): In all, 3784 orchidopexies were performed over 22 years. The median age at orchidopexy fell between 1993 and 2014. There was an initial drop in the median age for orchidopexy between 2000 and 2010 (36 months) compared with 1993 and 2000 (48 months) ($P < 0.05$); however, beyond the corresponding target age (<18 months). The age for orchidopexy between 2010 and 2014 was also beyond the corresponding target age (6-9 months). The survey of the general public showed that 0.98% had knowledge of UDT and none of them knew about the target age for orchidopexy in the survey of 5393 cases. In all, 63.46% of them were told about the UDT by healthcare practitioners at the 1-4 months postnatal baby check. Furthermore, only 2% of the healthcare practitioners knew the recommended age for orchidopexy was 6-9 months and only 14.3% of them would directly make a surgical referral to paediatric surgery specifically at this point.

Conclusion(s): The recommended orchidopexy age is not being achieved and we recognise the national need to address this. The approach should include the right cognition of cryptorchidism among the general public and earlier primary care referral directly from the routine postnatal baby check to a specialist centre prepared to undertake surgery in this age group.

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Institution

(Wei, Wu, Wang, Lin, He, Li, Liu, Hua, Lu, Zhang, Wei) Ministry of Education Key Laboratory of Child Development and Disorders, Key Laboratory of Pediatrics in Chongqing,

CSTC2009CA5002, China International Science and Technology Cooperation base of Child's Development and Critical Disorders, Children's Hospital of Chongqing Medical University,

Chongqing, China (Wei, Wu, Wang, Liu, Wei) Department of Pediatric Surgery, Children's Hospital of Chongqing Medical University, Chongqing, China

(Lin, Li, Hua, Zhang, Wen, Wei) Department of Respiratory Medicine, Children's Hospital of Chongqing Medical University, Chongqing, China

(He, Liu, Lu, Wei) Department of Pathology, Children's Hospital of Chongqing Medical University, Chongqing, China

Publisher

Blackwell Publishing Ltd (E-mail: customerservices@oxonblackwellpublishing.com)

Year of Publication

2016

490.

Testicular growth during puberty in boys with and without a history of congenital cryptorchidism. Sadov S., Koskenniemi J.J., Virtanen H.E., Perheentupa A., Petersen J.H., Skakkebaek N.E., Main K.M., Toppari J.

Embase

Journal of Clinical Endocrinology and Metabolism. 101 (6) (pp 2570-2577), 2016. Date of Publication: June 2016.

[Article]

AN: 610763777

Context: The pattern of testicular growth during puberty may provide important information about early testicular damage and reproductive potential in adulthood.

Objective(s): To evaluate pubertal testicular growth in boys with congenital cryptorchidism and controls.

Design(s): Longitudinal case-control study.

Setting(s): Andrological Research Center, University of Turku.

Participant(s): Altogether, 119 boys participated: 51 cases with a history of congenital cryptorchidism and 65 controls fulfilled the inclusion criteria.

Intervention(s): None.

Main Outcome Measure(s): Testicular volume by an orchidometer (mL) and ultrasound (mL), testicular length by a ruler (mm), and onset of pubertal testicular growth (y). Longitudinal testicular growth was analyzed with a nonlinear mixed-effect model.

Result(s): The mean age of the onset of pubertal testicular growth (age at the attainment of ≥ 3 mL by orchidometer) was 11.7 and 11.8 years in cryptorchid cases and controls, respectively. The difference between cases and controls was not significant. Modeled postpubertal testicular size was smaller among bilaterally and unilaterally undescended testis than in controls. There was a high level of agreement between testicular sizes of 3 mL by orchidometer and 25 mm by ruler as cut-offs in definition of the onset of puberty. An orchidometer size of 3 mL and ruler

length of 25 mm corresponded to 1.6 and 1.7 mL by ultrasound (with Lambert's formula), respectively.

Conclusion(s): Testicular growth in puberty was impaired in congenitally cryptorchid boys. This suggests a poor perinatal development of the cryptorchid testis. The timing of the onset of pubertal testicular growth, however, did not differ which suggests an intact hypothalamic-pituitary axis.

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Status

Embase

Institution

(Sadov, Koskenniemi, Virtanen, Perheentupa, Toppari) Departments of Physiology and Pediatrics, University of Turku and Turku University Hospital, Obstetrics and Gynaecology, Turku FI-20520, Finland (Koskenniemi, Petersen, Skakkebaek, Main) Department of Growth and Reproduction, International Center for Research, Research Training in Endocrine Disruption of Male Reproduction and Child Health (EDMaRC), University of Copenhagen, Rigshospitalet, Copenhagen DK-2100, Denmark

Publisher

Endocrine Society (E-mail: mzendell@endo-society.org)

Year of Publication

2016

491.

Why do undescended testes and posterior urethral valve occur together?.

Wong J., Punwani V., Lai C., Chia J., Hutson J.M.

Embase

Pediatric Surgery International. 32 (5) (pp 509-514), 2016. Date of Publication: 01 May 2016.

[Article]

AN: 609900079

Background/Aim: Undescended testis (UDT) occurs in ~2 % of newborn males, and occasionally these infants also have posterior urethral valve (PUV). The cause of this relationship is uncertain.

We aimed to review the literature to identify publications documenting co-occurrence of UDT and PUV, and to summarise the theories of co-occurrence.

Method(s): A search of the literature (Embase, Medline, Pubmed; 1947-2015) was undertaken to identify publications describing the link between UDT in PUV patients, as well as PUV in UDT patients. Ten publications in English were found with both UDT and PUV: 9 articles describing the frequency of UDT in patients with PUV, and 1 article examining the frequency of PUV in infants with UDT.

Result(s): UDT occurred in 12-17 % of PUV compared with 1-2 % in the control population, consistent with a 10-fold increase. PUV occurred in 1.2 % of UDT patients compared with 0.01 % in the control population, consistent with a 100-fold increase.

Discussion(s): PUV leads to a 10-fold increase in occurrence of UDT, while the presence of UDT causes a 100-fold increase in occurrence of PUV. Four main theories of causation have been proposed, each of which have some merit but little supporting evidence, leaving the cause of simultaneous occurrence of PUV and UDT uncertain.

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Embase

Institution

(Wong, Punwani, Lai, Chia) University of Melbourne, Melbourne, Australia (Hutson) Department of Paediatrics, University of Melbourne, Melbourne, Australia

(Hutson) Urology Department, The Royal Children's Hospital, 50 Flemington Road, Parkville, Melbourne, VIC 3052, Australia

(Hutson) Douglas Stephens Surgical Research Group, Murdoch Childrens Research Institute, Melbourne, Australia

Publisher

Springer Verlag (E-mail: service@springer.de)

Year of Publication

2016

Clinical delineation of the PACS1-related syndrome-Report on 19 patients.

Schuurs-Hoeijmakers J.H.M., Landsverk M.L., Foulds N., Kukolich M.K., Gavrilova R.H., Greville-Heygate S., Hanson-Kahn A., Bernstein J.A., Glass J., Chitayat D., Burrow T.A., Husami A., Collins K., Wusik K., van der Aa N., Kooy F., Brown K.T., Gadzicki D., Kini U., Alvarez S., Fernandez-Jaen A., Mcgehee F., Selby K., Tarailo-Graovac M., Van Allen M., van Karnebeek C.D.M., Stavropoulos D.J., Marshall C.R., Merico D., Gregor A., Zweier C., Hopkin R.J., Chu Y.W.-Y., Chung B.H.-Y., de Vries B.B.A., Devriendt K., Hurles M.E., Brunner H.G.

Embase

American Journal of Medical Genetics, Part A. 170 (3) (pp 670-675), 2016. Date of Publication: 01 Mar 2016.

[Article]

AN: 608762990

We report on 19 individuals with a recurrent de novo c.607C>T mutation in PACS1. This specific mutation gives rise to a recognizable intellectual disability syndrome. There is a distinctive facial appearance (19/19), characterized by full and arched eyebrows, hypertelorism with downslanting palpebral fissures, long eye lashes, ptosis, low set and simple ears, bulbous nasal tip, wide mouth with downturned corners and a thin upper lip with an unusual "wavy" profile, flat philtrum, and diastema of the teeth. Intellectual disability, ranging from mild to moderate, was present in all. Hypotonia is common in infancy (8/19). Seizures are frequent (12/19) and respond well to anticonvulsive medication. Structural malformations are common, including heart (10/19), brain (12/16), eye (10/19), kidney (3/19), and cryptorchidism (6/12 males). Feeding dysfunction is presenting in infancy with failure to thrive (5/19), gastroesophageal reflux (6/19), and gastrostomy tube placement (4/19). There is persistence of oral motor dysfunction. We provide suggestions for clinical work-up and management and hope that the present study will facilitate clinical recognition of further cases.

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Institution

(Schuurs-Hoeijmakers, de Vries, Brunner) Department of Human Genetics, Radboud University Medical Center, Nijmegen, Netherlands (Landsverk) Department of Pediatrics, Sanford School of Medicine, University of South Dakota, Sanford Children's Health Research Center, Sanford Research, Sioux Falls, SD, United States

(Foulds, Greville-Heygate) Wessex Clinical Genetics Services, University Hospital Southampton NHS Foundation Trust, Southampton, United Kingdom

(Foulds, Greville-Heygate) Faculty of Medicine, University of Southampton, Southampton, United Kingdom

(Kukolich) Clinical Genetics, Cook Children's Hospital, Fort Worth, TX, United States

(Gavrilova) Department of Neurology, Mayo Clinic, Rochester, MN, United States

(Gavrilova) Medical Genetics, Mayo Clinic, Rochester, MN, United States

(Hanson-Kahn) Department of Genetics, Stanford University School of Medicine, Stanford, CA, United States

(Hanson-Kahn, Bernstein) Department of Pediatrics, Stanford University School of Medicine, Stanford, CA, United States

(Glass, Burrow, Husami, Collins, Wusik, Hopkin) Division of Human Genetics, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, United States

(Chitayat) Department of Obstetrics and Gynecology, Mount Sinai Hospital, University of Toronto, Toronto, ON, Canada

(Chitayat) Division of Clinical and Metabolic Genetics, The Hospital for Sick Children, University of Toronto, Toronto, ON, Canada

(Burrow) Department of Pediatrics, University of Cincinnati College of Medicine, Cincinnati, OH, United States

(van der Aa, Kooy) Department of Medical Genetics, University Hospital Antwerp, Antwerp, Belgium

(Brown) Southwest Thames Regional Genetics Centre, St George's Healthcare NHS Trust, London, United Kingdom

(Gadzicki) MVZ Endokrinologikum Hannover, Hannover, Germany

(Kini) Department of Clinical Genetics, Oxford University Hospitals NHS Trust, Oxford, United Kingdom

(Alvarez) NIMgenetics, Madrid, Spain

(Fernandez-Jaen) School of Medicine, European University of Madrid, Spain

(Fernandez-Jaen) Neuropediatric Department, Quiron University Hospital, Spain

(Mcgehee) Cook Children's Physician Network, United States

(Selby) Child and family Research Institute, Department of Pediatrics, University of British Columbia, Vancouver, BC, Canada

(Selby) Division of Pediatric Neurology, Department of Pediatrics, B.C. Children's and Women's Hospital, Vancouver, BC, Canada

(Tarailo-Graovac, van Karnebeek) Centre for Molecular Medicine and Therapeutics (TIDE-BC), Department of Pediatrics and Department of Medical Genetics, University of British Columbia, Vancouver, BC, Canada

(Van Allen) Child and family Research Institute, Department of Medical Genetics, University of British Columbia, Vancouver, BC, Canada

(Stavropoulos, Marshall) Genome Diagnostics, Department of Paediatric Laboratory Medicine, the Hospital for Sick Children, Toronto, ON, Canada
(Merico) The Centre for Applied Genomics, Genetics and Genome Biology, The Hospital for Sick Children, Toronto, ON, Canada
(Gregor, Zweier) Institute of Human Genetics, Friedrich-Alexander-Universitat Erlangen-Nurnberg, Erlangen, Germany
(Chu, Chung) Department of Paediatrics and Adolescent Medicine, Centre for Genomic Sciences, LKS Faculty of Medicine, The University of Hong Kong, Hong Kong
(Devriendt) Centre for Human Genetics, KU Leuven, Leuven, Belgium
(Hurles) Human Genetics, Wellcome Trust Sanger Institute, Cambridge, United Kingdom
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493.

A systematic review and meta-analysis of comparative studies assessing the efficacy of luteinizing hormone-releasing hormone therapy for children with cryptorchidism.

Li T., Gao L., Chen P., Bu S., Cao D., Yang L., Wei Q.

Embase

International Urology and Nephrology. 48 (5) (pp 635-644), 2016. Date of Publication: 01 May 2016.

[Review]

AN: 608644314

Purpose: To assess the efficacy of intranasal luteinizing hormone-releasing hormone (LHRH) therapy for cryptorchidism.

Material(s) and Method(s): Eligible studies were identified by two reviewers using PubMed, Embase, and Web of Science databases. Primary outcomes were complete testicular descent rate, complete testicular descent rate for nonpalpable testis, and pre-scrotal and inguinal testis. Secondary outcomes included testicular descent with different medicines strategy and a subgroup analysis.

Result(s): Pooled data including the 1255 undescended testes showed that complete testicular descent rate was 20.9 % in LHRH group versus 5.6 % in the placebo group, which was significantly different [relative risk (RR) 3.94, 95 % confidence interval (CI) 2.14-7.28, P < 0.0001]. There was also a significant difference in the incidence of pre-scrotal and inguinal position testis descent, with 22.8 % in the LHRH group versus 3.6 % in the placebo group (RR 5.79, 95 % CI 2.94-11.39, P < 0.00001). However, side effects were more frequent in the LHRH group (RR 2.61, 95 % CI 1.52-4.49, P = 0.0005). There were no significant differences for nonpalpable testes.

Conclusion(s): LHRH had significant benefits on testicular descent, particularly for inguinal and pre-scrotal testes, which was also accompanied by temporary slight side effects.

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Embase

Institution

(Li, Gao, Chen, Bu, Cao, Yang, Wei) Department of Urology, Institute of Urology, West China Hospital, Sichuan University, No. 37 Guoxue Xiang, Chengdu 610041, China

Publisher

Springer Netherlands

Year of Publication

2016

494.

Superb microvascular imaging for the detection of parenchymal perfusion in normal and undescended testes in young children.

Lee Y.S., Kim M.-J., Han S.W., Lee H.S., Im Y.-J., Shin H.J., Lee M.-J.

Embase

European Journal of Radiology. 85 (3) (pp 649-656), 2016. Date of Publication: 01 Mar 2016.

[Article]

AN: 608238032

Objectives To compare the detectability of perfusion difference between normal and undescended testes (UDT) in young children using conventional Power Doppler Imaging (PDI) and Superb Microvascular Imaging (SMI). **Methods** We prospectively performed testicular ultrasonography including PDI and SMI for the evaluation of microvascular flow in young children. Microvascular flow was categorized into four grades (grade 0-4). Statistical analysis was performed to compare the differences between undescended and normal testes. **Results** We imaged 40 testes from 20 boys (age, 2-29 months). Testes sizes and volumes were similar between the 29 normal and 11 UDT. PDI demonstrated low grade flow in most normal (19/29) and UDT (11/11) without difference ($P = 0.130$). However, SMI detected differences in flow grades between normal and UDT ($P < 0.001$). In univariate analysis, age (odds ratio [OR], 0.829; $P = 0.012$) and low grade flow on SMI (OR of grade 0, 51.886 with $P < 0.001$ and OR of grade 1, 14.29 with $P = 0.017$) were associated with UDT. These parameters were also significant in multivariate analysis with larger area under the curve, compared with the results using PDI (0.892 vs. 0.726, $P = 0.002$). **Conclusions** SMI can detect perfusion difference between normal and UDT in young children better than PDI.

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Status

Embase

Author NameID

Lee, Mi-Jung; ORCID: <http://orcid.org/0000-0003-3244-9171>

Institution

(Lee, Han, Im) Department of Urology, Urological Science Institute, Severance Children's Hospital, Yonsei University, College of Medicine, 50-1 Yonsei-Ro, Seodaemun-Gu, Seoul 120-752, South Korea (Kim, Shin, Lee) Department of Radiology, Research Institute of Radiological Science, Severance Children's Hospital, Yonsei University, College of Medicine, 50-1 Yonsei-Ro, Seodaemun-Gu, Seoul 120-752, South Korea

(Lee) Biostatistics Collaboration Unit, Yonsei University, College of Medicine, 50-1 Yonsei-Ro, Seodaemun-Gu, Seoul 120-752, South Korea

Publisher

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Year of Publication

2016

495.

Causes of hypogonadotropic hypogonadism predict response to gonadotropin substitution in adults.

Rohayem J., Sinthofen N., Nieschlag E., Kliesch S., Zitzmann M.

Embase

Andrology. 4 (1) (pp 87-94), 2016. Date of Publication: 01 Jan 2016.

[Article]

AN: 607874422

Germ cell and Sertoli cell proliferation and maturation in human testes occur in three main waves, during the late fetal and early neonatal period and at early puberty. They are triggered by periods of increased activity of the hypothalamic-pituitary-gonadal (HPG) axis. In hypogonadotropic hypogonadism (HH), these processes are variably disturbed. The objective of this study was to explore whether success of gonadotropin replacement in HH men is predictable by the origin of HH, indicating time of onset and severity of GnRH/gonadotropin deficiency. The data of 51 adult HH patients who had undergone one cycle of hCG/FSH treatment were reviewed. Five groups were established, according to the underlying HH origin. Therapeutic success by final bi-testicular volumes (BTVs) final sperm concentrations (SC) and conception rates were compared and related to baseline parameters, indicative of the degree of HPG-axis disruption. Overall, BTVs rose from 13 +/- 15 to 27 +/- 15 mL, spermatogenesis was induced in 98%, with mean SCs of 15 +/- 30 mill/mL, spontaneous pregnancies in 37% and additional 18% via intracytoplasmic sperm injection. Kallmann syndrome patients had the poorest responses (BTV: 16.9 +/- 10 mL; SC: 3.5 +/- 5.6 mill/mL), followed by patients with congenital/infancy-acquired multiple pituitary hormone deficiencies (MPHD) and patients with HH+absent puberty (BTV: 21 +/- 14/24 +/- 9 mL; SC: 5.5 +/- 6.5/ 14.5 +/- 23.8 mill/mL). HH men with pubertal arrest and with post-pubertally acquired MPHD had the best results (BTV: 36 +/- 14/38 +/- 16 mL; SC: 25.4 +/- 34.2/29.9 +/- 50.5 mill/mL). Earlier conception after 20.3 +/- 11.5 months (vs. 43.1 +/- 43.8; p = 0.047) of gonadotropin treatment with higher pregnancy rates (62% vs. 42%) was achieved in the two post-pubertally acquired HH subgroups, compared to the three pre-pubertally acquired. Therapeutic success was higher in patients without previously undescended testes, with higher baseline BTVs (pre- vs. post-pubertal HH: 5 +/- 4 mL vs. 26 +/- 16 mL; p < 0.0001) and higher baseline inhibinB levels (pre- vs. post-pubertal HH: 16.6 vs. 144.5 pg/mL; p = 0.0004). The cause of HH is a valuable predictor of outcome of gonadotropin replacement in adults.

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Status

Embase

Institution

(Rohayem, Sinthofen, Nieschlag, Kliesch, Zitzmann) Centre of Reproductive Medicine and Andrology, Department of Clinical Andrology, University of Muenster, Muenster, Germany
(Nieschlag) Center of Excellence in Genomic Medicine Research, King Abdulaziz University, Jeddah, Saudi Arabia

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2016

496.

Ethnic patterns of hypospadias in New Zealand do not resemble those observed for cryptorchidism and testicular cancer: Evidence of differential aetiology?.

Gurney J.K., Stanley J., Shaw C., Sarfati D.

Embase

Andrology. 4 (1) (pp 82-86), 2016. Date of Publication: 01 Jan 2016.

[Article]

AN: 607874420

It has been proposed that hypospadias, cryptorchidism, poor semen quality and testicular cancer might share common prenatal causes. We have previously demonstrated similar ethnic patterns for the incidence of testicular cancer and cryptorchidism - a known risk factor for testicular cancer. If the underlying exposure(s) that cause hypospadias, cryptorchidism and testicular cancer are shared, then we would expect the incidence relationship between ethnic groups to follow the same pattern across all three conditions. We followed a birth cohort of 318 345 eligible male neonates born in New Zealand between 2000-2010, and linked routinely collected maternity records with inpatient hospitalization and mortality records through to 2011. We searched hospitalization records for diagnoses of hypospadias, and used mortality records for censoring. We used Poisson regression methods to compare the relative risk of hypospadias between ethnic groups, adjusting for perinatal risk factors and total person time. We observed that

European/Other children had the highest risk of hypospadias, with Maori, Pacific and Asian boys having around 40% lower risk of disease compared with this group (adjusted relative risk [RR]: Maori 0.62, 95% CI 0.55-0.70; Pacific 0.62, 95% CI 0.53-0.72; Asian 0.57, 95% CI 0.47-0.69). This contrasts substantially with our previous observations for cryptorchidism and testicular cancer, where Maori males have the greatest risk. Our observations suggest that - at least in New Zealand - the exposures that drive the development of hypospadias may differ to those that drive the development of cryptorchidism and/or testicular cancer.

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Embase

Institution

(Gurney, Stanley, Shaw, Sarfati) Department of Public Health, University of Otago, Wellington, New Zealand

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497.

Consequences of bilateral cryptorchidism in adults.

Adomaitis R., Vincel B., Eidukaite A., Ostaneviciute E., Kirka R., Bilius V., Malcius D., Verkauskas G., Hadziselimovic F.

Embase

Andrologia. 48 (9) (pp 933-938), 2016. Date of Publication: 01 Nov 2016.

[Article]

AN: 607781512

Bilateral cryptorchidism treatment results are often shadowed by the majority of unilateral cases. We report the long-term follow-up results of boys treated for bilateral cryptorchidism during childhood. Patients treated in two main paediatric surgery centres were selected from medical registries and invited for a clinical examination including scrotal ultrasound, salivary testosterone

measurement and a semen sample. Thirty-six men (38.3%) replied to the written invitation, and 21 agreed to be examined. The mean age at orchidopexy was 74 months (range 24-138). Sperm count was 0.42×10^6 (SD $\pm 0.64 \times 10^6$) ml⁻¹. The correlation between total testicular volume and total sperm count was statistically significant ($r = 0.481$; $P = 0.032$). These results show that surgical treatment of bilateral cryptorchidism after the age of 2 years does not prevent infertility. Sperm count and endocrine evaluation advocated after the treatment of bilateral cryptorchidism in all adult patients.

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Status

Embase

Institution

(Adomaitis) Centre of Urology, Faculty of Medicine, Vilnius University, Vilnius, Lithuania (Vincel,

Verkauskas) Children's Surgery Centre, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

(Eidukaite) Centre for Innovative Medicine, State Research Institute, Vilnius, Lithuania

(Ostaneviciute) Centre of Laboratory Medicine, Vilnius University, Vilnius, Lithuania

(Kirka) Faculty of Mathematics and Informatics, Vilnius University, Vilnius, Lithuania

(Bilius) Department of Pediatrics, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

(Malcius) Department of Pediatric Surgery, Lithuanian University of Health Sciences, Kaunas, Lithuania

(Hadziselimovic) Institute for Andrology, Liestal, Switzerland

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498.

Surgical Management of the Undescended Testis: Recent Advances and Controversies.

Elder J.S.

Embase

European Journal of Pediatric Surgery. 26 (5) (pp 418-426), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 612276569

Undescended testis (UDT) is the most common disorder of sexual development in boys and affects 3.5% of male newborns. Although approximately half of newborn UDTs descend spontaneously, some boys develop an ascending testis later in childhood. Recent guideline recommendations advocate orchiopexy by 18 months of age to maximize potential for fertility and perhaps reduce the risk for testicular carcinoma in the future. For palpable testes, a standard inguinal approach is appropriate. However, the prescrotal approach is often effective for low inguinal testes and reduces surgical time and patient discomfort with an equivalent success rate in boys with an ascending testis. Some advocate monitoring until adolescence to determine whether the testis will spontaneously descend into the scrotum, but data do not support this approach. Instead, prompt orchiopexy is recommended. In boys with a nonpalpable testis, approximately 50% are abdominal or high in the inguinal canal and 50% are atrophic, typically in the scrotum. Routine inguinal/scrotal ultrasound is not recommended, although in an older boy who is overweight, it is appropriate. If the patient has contralateral testicular hypertrophy, scrotal exploration is appropriate, and removal of the testicular remnant and contralateral scrotal orchiopexy to prevent future contralateral testicular torsion is recommended. In most cases, diagnostic laparoscopy is advised to determine whether the testis is abdominal. For the abdominal testis, there are numerous treatment options. If the testis is mobile or a peeping testis just distal to the internal inguinal ring, standard one-stage laparoscopic or open orchiopexy should be attempted using the Prentiss maneuver. If the testicular vessels are short or the testis is not mobile, a two-stage Fowler-Stephens orchiopexy is appropriate. The second stage can be performed laparoscopically or open. Another option is microvascular testicular autotransplantation, which is a technically demanding procedure. Surgical results of abdominal orchiopexy are highly variable, short term, and highly subjective. Prospective clinical trials with follow-up into adolescence and adulthood are necessary to assess the success of various surgical approaches.

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Embase

Institution

(Elder) Division of Pediatric Urology, Massachusetts General Hospital, Boston, MA, United States

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Year of Publication

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499.

Hormonal Aspects of the Pathogenesis and Treatment of Cryptorchidism.

Cortes D., Holt R., De Knecht V.E.

Embase

European Journal of Pediatric Surgery. 26 (5) (pp 409-417), 2016. Date of Publication: 01 Oct 2016.

[Review]

AN: 612276167

A normal functioning hypothalamic-pituitary-testicular axis is required for normal testicular descent. The percentage of cases that result from a disturbance in this axis remains controversial. Much has yet to be learnt about cryptorchidism, but it seems that the existence of Ad spermatogonia is essential for later fertility. Bilateral cryptorchid patients have a high risk of later infertility, even though they undergo early surgery for cryptorchidism. It is possible today to distinguish - to a certain extent - between three different groups of cryptorchid patients based on testicular histology, gonadotropins, and inhibin B at the time of early surgery: Group 1, patients suspected of prepubertal transient hypothalamic-pituitary-testicular hypofunction and a high risk of later infertility; Group 2, patients with hypergonadotropic hypogonadism and a primary testicular dysfunction; and Group 3, patients with normal histology and normal serum levels of inhibin B and gonadotropins at the time of early surgery and a low risk of later infertility. Given the potential adverse effects of hormonal treatment, attention should be directed toward small doses of adjuvant gonadotropin-releasing hormone (GnRH) treatment for those who might benefit the most, that is, bilateral cryptorchid boys at early surgery without evidence of normal maturation of gonocytes into Ad spermatogonia. Optimally, gonadotropin levels in such patients should be measured to ensure that levels are not compensatorily elevated, thereby supporting the suspicion of hypothalamic-pituitary-testicular hypofunction. Studies of GnRH-supplementary treatment should include testicular biopsy at surgery and at follow-up in childhood as well as examinations of fertility potential in adulthood.

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Embase

Institution

(Cortes, Holt, De Knecht) Department of Pediatrics, Hvidovre Hospital, University of Copenhagen, Copenhagen, Denmark (Cortes) Department of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

Publisher

Georg Thieme Verlag (E-mail: kunden.service@thieme.de)

Year of Publication

2016

500.

What Animal Models of Testicular Descent and Germ Cell Maturation Tell Us about the Mechanism in Humans.

Hutson J.M., Li R., Vikraman J., Loebenstein M.

Embase

European Journal of Pediatric Surgery. 26 (5) (pp 390-398), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 612276091

Testicular descent occurs in most mammals in two main steps that have different hormonal control and anatomical processes. The evolution of testicular descent reveals the same basic processes in humans and animals, with minor differences in timing and anatomy, especially the location of the scrotum and the processus vaginalis. Animal models are useful as they reveal some embryological processes that cannot be studied easily in humans, such as the potential role of the mammary line and the role of the genitofemoral nerve. Postnatal germ cell development is very similar in animal models and humans, except for the timing of arrival of the testis into the scrotum, which is before birth in humans versus around puberty in rodents. Once all the minor differences between animal models and humans are taken into account, animal experimentation

has provided amazing insights into the mechanisms of testicular descent, and recently, how the postnatal germ cell develops in normally descended and undescended testes.

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Status

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Institution

(Hutson, Li) F. Douglas Stephens Surgical Research Group, Murdoch Childrens Research Institute, Victoria, Australia (Hutson, Li, Vikraman) Department of Urology, Royal Children's Hospital Melbourne, 50 Flemington Road, Parkville, Victoria 3052, Australia

(Hutson) Department of Paediatrics, University of Melbourne, Victoria, Australia

(Loebenstein) Department of Medicine, Faculty of Medicine, Dentistry and Health Sciences, University of Melbourne, Victoria, Australia

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501.

Comments to Recent Guidelines on Undescended Testis.

Kutasy B., Thorup J.M., Wester T.

Embase

European Journal of Pediatric Surgery. 26 (5) (pp 432-435), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 612275844

During the past couple of decades, our understanding of the treatment of undescended testis (UDT) has hugely expanded and it is still dynamically changing: new diagnostic tools are available, and experimental procedures are becoming a real-life treatment options. Our community needs to continuously update our guidelines. It is also our responsibility to build up, not a uniform, but a patient-oriented guideline which can provide information for both primary care

providers and pediatric surgeons. Here, in Europe, we endeavor to change the different national guidelines to one common European pediatric surgical guideline in the treatment of UDT.

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Status

Embase

Institution

(Kutasy, Wester) Department of Pediatric Surgery, Karolinska University Hospital, Stockholm, Sweden (Thorup) Department of Pediatric Surgery, Copenhagen University Hospital, Rigshospitalet, Copenhagen, Denmark

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Year of Publication

2016

502.

Long-Term Follow-Up after Treatment of Cryptorchidism.

Thorup J., Cortes D.

Embase

European Journal of Pediatric Surgery. 26 (5) (pp 427-431), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 612123764

When considering long-term prognosis and results in adult age following treatment of cryptorchidism in childhood there are three main issues to be discussed: cosmetics, fertility, and malignancy. In the present review, the most recent research on the topics related to summaries of well-known knowledge on the field is presented. To some extent a smaller testis in a higher scrotal position than normal must be accepted as a fair cosmetic result after orchidopexy in childhood. The smaller testis size is related to the impaired fertility potential of the testis. In cases with atrophy, testicular prosthesis implantation is an option with good operative results. The risk of impaired fertility potential in adults treated in childhood for cryptorchidism is still significant and

worst in bilateral disease. We need repetitive solid long-term follow-up data to show that orchidopexy performed within first year of life has markedly improved the fertility potential. Men previously having orchidopexy for cryptorchidism related to intra-abdominal testes, abnormal external genitalia and/or abnormal karyotype, and/or hypospadias are of special increased risk of developing testicular cancer. In these cases intratubular germ cell neoplasia may be diagnosed in prepubertal age. Early orchidopexy may lower the risk of developing testicular cancer.

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Embase

Institution

(Thorup) Department of Pediatric Surgery, Rigshospitalet, Copenhagen, Denmark (Thorup, Cortes) Institute for Clinical Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

(Cortes) Department of Pediatrics, University Hospital, Hvidovre, Denmark

Publisher

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Year of Publication

2016

503.

The Sertoli cell hormones inhibin-B and anti Mullerian hormone have different patterns of secretion in prepubertal cryptorchid boys.

Cortes D., Clasen-Linde E., Hutson J.M., Li R., Thorup J.

Embase

Journal of Pediatric Surgery. 51 (3) (pp 475-480), 2016. Date of Publication: 01 Mar 2016.

[Article]

AN: 608876148

Objectives and hypotheses The Sertoli-cells produce inhibin-B and Anti-Mullerian-Hormone (AMH). Much is still unknown about these hormones in prepubertal cryptorchids. The Sertoli-cells are mandatory for germ cell development. The aim of the study was to investigate if there are

differences in secretion pattern of Sertoli-cell hormones and their gonadotropin feed-back mechanisms. Methods Included were 94 prepubertal cryptorchid boys 0.5-13.1 years with measurements of serum-inhibin-B, Anti-Mullerian-Hormone (AMH), Luteinizing Hormone (LH) and Follicle Stimulation Hormone (FSH). The serum values were measured using commercially available kits. The hormonal values were related to age-matched normal values. Testicular biopsy was taken at orchiopexy. Results Inhibin-B positively correlated to AMH for 1-13 year-old patients ($p < 0.0001$), but not for 0.5-1 year-old patients ($p = 0.439$). For 0.5-1 year-old patients inhibin-B-values tended to decrease ($p = 0.055$), in contrast to AMH-values ($p = 0.852$). LH was elevated more often than FSH ($p = 0.014$). FSH and LH were positively associated in patients both 0.5-1 year ($p = 0.042$) and 1-13 years of age ($p < 0.0001$). LH correlated positively to inhibin-B ($p = 0.001$). In contrast, FSH did not correlate to inhibin-B or AMH ($p = 0.755$ and $p = 0.528$). The number of A-dark spermatogonia per tubular transverse section was positively correlated to inhibin-B serum level. Conclusion Our new finding of an association between LH and inhibin-B in infancy of cryptorchid boys may be essential for the transformation of gonocytes to A-dark spermatogonia. Previously, LH associated to inhibin-B was described in early puberty only. During the first year of life inhibin-B values decreased faster than AMH. The AMH-levels may just reflect the increased Sertoli cell number that occurs during the first 3 months of life.

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Status

Embase

Institution

(Cortes, Thorup) 4272 Department of Pediatric Surgery, Rigshospitalet, 9 Blegdamsvej, Copenhagen DK-2100, Denmark (Cortes) Section of Endocrinology, Department of Pediatrics, Hvidovre University Hospital, Copenhagen, Denmark

(Cortes, Thorup) Faculty of Health Science, University of Copenhagen, Denmark

(Clasen-Linde) Department of Pathology, Rigshospitalet, Copenhagen, Denmark

(Hutson, Li) Douglas Stephens Surgical Research Laboratory, Murdoch Childrens Research Institute, Melbourne, Australia

(Hutson) Department of Paediatric Urology, Royal Children's Hospital, Melbourne, Australia

(Hutson) Department of Paediatrics, University of Melbourne, Australia

Publisher

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2016

504.

Association between two single nucleotide polymorphisms of interleukin-27 gene and increased cryptorchidism risk.

Chen P., Pu Y., Zhou B., Wang Y., Tang T., Zhang P., Zhang L.

Embase

Andrologia. 48 (2) (pp 193-197), 2016. Date of Publication: 01 Mar 2016.

[Article]

AN: 604496762

Growing evidences have suggested the association between interleukin-27 and cryptorchidism. We aimed to investigate the relationship between IL-27 polymorphisms and cryptorchidism susceptibility. A total of 519 males were enrolled in a case-control study (150 cases and 369 normal subjects). The variants were discriminated using polymerase chain reaction-restriction fragment length polymorphism methods. The proportions of the major allele for rs153109 and rs17855750 were A and T with frequencies of 0.56 and 0.85 in cases and 0.51 and 0.91 in controls respectively (P values = 0.002, P value = 0.002). The heterozygous genotype of rs153109 and 17855750 was A/G and T/G with frequencies of 0.62 and 0.25 in cases and 0.39 and 0.17 in controls respectively (P values <0.001, P values <0.001). The A allele and A/G genotype of rs153109 polymorphisms contribute to increase cryptorchidism susceptibility, and G allele and T/G genotype of rs17855750 also contribute to increase cryptorchidism susceptibility, which implies that these allele and genotypes may be risk factors for the development of cryptorchidism.

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Embase

Institution

(Chen, Pu, Zhang) Department of Forensic Biology, West China School of Preclinical and Forensic Medicine, Sichuan University, Chengdu, China (Zhou, Wang, Zhang) Laboratory of Molecular Translational Medicine, Sichuan University, Key Laboratory of Obstetric, Gynecologic and Pediatric Diseases and Birth Defects of Ministry of Education, West China Institute of Women and Children's Health, West China Second University Hospital, Chengdu, China

(Tang) Department of Urology, Affiliated Hospital of North Sichuan Medical College, Nanchong, China

(Zhang) Department of Urology, West China Hospital, Sichuan University, Chengdu, China

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505.

Testicular biopsy in prepubertal boys: A worthwhile minor surgical procedure?.

Faure A., Bouty A., O'Brien M., Thorup J., Hutson J., Heloury Y.

Embase

Nature Reviews Urology. 13 (3) (pp 141-150), 2016. Date of Publication: 01 Mar 2016.

[Review]

AN: 607841787

No consensus exists regarding the precise role of testicular biopsy in prepubertal boys, although it is considered useful for assessing the potential consequences of undescended testes on fertility. Current scientific knowledge indicates that surgeons should broaden indications for this procedure. For example, the use of immunohistochemical markers such as OCT3/4, TSPY, Kit ligand (SCF) and ALPP (PLAP) has considerably facilitated the detection of germ cell tumour precursors, such as carcinoma in situ and/or gonadoblastoma. These markers are very important for evaluating malignancy risk in undervirilized patients with 46,XY disorders of sexual development. Testicular histology is also of considerable value in the prediction of both fertility potential and risk of cancer in individuals with undescended testes, particularly those with intraabdominal undescended testes. New possibilities for the preservation of fertility after gonadotoxic chemotherapy-even for prepubertal boys-are emerging. Cryopreservation of testicular tissue samples for the preservation of fertility-although still an experimental method at present-is appealing in this context. In our opinion, testicular biopsy in prepubertal boys is a minor procedure that can provide valuable information for predicting the risk of malignancy and fertility, and might be useful in fertility preservation in the near future.

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Institution

(Faure, Bouty, O'Brien, Hutson, Heloury) Royal Children's Hospital, Department of Paediatric Urology, 50 Flemington Road, Parkville, Melbourne, VIC 3052, Australia (Thorup) Department of Paediatric Surgery, Rigshospitalet, 9 Blegdamsvej, Copenhagen, Denmark

Publisher

Nature Publishing Group (Houndmills, Basingstoke, Hampshire RG21 6XS, United Kingdom)

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2016

506.

The anatomic findings during operative exploration for non-palpable testes: A prospective evaluation.

Gonzalez K.W., Dalton B.G., Snyder C.L., Leys C.M., St. Peter S.D., Ostlie D.J.

Embase

Journal of Pediatric Surgery. 51 (1) (pp 128-130), 2016. Date of Publication: 01 Jan 2016.

[Conference Paper]

AN: 607549199

Background We conducted a randomized trial comparing 1 and 2-stage laparoscopic orchiopexy for intra-abdominal testes. During recruitment, it became apparent that most patients with non-palpable testes do not require vascular division. In this report, we outline the location and quality of testes found during operative exploration in patients who consented for the study but were not randomized. Methods Analysis was performed on 80 patients undergoing operative exploration for non-palpable testes between 2007 and 2014. The location and pathology of undescended testes were analyzed. Results There were 87 preoperative non-palpable testes in 80 patients that were consented but not randomized to 1 or 2 stage orchiopexy with vascular division. Forty (46%) of nonrandomized testes were atrophic or absent, and 47 (54%) were normal in appearance. Sixty eight testes were evaluated via laparoscopy. The most common location for normal (81%) and absent/atrophic (70%) testes was the inguinal canal. Atrophic testes were more often left sided (72.5%) with normal testes equally divided. Patients with atrophic or absent testicles were

more likely to have a closed internal ring ($p < 0.01$). Conclusion This study demonstrates the majority of patients undergoing operative exploration for non-palpable testes will not require vascular division, and instead would be either atrophic or able to undergo traditional orchiopexy.

Level of Evidence: III.

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Institution

(Gonzalez, Dalton, Snyder, St. Peter) Department of Pediatric Surgery, Children's Mercy Hospital, Kansas City, MO, United States (Leys, Ostlie) Department of Surgery, University of Wisconsin, School of Medicine and Public Health, Madison, WI, United States

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2016

507.

NNT mutations: A cause of primary adrenal insufficiency, oxidative stress and extraadrenal defects.

Roucher-Boulez F., Mallet-Motak D., Samara-Boustani D., Jilani H., Ladjouze A., Souchon P.-F., Simon D., Nivot S., Heinrichs C., Ronze M., Bertagna X., Groisne L., Leheup B., Naud-Saudreau C., Blondin G., Lefevre C., Lemarchand L., Morel Y.

Embase

European Journal of Endocrinology. 175 (1) (pp 73-84), 2016. Date of Publication: July 2016.

[Article]

AN: 611185112

Objective: Nicotinamide nucleotide transhydrogenase (NNT), one of the several genes recently discovered in familial glucocorticoid deficiencies (FGD), is involved in reactive oxygen species detoxification, suggesting that extra-adrenal manifestations may occur, due to the sensitivity to oxidative stress of other organs rich in mitochondria. Here, we sought to identify NNT mutations

in a large cohort of patients with primary congenital adrenal insufficiency without molecular etiology and evaluate the degree of adrenal insufficiency and onset of extra-adrenal damages.

Method(s): Sanger or massive parallel sequencing of NNT and patient monitoring.

Result(s): Homozygous or compound heterozygous NNT mutations occurred frequently (26%, 13 unrelated families, 18 patients) in our cohort. Seven new mutations were identified: p.Met337Val, p.Ala863Glu, c.3G>A (p.Met1?), p.Arg129*, p.Arg379*, p.Val665Profs*29 and p.Ala704Serfs*19. The most frequent mutation, p.Arg129*, was found recurrently in patients from Algeria. Most patients were diagnosed belatedly (8-18 months) after presenting severe hypoglycemia; others experiencing stress conditions were diagnosed earlier. Five patients also had mineralocorticoid deficiency at onset. One patient had congenital hypothyroidism and two cryptorchidism. In follow-up, we noticed gonadotropic and genitalia impairments (precocious puberty, testicular inclusions, interstitial Leydig cell adenoma, azoospermia), hypothyroidism and hypertrophic cardiomyopathy. Intrafamilial phenotype heterogeneity was also observed.

Conclusion(s): NNT should be sequenced, not only in FGD, but also in all primary adrenal insufficiencies for which the most frequent etiologies have been ruled out. As NNT is involved in oxidative stress, careful follow-up is needed to evaluate mineralocorticoid biosynthesis extent, and gonadal, heart and thyroid function.

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Status

Embase

Institution

(Roucher-Boulez, Mallet-Motak, Jilani, Morel) Molecular Endocrinology and Rare Diseases, Lyon University Hospital, Bron, France (Roucher-Boulez, Morel) Claude Bernard Lyon 1 University, Lyon, France

(Samara-Boustani) Pediatric Endocrinology, Gynecology and Diabetology, Necker University Hospital, Paris, France

(Ladjouze) Pediatric Department, Bab El Oued University Hospital, Alger, Algeria

(Souchon) Pediatric Endocrinology and Diabetology, American Memorial Hospital, Reims, France

(Simon) Pediatric Endocrinology, Robert Debre Hospital, Paris, France

(Nivot) Department of Pediatrics, Rennes Teaching Hospital, Rennes, France

(Heinrichs) Pediatric Endocrinology, Queen Fabiola Children's University Hospital, Brussels, Belgium

(Ronze) Endocrinology Department, L.-Hussel Hospital, Vienne, France

(Bertagna) Endocrinology Department, Cochin University Hospital, Paris, France

(Groisne) Endocrinology Department, Lyon University Hospital, Bron-Lyon, France

(Leheup) Paediatric and Clinical Genetic Department, Nancy University Hospital, Vandoeuvre les Nancy, France

(Naud-Saudreau, Blondin) Pediatric Endocrinology and Diabetology, Bretagne Sud Hospital Center, Lorient, France

(Lefevre) Pediatric Endocrinology, Jeanne de Flandre Hospital, Lille, France

(Lemarchand) Pediatric Department, La Rochelle-Re-Aunis Hospital Group, La Rochelle, France

Publisher

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508.

Perspectives in pediatric pathology, chapter 14. Natural history of undescended testes.

Nistal M., Paniagua R., Gonzalez-Peramato P., Reyes-Mugica M.

Embase

Pediatric and Developmental Pathology. 19 (3) (pp 183-201), 2016. Date of Publication: May-June 2016.

[Review]

AN: 611174616

Cryptorchidism is one of the most frequent problems encountered in pediatric urology. Its causes, associated lesions, and prognosis in terms of fertility have been a source of interest and discrepancies for pediatric pathologists and urological surgeons.

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Status

Embase

Institution

(Nistal, Gonzalez-Peramato) Department of Pathology, Hospital la Paz, Universidad Auto Noma de Madrid, Calle Arzobispo Morcillo No. 2, Madrid 28029, Spain (Paniagua) Department of Cell Biology, Universidad de Alcalá, Madrid, Spain

(Reyes-Mugica) Department of Pathology, Children's Hospital of Pittsburgh, University of Pittsburgh Medical Center, Pittsburgh, PA, United States

Publisher

Society for Pediatric Pathology (P. O. Box 7075, Lawrence KS 66044, United States)

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509.

Increased prevalence of some birth defects in Korea, 2009-2010.

Lamichhane D.K., Leem J.-H., Park M., Kim J.A., Kim H.C., Kim J.H., Hong Y.-C.

Embase

BMC Pregnancy and Childbirth. 16 (1) (no pagination), 2016. Article Number: 61. Date of Publication: 22 Mar 2016.

[Article]

AN: 611090569

Background: Birth defects are a leading cause of neonatal and infant mortality, and several studies have indicated an increase in the prevalence of birth defects; more recent investigations have suggested that the trends of some defects are increasing in rapidly industrialized areas.

This study estimates the prevalence rate and types of birth defects in Korea.

Method(s): This study used medical insurance benefit data of 403,250 infants aged less than one year from the National Health Insurance Corporation from seven metropolitan areas in Korea for 2009 and 2010.

Result(s): The prevalence rate of birth defects was 548.3 per 10,000 births (95 % CI: 541.1-555.6), 306.8 among boys and 241.5 among girls. Anomalies of the circulatory system (particularly septal defects) were the most common (180.8 per 10,000), followed by defects of the genitourinary tract (130.1 per 10,000) (particularly obstructive genitourinary and undescended testis), musculoskeletal system (105.7 per 10,000), digestive system (24.7 per 10,000), and central nervous system (15.6 per 10,000).

Conclusion(s): Relatively higher rates of some birth defects were found in the metropolitan areas. The high differences of birth prevalences for septal heart defects and undescended testis are probably due in part to progress in clinical management and more frequent prenatal diagnosis. Environmental exposure might play a critical role in the development of some birth defects. In attempting to describe the prevalence and spatio-temporal variations of birth defects in Korea, establishment of a registry system of birth defects and environmental surveillance are needed.

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Status

Embase

Institution

(Lamichhane, Leem, Park, Kim, Kim) School of Medicine, Inha University, Department of Social and Preventive Medicine, Incheon, South Korea (Leem, Kim) School of Medicine, Inha University, Department of Occupational and Environmental Medicine, 7-206 3rd St., Shinhung Dong, Jung Gu, Incheon, South Korea

(Kim) Sejong University, 209 Neungdong-ro, Department of Integrative Bioscience and Biotechnology, Gwangjin-gu, Seoul 05006, South Korea

(Hong) College of Medicine, Seoul National University, Department of Preventive Medicine, Seoul, South Korea

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510.

Chromosome Xq28 duplication encompassing MECP2: Clinical and molecular analysis of 16 new patients from 10 families in China.

Yi Z., Pan H., Li L., Wu H., Wang S., Ma Y., Qi Y.

Embase

European Journal of Medical Genetics. 59 (6-7) (pp 347-353), 2016. Date of Publication: 01 Jun 2016.

[Article]

AN: 610444272

Introduction: Chromosome Xq28 duplications encompassing methyl-CpG-binding protein 2 gene (MECP2) are observed most in males with a severe neurodevelopmental disorder associated with hypotonia, spasticity, severe learning disability, delayed psychomotor development, and recurrent pulmonary infections. Most female carriers are asymptomatic due to extremely or completely skewed X-inactivation.

Method(s): A retrospective clinical and molecular study was conducted to examine 16 patients and two fetuses from 10 families who were identified among patients with Xq28 duplications who presented at genetic clinics.

Result(s): Of all 16 patients, 10 had a family history. Only one patient was female. All of the patients had no relevant pre-natal history. All of the patients exhibited severe psychomotor developmental delay, infantile hypotonia and recurrent infections. Some of the patients exhibited cardiac abnormalities, gastrointestinal mobility problems, hydrocele of tunica vaginalis, cryptorchidism, and autistic phenotypes. Additionally, neonatal kidney calculus, premature closure of the fontanel and pulmonary sequestration were found in the patients. Duplication sizes in these patients range from 0.21 to 14.391 Mb (most were smaller than 1 Mb), and all the duplications included host cell factor C1 (HCFC1), interleukin-1 receptor-associated kinase 1 (IRAK1), and MECP2. Bioinformatics analysis revealed that approximately half of the distal breakpoints were located within the low-copy repeats (LCRs), which may be involved in the recombination. The two fetuses were found to be healthy in the prenatal diagnosis.

Conclusion(s): This is the first large cohort of patients with MECP2 duplication syndrome, including a female, reported in China. Interestingly, neonatal kidney calculus, premature closure of the fontanel and pulmonary sequestration were first reported in this syndrome. However, it was difficult to distinguish if these patients represented unique cases or if these phenotypes can be considered as part of the syndrome. The correlation between the infrequent phenotypes and duplications/genes in the duplication region needs further systematic delineation. In conclusion, our study suggested that it is important to emphasize molecular genetic analysis in patients with developmental delay/intellectual disability and recurrent infections and that it is especially important for familial female carriers to accept prenatal diagnosis.

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Institution

(Yi, Pan, Li, Wu, Wang, Ma, Qi) Department of Central Laboratory, Peking University First Hospital, Beijing, China

Publisher

Elsevier Masson SAS (62 rue Camille Desmoulins, Issy les Moulineaux Cedex 92442, France)

Year of Publication

2016

Are anti-Mullerian hormone and its receptor polymorphism associated with the hormonal condition of undescended testes?.

Komarowska M.D., Milewski R., Charkiewicz R., Matuszczak E., Sulewska A., Zelazowska-Rutkowska B., Hermanowicz J., Niklinski J., Debek W., Hermanowicz A.

Embase

Advances in Medical Sciences. 61 (2) (pp 288-292), 2016. Date of Publication: 01 Sep 2016.

[Article]

AN: 610222439

Purpose Numerous genetic and endocrine factors are involved in the process of testicular descent, but only a few genetic causes have been reported in human. The aim of this study was to investigate the density and distribution of single nucleotide polymorphisms (SNPs) anti-Mullerian hormone (AMH) and AMHRII receptors in cryptorchid patients and determine potential hormone imbalance connected with undescended testes by assessing the levels of AMH, Insulin-like factor 3 (INSL3) and inhibin B. Materials and methods The serum hormone levels (AMH, INSL3 and inhibin B) were compared in the two groups - cryptorchidism (n = 105) and control group (n = 58). The frequency of AMHRII -482 A > G, AMHRII IVS 10 + 77 A > G, AMHRII IVS 5-6 C > T, and AMH Ile49Ser polymorphisms among cryptorchid boys were compared with the control group. Results None of the hormones levels were different between the cryptorchid and the control groups. All cases of IVS 5-6 C > T homozygote and heterozygote mutation were accompanied by an IVS 10 + 77 A > G and 482 A > G homozygote and heterozygote mutation. Interestingly, in most cases of all four polymorphisms, homozygote recessive genotype was associated with cases of cryptorchidism. However, the groups of patients were too small to draw definite conclusions. Conclusion The AMHRII -482 A > G, AMHRII IVS 10 + 77 A > G, AMHRII IVS 5-6 C > T and AMH Ile49Ser genotypes should be determined in a much larger group of boys with cryptorchidism.

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In-Process

Institution

(Komarowska, Matuszczak, Debek, Hermanowicz) Department of Pediatric Surgery, Medical University of Bialystok, ul. Waszyngtona 17, Bialystok 15-274, Poland (Milewski) Department of Statistics and Medical Informatics, Medical University of Bialystok, Bialystok, Poland (Charkiewicz, Sulewska, Niklinski) Department of Clinical Molecular Biology, Medical University of Bialystok, Bialystok, Poland (Zelazowska-Rutkowska) Department of Pediatric Laboratory Diagnostics, Medical University of Bialystok, Bialystok, Poland

(Hermanowicz) Department of Clinical Pharmacy, Medical University of Bialystok, Bialystok, Poland

Publisher

Medical University of Bialystok (Kilinskiego 1, Bialystok 15-089, Poland)

Year of Publication

2016

512.

Long-term efficacy of recombinant human growth hormone therapy in short-statured patients with Noonan syndrome.

Jeong I., Kang E., Cho J.H., Kim G.-H., Lee B.H., Choi J.-H., Yoo H.-W.

Embase

Annals of Pediatric Endocrinology and Metabolism. 21 (1) (pp 26-30), 2016. Date of Publication: March 2016.

[Article]

AN: 610087945

Purpose: Noonan syndrome (NS) is characterized by short stature, heart anomalies, developmental delays, dysmorphic features, cryptorchidism, and coagulation defects. Several studies reported the short-term effects of recombinant human growth hormone (rhGH) treatment on the improvement of height. This study was performed to evaluate the long-term efficacy of rhGH in children with NS in Korea.

Method(s): This study included 15 prepubertal NS children who received rhGH subcutaneously at a dose of 50-75 mug/kg/day for 6 days a week for at least >3 years. Pre- and posttreatment data, such as height, weight, bone age, insulin-like growth factor 1 (IGF-1), and IGF binding protein 3 (IGFBP-3) levels, were collected every 6 months.

Result(s): Chronologic age and bone age at the start of treatment were 7.97+/-1.81 and 5.09+/-2.12 years, respectively. Height standard deviation score (SDS) was increased from -2.64+/-0.64 to -1.54+/-1.24 years after 3 years ($P<0.001$). Serum IGF-1 SDS levels were elevated from -1.28+/-1.03 to -0.10+/-0.94 ($P<0.001$). Height SDS was more increased in subjects without PTPN11 mutations compared to those with mutations after 3 years ($P=0.012$). However, the other parameters, including bone age, IGF-1 SDS, and IGFBP-3 SDS, were not significantly different between patients with and without PTPN11 mutations.

Conclusion(s): Although this study included a relatively small number of patients, long-term rhGH therapy in NS patients was safe and effective at improving height, growth velocity, and serum IGF-1 levels, in accordance with previous studies. However, the meticulous monitoring of potential adverse events is still needed because of high dose of rhGH and preexisting hyperactivity of RAS-MAPK pathway. Patients with PTPN11 mutations demonstrated a decreased response to rhGH therapy compared to those without mutations.

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Embase

Institution

(Jeong, Kang, Cho, Lee, Choi, Yoo) Department of Pediatrics, Asan Medical Center Children's Hospital, University of Ulsan College of Medicine, Seoul, South Korea (Kim) Medical Genetics Center, Asan Medical Center Children's Hospital, University of Ulsan College of Medicine, Seoul, South Korea

Publisher

Korean society of pediatric endocrinology (E-mail: kspendo@gmail.com)

Year of Publication

2016

513.

The Effectiveness of hCG and LHRH in Boys with Cryptorchidism: A Meta-Analysis of Randomized Controlled Trials.

Bu Q., Pan Z., Jiang S., Wang A., Cheng H.

Embase

Hormone and Metabolic Research. 48 (5) (pp 318-324), 2016. Date of Publication: 06 Apr 2016.

[Article]

AN: 609712216

To systematically review the efficacy of hCG and LHRH on testicular descent in boys with cryptorchidism, comprehensive search was performed to identify randomized controlled trials (RCTs) in PubMed, EMBASE, the Cochrane Library, Wanfang Database, and China National Knowledge Infrastructure (CNKI) up to March 2014. Outcomes included testicular complete descent rate (TCDR) and cure rate of patients. Study quality was evaluated using the Jadad

scale. Meta-analysis was performed using Review Manager software. Finally, 13 studies were included. hCG and LHRH increased TCDR comparing with control group. The success rate of hCG and LHRH was 24 and 19%, respectively. Further, hCG and LHRH had significant effect on bilateral cryptorchidism, but not on unilateral cryptorchidism. All side effects were transitory and not severe, but if they have long-term harms were not clear. hCG and LHRH can effectively increase TCDR and there was no significant difference between them. However, the hormones cannot be recommended for everyone because of their low success rates and potential long-term harms. Further studies are needed to determine the efficacy of hormonal treatment for subtypes of cryptorchidism.

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Embase

Institution

(Bu) Department of Genetics, Northwest Women's and Children's Hospital, Xi'an, China (Pan, Wang, Cheng) Department of Clinical Pharmacy, Xi'an Jiaotong University Affiliated Children's Hospital, No.69 Xijuyuan Road, Xi'an 710003, China

(Jiang) Department of Psychiatry, University of Illinois at Chicago, Chicago, United States

Publisher

Georg Thieme Verlag (E-mail: kunden.service@thieme.de)

Year of Publication

2016

514.

How should we monitor boys with testicular microlithiasis?.

Yesil S., Tanyildiz H.G., Sahin G.

Embase

Pediatric Hematology and Oncology. 33 (3) (pp 171-177), 2016. Date of Publication: 02 Apr 2016.

[Article]

AN: 609208380

Testicular microlithiasis (TM), a rare condition characterized by calcification within the seminiferous tubules, is associated with benign and malignant disorders of the testis. We review current practices of following up pediatric patients diagnosed TM incidentally on scrotal

ultrasonography (US). We analyzed retrospectively patient characteristics, family history, indications for US, pathological features, US findings, outcome, and follow-up. At our institution, 2875 scrotal US examinations were performed on 2477 children with various scrotal complaints from 2008 to 2015. Testicular microlithiasis was detected in 81 patients (i.e., an incidence of 3.27%). Every 6 months, each patient underwent a clinical and ultrasonographic evaluation as well as serum tumor markers determination to detect a potential malignancy. Seventy-eight patients who had undergone scrotal US at least twice were included in this study. We evaluated the US studies for the type of TM (diffuse and focal) and change in follow-up studies. Testicular microlithiasis was typically diffuse (n = 56, 71.8%) and bilateral (n = 45, 57.7%), and it was detected the most frequently in the 9-11-year age group (27 patients, 34.6%). The most common comorbid conditions included undescended testes (31 patients, 39.7%) and hydrocele (11 patients, 14.1%). We found that serum tumor markers were within normal limits both at diagnosis and upon follow-up. No testicular tumors or new abnormal symptoms developed during the clinical follow-up. There is no convincing evidence that TM alone is premalignant in a pediatric population. In terms of follow-up, we advise regular self-examinations and annual US in the absence of risk factors.

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Institution

(Yesil, Tanyildiz, Sahin) Department of Pediatric Oncology and Hematology, Dr. Sami Ulus Maternity and Children's Health and Diseases Training and Research Hospital, Ankara, Turkey

Publisher

Taylor and Francis Ltd (E-mail: healthcare.enquiries@informa.com)

Year of Publication

2016

515.

Age at surgery and outcomes of an undescended testis.

Schneuer F.J., Holland A.J.A., Pereira G., Jamieson S., Bower C., Nassar N.

Embase

Pediatrics. 137 (2) (no pagination), 2016. Article Number: e20152768. Date of Publication: February 2016.

[Article]

AN: 608565823

BACKGROUND: Undescended testis (UDT) is the most common genital anomaly in boys. Current guidelines recommend surgery before 12 months of age to maximize fertility and potentially reduce the risk of future malignancy. We investigated the prevalence of UDT and examined rates of surgery and age at surgery in an Australian population.

METHOD(S): UDT was identified from all live-born infants in New South Wales, Australia, from 2001 to 2011 using routinely collected record-linked birth and hospital data. The prevalence of UDT, surgery rates, age at surgery, postsurgical outcomes, and risk factors for surgery performed later than the recommended age were evaluated.

RESULT(S): There were 10 875 (2.1%) boys with a recorded diagnosis of UDT. Corrective surgery was performed in 4980 (45.8%), representing a cumulative prevalence of 9.6 per 1000 male births. Five percent of surgeries were orchidectomies, and 9% of boys had revision surgery. Median age at surgery was 16.6 months (interquartile range 11.8 to 31.0 months), decreasing from 21 months for boys born in 2001 to 13 months for boys born in 2010. Among those boys having surgery before 36 months (n = 3897), 67% had corrective surgery after the recommended 12 months of age; socioeconomic disadvantage, regional/ remote area of residence, and lack of private health insurance were risk factors for having corrective surgery after 12 months.

CONCLUSION(S): One in 50 boys born are diagnosed with UDT; two-thirds had no report of corrective surgery. The age at surgery is decreasing; however, two-thirds of surgeries are performed after 12 months of age.

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PMC Identifier

26801912 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=26801912>]

Status

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Institution

(Schneuer, Nassar) Clinical and Population Perinatal Health Research, Kolling Institute, University of Sydney, Northern Sydney Local Health District, Charles Perkins Centre, St. Leonards, NSW 2006, Australia (Schneuer, Nassar) Menzies Centre for Health Policy, School of Public Health, Sydney Medical School, University of Sydney, NSW, Australia (Holland) Discipline of Paediatrics and Child Health, Children's Hospital at Westmead, Sydney Medical School, University of Sydney, NSW, Australia (Pereira) School of Public Health, Curtin University, Bentley, WA, Australia

(Jamieson, Bower) Telethon Kids Institute, University of Western Australia, Crawley, WA, Australia

Publisher

American Academy of Pediatrics (141 Northwest Point Blvd, P.O. Box 927, Elk Grove Village IL 60007-1098, United States)

Year of Publication

2016

516.

Opinion: Comment on evaluation and treatment of cryptorchidism: AUA/AAP and nordic consensus guidelines.

Hadziselimovic F.

Embase

Urologia Internationalis. 96 (3) (pp 249-254), 2016. Date of Publication: 01 Apr 2016.

[Review]

AN: 607997816

The ultimate goal in the treatment of cryptorchidism is to achieve normal fertility. However, in a substantial number of cryptorchid males, early and apparently successful orchidopexy does not improve fertility as it does not address the underlying pathophysiological cause, namely, the impaired transformation of gonocytes into Ad spermatogonia. It is important to realize that over half the patients presenting with unilateral cryptorchidism and the majority of those presenting with bilateral cryptorchidism have abnormal spermiogram which indicates that unilateral cryptorchidism is in fact a bilateral disease and therefore a serious andrological problem. More importantly, only testicular biopsy can nowadays determine which patient should benefit from hormonal therapy. This means that the rationale behind testicular biopsy is both diagnostic and therapeutic, particularly since LH-RHa hormonal therapy is a worthwhile solution to this andrological problem. In boys with a high risk of azoospermia development, adequate treatment with low doses of LH-RHa allowed 86% of subjects to achieve a normal sperm count. This strongly contrasts with the results of the 'surgery-only' group where not a single patient had a normal spermiogram and 20% suffered from azoospermia. Testicular biopsy is all the more justified that it allowed the detection of in situ carcinoma in 0.6% of all the cryptorchid boys studied. Even if hormonal pre-treatment only achieves successful epididymo-testicular descent in

20% of cases, this treatment should remain the first therapeutic choice because it may avoid resorting to surgery. In addition, it has no adverse effect on fertility and, in unsuccessful cases, facilitates orchidopexy and considerably helps reduce the incidence of post-surgical testicular atrophy, whether unilateral or, and this is a much more serious event, bilateral.

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Institution

(Hadziselimovic) Institute for Cryptorchidism Research, Kindermedizinisches Zentrum Liestal, Liestal CH-4410, Switzerland

Publisher

S. Karger AG

Year of Publication

2016

517.

Educational survey of regional general practitioner's management of paediatric patients with undescended testis.

Cho A., Ball M., Read K., Tharmapooopathy P., Ross A.R., Mathur A., Minocha A., Tsang T., Kulkarni M.

Embase

Journal of Pediatric Urology. 12 (3) (pp 151.e1-151.e7), 2016. Date of Publication: 01 Jun 2016.

[Article]

AN: 607644192

Introduction Recent recommendations have lowered the ideal age of surgery for undescended testis (UDT) to 3-6 months of age. However, many publications demonstrate that age at surgery is still above the recommended age of 1 year as originally suggested in 1996. **Aim** Through a web-based educational survey, we aimed to combine questions regarding General Practitioner's (GPs) management of these patients with educational slides with advice to update them with current recommendations. **Methods** The regional GPs were invited by email and letter to undertake the web-based questionnaire devised using SurveyMonkey. Educational slides were shown after each questionnaire slide. Feedback was immediate and a one-page summary was

emailed to the GP on completion. A pre- and post-educational intervention audit was undertaken to ascertain the change in age of referral for patients <5 years of age. Results 144 (36%) of 401 GPs undertook this survey. 84% were happy assessing infants (<1year) with UDT. 16% were unhappy discussing management with parents for palpable UDT. 52% were happy discussing malignant risk with parents. 80% thought that ultrasonography was routinely used. Optimal referral time was thought to be 6-12 months (42%) and time of surgery was 1-2 years (50%). 72% would refer a patient with palpable UDT after 6 months of age. Only 41% were happy to assess testicular size at puberty. 98% found this format of an educational survey was helpful. The average age of referral for patients <5 years improved significantly after educational intervention from 2.8 years in 2010 to 1.25 years in 2013 ($p < 0.01$). Discussion With an interactive survey, we were able assess and also educate the regional GPs with regard to management of paediatric patients with UDT. There is a varied range of knowledge and practice demonstrated which we hoped to standardise and thereby increase efficiency and decrease the age of referral. A large majority would refer patients with UDT after 6 months of age that would make the target of surgery <6 months unachievable unless they are aware of current recommendations. This study is limited by a 36% response rate but that is comparable to other surveys. Also, referrals come from other sources that were not included. The causal effect of the educational survey is hypothesised. Conclusion This educational survey has confirmed the varied management by GPs. The referral age was demonstrated to be reduced after this intervention and this process was widely accepted by GPs that undertook the educational survey.[Figure presented]

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(Cho, Ross, Mathur, Minocha, Tsang, Kulkarni) Department of Paediatric Surgery, Norfolk & Norwich University Hospital, Norwich, United Kingdom (Ball, Read, Tharmapooopathy) University of East Anglia, Norwich, United Kingdom

Publisher

Elsevier Ltd

Year of Publication

2016

Can separation of the scrotal sac in proximal hypospadias reliably predict the need for urethral plate transection?.

Arnaud A., Ferdynus C., Harper L.

Embase

Journal of Pediatric Urology. 12 (2) (pp 121.e1-121.e5), 2016. Date of Publication: 01 Apr 2016.

[Article]

AN: 607443520

Introduction One of the main challenges in proximal hypospadias repair is correcting curvature. The best technique to achieve this remains the object of debate. Indeed, some authors believe the urethral plate should be kept and used as often as possible. In some cases, however, even after extensive mobilization and dorsal plication, significant curvature remains and it is necessary to transect the urethral plate. Having a reliable pre-dissection marker of the need for urethral transection would be useful in choosing a technique. We wanted to determine if presence of marked separation of the scrotal sac (SSS), also referred to as bifid scrotum, could reliably predict the need for urethral plate transection. Study design We prospectively enrolled a series of boys with proximal hypospadias. We noted age, degree of hypospadias, meatal position, presence of cryptorchidism, and presence or absence of SSS. During surgery we fully degloved the penile shaft, freeing all ventral tissues, and radically dissected the more proximal bulbar urethra. We then performed an erection test. If there was residual curvature $<30^\circ$ we performed a dorsal plication, if it was $>30^\circ$ we transected the urethral plate. Results Twenty-nine patients were included, of whom 18 presented SSS. The average age was comparable in both groups, as was type of hypospadias and meatal position. We estimated transection of the urethral plate to be necessary in 15 out of the 18 children with SSS, and 2 out of the 11 children without SSS. The relative risk for requiring urethral plate transection in case of SSS in this series was 4.58. Conclusion Techniques that commit to urethral plate transection are criticized because they preclude using the urethral plate. In our study presence of SSS was predictive for the need to transect the plate. Obviously one can decide to keep the urethral plate at all cost, and mobilize it more than we did, or accept more residual curvature, but in reality our aim was to determine a preoperative marker allowing us to define a patient category. We believe presence of SSS is a marker of severity, and that this "severity" translates into "a less usable urethra". As recent studies caution us about the evolution of the reconstructed native urethra and the possibility that it may not grow as well as the other penile tissues, we believe this extra information could influence the surgeon's decision as to the most appropriate technique for each patient. Results of bivariate analysis. Results

Total	No SSS	SSS	SpAge, years
4.23	4.44	7.06	8.68

Urethral transection

Total	No SSS	SSS	SpAge, years
17 (58.6%)	2 (18.2%)	15 (83.3%)	0.001

A p-value <0.05 was considered statistically significant.

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Institution

(Arnaud, Harper) Department of Pediatric Surgery, CHU F Guyon, Bellepierre 97405, Reunion
(Ferdynus) Methodology Unit (USM), CHU F Guyon, Bellepierre, Reunion

Publisher

Elsevier Ltd

Year of Publication

2016

519.

Evaluation of external genital anomalies and the underlying factors in male newborns.

Pakniyat A., Fallah M.R., Fakour Z., Moloudi F., Khezri S., Masoudi S.

Embase

Iranian Journal of Neonatology. 7 (1) (pp 52-57), 2016. Date of Publication: March 2016.

[Article]

AN: 609360499

Background: External genital anomalies are the most common congenital disorders in male infants with unknown etiology in the majority of cases. According to recent reports, incidence rate of these anomalies is rising in different countries. This study aimed to evaluate the prevalence of external genital anomalies and possible underlying factors in male newborns.

Method(s): This cross-sectional study was conducted on all the male neonates born in Motahari Hospital of Urmia during October 2009-June 2010. Neonatal screening results and demographic data were recorded separately in a questionnaire for each neonate. Data analysis was performed in SPSS V.20 using descriptive statistics (mean, frequency, percentage, and standard deviation) and Chi-square test to evaluate the correlations between quantitative variables.

Result(s): In this study, external genital anomalies were identified in 83 male neonates (8.3%). Moreover, 59 infants (5.9%) had cryptorchidism, 18 (1.8%) had hypospadias and 12 infants (1.2%) had microphallus. Moreover, a significant correlation was observed between the incidence of cryptorchidism and maternal comorbidities, low birth weight, prematurity, maternal gestational hypertension, and diabetes mellitus.

Conclusion(s): According to the results of this study, cryptorchidism is the most prevalent external genital anomaly in male newborns, followed by hypospadias. Occurrence of these anomalies is associated with several risk factors, such as prematurity, low birth weight, maternal diseases (e.g., diabetes and hypertension), and use of medications during pregnancy. In addition, cryptorchidism had a significant correlation with smoking habits, and further studies with larger sample sizes are required in this regard.

Status

Embase

Institution

(Pakniyat) Student Research Committee, Emergency Department, Arak University of Medical Science, Arak, Iran, Islamic Republic of (Fallah) Nephrology and Kidney Transplant Research Center, Urmia University of Medical Sciences, Urmia, Iran, Islamic Republic of

(Fakour) Department of Pediatrics, Urmia University of Medical Sciences, Urmia, Iran, Islamic Republic of

(Moloudi, Khezri, Masoudi) Urmia University of Medical Sciences, Urmia, Iran, Islamic Republic of

Publisher

Mashhad University of Medical Sciences (P.O. Box: 445, Daneshgah Avenue, Mashhad, Iran, Islamic Republic of)

Year of Publication

2016

520.

Can hypertrophy of the contralateral testis predict the absence of a viable testis in infancy with cryptorchidism: A prospective analysis.

Son H.S., Lee Y.S., Im Y.J., Kim S.W., Chi B.H., Han S.W.

Embase

PLoS ONE. 11 (3) (no pagination), 2016. Article Number: e0151528. Date of Publication: March 2016.

[Article]

AN: 609296315

This prospective study aimed to evaluate whether Contralateral compensatory testicular hypertrophy (CTH) is valid as a predictive tool for a non-viable testis in children aged between 6

and 18 months, and whether CTH is affected by mini-puberty. Seventy-two testes from 60 boys aged between 6 and 18 months were categorized into three groups: 24 testes contralateral to surgically removed non-viable testes (NVTs), 24 testes contralateral to surgically corrected undescended testes (UDTs), and 24 testes from a normal controls. Contralateral testicular length and volume were measured with ultrasonography and compared among the groups. Group 1 (NVT) had a significantly longer length and larger volume than group 2 (UDT). The length and volume of each group among three developmental periods (6-10, 10-14, and 14-18 months) were also analyzed. In the controls, the length was significantly larger at 6-10 months than at 10-14 months in accordance with previously reported changes in testicular size due to the effect of "mini-puberty." The volume of controls showed a similar pattern, though without statistical significance. However, this pattern was not observed in the NVT and UDT groups. A receiver operating curve revealed that a testicular length of 16.1 mm or a volume of 0.59 ml had the highest sensitivity and specificity for predicting NVTs. The CTH was also found to be valid as a predictive tool for a NVT in children of ages 6 to 18 months, as the effect of mini-puberty appeared to be absent in the NVT and UDT groups. However, the cut-off values were less than those of previous reports. The proper cut-off level according to the age and measurement method should be applied in this developmental period.

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Institution

(Son, Lee, Im, Kim, Chi, Han) Department of Urology, Urological Science Institute, Yonsei University, College of Medicine, Seoul, South Korea

Publisher

Public Library of Science (E-mail: plos@plos.org)

Year of Publication

2016

521.

Organic food consumption during pregnancy and hypospadias and cryptorchidism at birth: The Norwegian mother and child cohort study (MoBa).

Brantsaeter A.L., Torjusen H., Meltzer H.M., Papadopoulou E., Hoppin J.A., Alexander J., Lieblein G., Roos G., Holten J.M., Swartz J., Haugen M.

Embase

Environmental Health Perspectives. 124 (3) (pp 357-364), 2016. Date of Publication: March 2016.
[Article]

AN: 608769098

Background: The etiologies of the male urogenital anomalies hypospadias and cryptorchidism remain unclear. It has been suggested that maternal diet and environmental contaminants may affect the risk of these anomalies via placental or hormonal disturbances.

Objective(s): We examined associations between organic food consumption during pregnancy and prevalence of hypospadias and cryptorchidism at birth.

Method(s): Our study includes 35,107 women participating in the Norwegian Mother and Child Cohort Study (MoBa) who delivered a singleton male infant. Information about use of six groups of organically produced food (vegetables, fruit, bread/cereal, milk/dairy products, eggs, and meat) during pregnancy was collected by a food frequency questionnaire. Women who indicated that they sometimes, often, or mostly consumed organic foods in at least one of the six food groups were classified as organic food consumers in analyses. Hypospadias and cryptorchidism diagnoses were retrieved from the Medical Birth Registry of Norway. We estimated odds ratios (ORs) and 95% confidence intervals (CIs) using multiple logistic regression.

Result(s): Seventy-four male newborns were diagnosed with hypospadias (0.2%), and 151 with cryptorchidism (0.4%). Women who consumed any organic food during pregnancy were less likely to give birth to a boy with hypospadias (OR = 0.42; 95% CI: 0.25, 0.70, based on 21 exposed cases) than women who reported they never or seldom consumed organic food.

Associations with specific organic foods were strongest for vegetable (OR = 0.36; 95% CI: 0.15, 0.85; 10 exposed cases) and milk/dairy (OR = 0.43; 95% CI: 0.17, 1.07; 7 exposed cases) consumption. No substantial association was observed for consumption of organic food and cryptorchidism.

Conclusion(s): Consumption of organically produced foods during pregnancy was associated with a lower prevalence of hypospadias in our study population. These findings were based on small numbers of cases and require replication in other study populations.

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Institution

(Brantsaeter, Torjusen, Meltzer, Papadopoulou, Haugen) Division of Environmental Medicine, Department of Exposure and Risk Assessment, Norwegian Institute of Public Health, Oslo, Norway (Torjusen, Roos) National Institute for Consumer Research (SIFO), Oslo, Norway (Hoppin) Center for Human Health and the Environment, Department of Biological Sciences, North Carolina State University, Raleigh, NC, United States

(Alexander) Office of the Director-General, Norwegian Institute of Public Health, Oslo, Norway

(Lieblein) Department of Plant Sciences, Norwegian University of Life Sciences, As, Norway

(Holten) Oikos-Organic Norway, Oslo, Norway

(Swartz) Department of Neuroscience Child and Adolescent Psychiatry, Uppsala University, Uppsala, Sweden

(Swartz) Vidarkliniken, Jarna, Sweden

Publisher

Public Health Services, US Dept of Health and Human Services

Year of Publication

2016

522.

A combined preperitoneal and inguinal approach for redo orchiopexy.

Sfoungaris D., Mouravas V.

Embase

Journal of Pediatric Urology. 12 (1) (pp 43.e1-43.e6), 2016. Date of Publication: 01 Feb 2016.

[Article]

AN: 605899311

Introduction Redo orchiopexy involves a hazardous dissection inside the inguinal canal (IC) where scar tissue encircles the testicular vessels (TesV), vas deferens (VD), and the testis. **Objective** The aim was to describe and evaluate a combined preperitoneal and inguinal approach (CPI) through a single cutaneous incision and accomplish this task as safely as possible, at the same time permitting additional maneuvers for cord lengthening. **Material and methods** We prospectively studied eight patients aged from 2.7 to 13 years (mean 7 years) reoperated for failed orchiopexy using the CPI approach. Reoperation took place 12 months to 11 years (mean 4.4 years) after the initial operation. Through a single transverse skin crease incision over the IC, at the level of the deep inguinal ring (DIR), we gained access to both the preperitoneal space

(PPS) and the IC. We first entered the PPS, the peritoneum is retracted, and the VD and TesV are seen entering the DIR. They are gently dissected and two vascular lacets are passed around them. We introduce the backside of an anatomic forceps through the DIR, just under the anterior IC wall, until it is impeded by adhesions and then incise above the forceps, thus protecting the cord structures. Through that opening we transpose one of the lacets that encircle the VD and TesV and exercise traction upon them (figure, 1), revealing step by step the points where adhesiolysis must take place (figure, 2). The testis is dissected last of all and delivered back, through the DIR, into the PPS. There, the TesV and VD are freed from their retroperitoneal attachments (figure, 3). Finally, the testis is fixed into a Dartos pouch. Results In all cases the testes were relocated to the scrotum without any mishaps. All testes were inside the scrotum at first month examination and with good consistency. At 6 months, one testis ascended at mid-scrotum. At 2 years they all retained their position and their good standing, according to clinical and ultrasonographic findings. Discussion Several procedures of redo orchioepexies have been published so far, most of them rely on the surgeon's dexterity for good results. The CPI procedure offers a practical maneuver to protect the cord elements while dissecting and also exposes all the regions where dissection will offer lengthening of the cord. Conclusion Our results have demonstrated that the CPI can be considered as a safe and efficient procedure for redo orchioepexy.

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Status

Embase

Author NameID

Sfoungaris, Dimitrios; ORCID: <http://orcid.org/0000-0001-5567-5118>

Institution

(Sfoungaris, Mouravas) Second Department of Pediatric Surgery, Aristotle University of Thessaloniki, General Hospital Papageorgiou, Ring Road Thessaloniki, Nea Efkarpia 56403, Greece

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Year of Publication

2016

Testicular hypertrophy as a predictor for contralateral monorchism: Retrospective review of prospectively recorded data.

Hodhod A., Capolicchio J.P., Jednak R., El-Sherbiny M.

Embase

Journal of Pediatric Urology. 12 (1) (pp 34.e1-34.e5), 2016. Date of Publication: 01 Feb 2016.

[Article]

AN: 605755218

Objectives Testicular hypertrophy has previously been evaluated as a predictor of monorchism. However, its implication in clinical practice is not well evaluated. The aim of the present study was to examine its value in planning the operative time. **Patients and methods** Medical charts of prospectively recorded data of 76 consecutive patients with unilateral impalpable testis from 2011 to 2014 were reviewed at the present institute. Inclusion criteria included prepubertal patients with non-palpable testes by examination under anesthesia. Contralateral testes were prospectively measured using a Takihara orchidometer. Orchiectomy or orchiopexy was performed according to the viability of the undescended testis (UDT). Collected data included age of surgery, contralateral testicular size, surgical time and laparoscopic findings. A ROC curve was used to define the best cut-off volume of the contralateral testis that can predict ipsilateral testicular viability. The Student's t-test was used to examine if this cut-off volume would be useful in allocating the operative time. **Results** Of 76 patients, four palpable testes by examination under anesthesia were excluded. The remaining 72 patients were included in the study. Ipsilateral normal viable testes were found in 26 (36.1%) patients, while 46 (63.9%) had non-viable testes (testicular nubbins or vanishing testes) (Figure). A contralateral testicular volume >2 ml was significantly predictive for monorchism with 71.7% sensitivity and 100% specificity (P < 0.001). The mean operative time for management of UDT with a contralateral size >2 ml was 50 min, which was significantly shorter than that for UDT with a contralateral size ≤2 ml, which was 88 min (P < 0.001). **Discussion** In previously published reports, the cut-off value for testicular hypertrophy that predicts monorchism greatly varied. This is likely due to the different methods used for testicular measurements that make it impractical to make a direct comparison. The usefulness of predicting monorchism before surgery has not previously been used as a guide for allocating operative time in the management of a unilateral non-palpable testicle. This study had some limitations, including a relatively small sample size and involvement of different surgeons, which may have affected the operative time. **Conclusion** Using the cut-off volume of a contralateral testis >2 ml as a predictor for monorchism can reduce the allocated operative time by approximately one third.

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Status

Embase

Institution

(Hodhod, Capolicchio, Jednak, El-Sherbiny) Department of Pediatric Surgery, Urology, McGill University, 2300 Tupper, Montreal, QC H3H1P3, Canada

Publisher

Elsevier Ltd

Year of Publication

2016

524.

Bisphenol A and other phenols in human placenta from children with cryptorchidism or hypospadias.

Fernandez M.F., Arrebola J.P., Jimenez-Diaz I., Saenz J.M., Molina-Molina J.M., Ballesteros O., Kortenkamp A., Olea N.

Embase

Reproductive Toxicology. 59 (pp 89-95), 2016. Date of Publication: January 01, 2016.

[Article]

AN: 608149157

Embryo-foetal exposure to low doses of endocrine disrupting chemicals (EDCs) has been related to reproductive tract diseases in experimental animals but not convincingly in human populations. The aim of this case-control study was to explore the relationship between exposure to non-persistent EDCs during pregnancy and male genital development. Exposure to bisphenol-A (BPA), benzophenones (BPs) [BP-1, BP-2, BP-3, BP-6, BP-8 and 4-hydroxybenzophenone (4-OH-BP),] and parabens (PBs) [methyl-, ethyl-, propyl- and butyl-PB] was analyzed by means of ultra-high performance liquid chromatography-tandem mass spectrometry in placenta samples from a subsample of 28 cases and 51 healthy controls nested in a cohort of newborns recruited between 2000 and 2002. The multivariable regression analyses indicated a statistically significant association between exposure to BPA and propyl-PB and the risk of malformations [adjusted odd ratio (95% CIs) in the third tertile of exposure: 7.2 (1.5-35.5) and 6.4 (1.2-35.5) for BPA and propyl-PB, respectively].

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Institution

(Fernandez, Arrebola, Jimenez-Diaz, Saenz, Molina-Molina, Olea) Instituto de Investigacion Biosanitaria (ibs.GRANADA), University of Granada, San Cecilio University Hospital, Granada 18071, Spain (Fernandez, Arrebola, Molina-Molina, Olea) CIBER en Epidemiologia y Salud Publica (CIBERESP), Madrid, Spain

(Jimenez-Diaz, Ballesteros) Research Group of Analytical Chemistry and Life Sciences, Department of Analytical Chemistry, University of Granada, Granada, Spain

(Kortenkamp) Institute of the Environment, Health and Societies, Brunel University, Kingston Lane, Uxbridge UB8 3PH, United Kingdom

Publisher

Elsevier Inc. (E-mail: usjcs@elsevier.com)

Year of Publication

2016

525.

Differences in the development of the processus vaginalis between children with undescended testis and inguinal hernia.

Sfoungaris D., Mouravas V., Koletsa T., Lambropoulos V., Kostopoulos I., Spyridakis I.

Embase

Journal of Clinical Neonatology. 5 (1) (pp 31-34), 2016. Date of Publication: January-March 2016.

[Article]

AN: 607895750

Background: It has been shown in several investigations that smooth muscle cells (SMCs) are present on the patent processus vaginalis (PV) peritonei in cases of inguinal hernia (IH) preventing its obliteration. The PV fails to obliterate in cases of undescended testis (UT) as well, but without causing herniation.

Material(s) and Method(s): We conducted a case control study in order to compare the status of the SMCs present on the PV in UT and IH cases and correlate it with the clinical outcome of herniation. Specimens were harvested from the hernia sacs of 26 boys with IH aged from 2 days to 16 years (mean 44.31 months) and the PV of 14 children with UT, aged from 13 months to 13 years (mean 30.28 months). They were examined histologically and immunohistochemically for

markers of mature SMC such as smooth muscle actin (SMA), desmin and h-caldesmon as well as for vimentin, an immature SMC marker.

Result(s): The expression of SMA, desmin, and h-caldesmon was present in all cases of IH and UT. Vimentin was expressed in 13 out of 14 (93%) UT specimens and in 12 out of 26 IH sacs (46.1%), (P = 0.0102). In IH and UT cases, when vimentin was not expressed, SMCs were organized in bundles.

Conclusion(s): The SMCs on the PV in UT cases reach a more advanced stage of dedifferentiation that corresponds to a status more close to that of the natural obliteration, compared to IH cases, preventing herniation to occur.

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Status

Embase

Institution

(Sfoungaris, Mouravas, Lambropoulos, Spyridakis) Second Department of Pediatric Surgery, Aristotle University of Thessaloniki, General Hospital Papageorgiou, Thessaloniki Ring Road, Nea Efkarpiia, Thessaloniki 56403, Greece (Koletsis, Kostopoulos) Department of Pathology, Aristotle University of Thessaloniki, University Campus, Thessaloniki 54124, Greece

Publisher

Medknow Publications (B9, Kanara Business Centre, off Link Road, Ghatkopar (E), Mumbai 400 075, India)

Year of Publication

2016

526.

Perfluorooctane sulfonate concentrations in amniotic fluid, biomarkers of fetal leydig cell function, and cryptorchidism and hypospadias in Danish boys (1980-1996).

Toft G., Jonsson B.A., Bonde J.P., Norgaard-Pedersen B., Hougaard D.M., Cohen A., Lindh C.H., Ivell R., Anand-Ive R., Lindhard M.S.

Embase

Environmental Health Perspectives. 124 (1) (pp 151-156), 2016. Date of Publication: January 2016.

[Article]

AN: 607485772

Background: Exposure to perfluorooctane sulfonate (PFOS) may potentially disturb fetal Leydig cell hormone production and male genital development.

Objective(s): We aimed to study the associations between levels of amniotic fluid PFOS, fetal steroid hormone, and insulin-like factor 3 (INSL3) and the prevalence of cryptorchidism and hypospadias.

Method(s): Using the Danish National Patient Registry, we selected 270 cryptorchidism cases, 75 hypospadias cases, and 300 controls with stored maternal amniotic fluid samples available in a Danish pregnancy-screening biobank (1980-1996). We used mass spectrometry to measure PFOS in amniotic fluid from 645 persons and steroid hormones in samples from 545 persons. INSL3 was measured by immunoassay from 475 persons. Associations between PFOS concentration in amniotic fluid, hormone levels, and genital malformations were assessed by confounder-adjusted linear and logistic regression.

Result(s): The highest tertile of PFOS exposure (> 1.4 ng/mL) in amniotic fluid was associated with a 40% (95% CI: -69, -11%) lower INSL3 level and an 18% (95% CI: 7, 29%) higher testosterone level compared with the lowest tertile (< 0.8 ng/mL). Amniotic fluid PFOS concentration was not associated with cryptorchidism or hypospadias.

Conclusion(s): Environmental PFOS exposure was associated with steroid hormone and INSL3 concentrations in amniotic fluid, but was not associated with cryptorchidism or hypospadias in our study population. Additional studies are needed to determine whether associations with fetal hormone levels may have long-term implications for reproductive health.

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Status

Embase

Institution

(Toft) Department of Occupational Medicine, Aarhus University Hospital, Aarhus, Denmark

(Toft) Department of Clinical Epidemiology, Aarhus University Hospital, Aarhus, Denmark

(Jonsson, Lindh) Division of Occupational and Environmental Medicine, Lund University, Lund, Sweden

(Bonde) Department of Occupational and Environmental Medicine, Copenhagen University Hospital, Bispebjerg, Copenhagen, Denmark

(Norgaard-Pedersen, Hougaard, Cohen) Danish Center for Neonatal Screening, Department of Clinical Biochemistry and Immunology, Statens Serum Institute, Copenhagen, Denmark

(Ivell, Anand-Ive) School of Biosciences, University of Nottingham, Nottingham, United Kingdom

(Lindhard) Department of Pediatrics, Regional Hospital of Randers, Randers, Denmark

(Lindhard) Perinatal Epidemiology Research Unit, Department of Pediatrics, Aarhus University Hospital, Skejby, Denmark

Publisher

Public Health Services, US Dept of Health and Human Services

Year of Publication

2016

527.

Selective use of laparoscopy in nonpalpable undescended testes.

Jamalalail Y.A., Guerra L.A., Leonard M.P.

Embase

Urology Annals. 8 (1) (pp 81-83), 2016. Date of Publication: January 2016.

[Article]

AN: 607439503

Introduction: Approximately, 20% of undescended testes (UDT) are nonpalpable. Surgical management of the nonpalpable testis comprises laparoscopy. The aim of this study was to determine if ultrasonography can be used as a preoperative tool to localize the nonpalpable inguinal testis, eliminating the need for laparoscopy.

Method(s): We identified 46 patients diagnosed with nonpalpable UDT between 2007 and 2012 who underwent an inguino-scrotal ultrasound preoperatively. We analyzed correlations between radiological and surgical findings.

Result(s): A total of 46 patients (53 UDT), median age 14 months (quartile 1st: 7; 3rd: 80) were included. Ultrasound localized the testis as intracanalicular in 24/53 (45.2%), intraabdominal in 10/53 (18.8%), scrotal in 1/53 (1.8%), and could not localize 18/53 (33.9%) testes. In 35/53 (66%) testes, the ultrasound location correlated with the surgical findings ($P < 0.001$). Ultrasound detection showed 96% sensitivity and 56% specificity for intracanalicular testes.

Conclusion(s): The use of preoperative ultrasound in this series was helpful in identifying the location of nonpalpable testes in children. In particular, the ultrasound finding of an intracanalicular testis may preclude the need for laparoscopy.

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Status

Embase

Institution

(Jamalalail) Department of Surgery, Division of Urology, King Fahad Medical City, Riyadh, Saudi Arabia (Guerra, Leonard) Department of Surgery, Division of Urology, Childrens Hospital of Eastern Ontario, University of Ottawa, Ottawa, ON, Canada

Publisher

Medknow Publications (B9, Kanara Business Centre, off Link Road, Ghatkopar (E), Mumbai 400 075, India)

Year of Publication

2016

528.

The value of positive Oct3/4 and D2-40 immunohistochemical expression in prediction of germ cell neoplasia in prepubertal boys with cryptorchidism.

Clasen-Linde E., Kvist K., Cortes D., Thorup J.

Embase

Scandinavian Journal of Urology. 50 (1) (pp 74-79), 2016. Date of Publication: 02 Jan 2016.

[Article]

AN: 606525822

Objective: Intratubular germ cell neoplasia (ITGCN) is a precursor to testicular germ cell cancer. Adult germ cell cancer immunohistochemical markers fail to detect ITGCN in prepubertal boys with congenital cryptorchidism, because positive immunohistochemistry is commonly seen below 18 months old, where most orchiopexies are performed. The aim of the study was to evaluate the ability of Oct3/4 and D2-40 immunohistochemical markers to detect ITGCN in boys older than 2 years with cryptorchidism.

Material(s) and Method(s): Histological sections from 309 testicular biopsies from 234 boys aged 1 month to 14 years, 6 months operated on for cryptorchidism were incubated with primary antibodies including anti-placental-like alkaline phosphatase, anti-Oct3/4, anti-C-kit and anti-D2-40 receptor.

Result(s): One 3-year, 8-month-old boy with 45X/46XY disorder of sexual development had ITGCN and all positive markers. Besides this case, none of the 192 testes except one from boys older than 2 years had any Oct3/4-or D2-40-positive germ cells identified. The germ cells of the

right testis from a 3-year, 7-month-old boy had weak Oct3/4 expression but were D2-40 negative. The prevalences of Oct3/4-and D2-40-positive staining of germ cells in testicular biopsies were, for each age group: < 6 months, 100% and 50%; 6 months to < 1 year, 65% and 16%; 1 to < 2 years, 15% and 3%; and 2 years to < 14 years, 6 months, 2% and 1%, respectively.

Conclusion(s): Oct3/4 and D2-40 immunohistochemical markers may be beneficial in detecting ITGCN in boys older than 2 years with cryptorchidism. Even when immunohistochemistry is applied, prepubertal ITGCN is so rarely demonstrated in cryptorchid testes that it is not plausible that ITGCN generally originates during fetal development in cryptorchidism.

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Status

Embase

Institution

(Clasen-Linde) Department of Pathology, Rigshospitalet, Copenhagen, Denmark (Kvist, Cortes, Thorup) Department of Pediatric Surgery, Rigshospitalet, Copenhagen DK-2100, Denmark

(Cortes, Thorup) Faculty of Health and Medical Science, University of Copenhagen, Copenhagen, Denmark

(Cortes) Section of Endocrinology, Department of Pediatrics, Hvidovre Hospital, Copenhagen, Denmark

Publisher

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Year of Publication

2016

529.

Subfertility and risk of testicular cancer in the EPSAM case-control study.

Grasso C., Zugna D., Fiano V., Rodriguez N.R., Maule M., Gillio-Tos A., Ciuffreda L., Lista P., Segnan N., Merletti F., Richiardi L.

Embase

PLoS ONE. 11 (12) (no pagination), 2016. Article Number: e0169174. Date of Publication:

December 2016.

[Article]

AN: 613932122

Background/objectives It has been suggested that subfertility and testicular cancer share genetic and environmental risk factors. We studied both subfertility and the strongest known testicular cancer susceptibility gene, the c-KIT ligand (KITLG), whose pathway is involved in spermatogenesis. **Methods** The EPSAM case-control study is comprised of testicular cancer patients from the Province of Turin, Italy, diagnosed between 1997 and 2008. The present analysis included 245 cases and 436 controls from EPSAM, who were aged 20 years or older at diagnosis/recruitment. The EPSAM questionnaire collected information on factors such as number of children, age at first attempt to conceive, duration of attempt to conceive, use of assisted reproduction techniques, physician-assigned diagnosis of infertility, number of siblings, and self-reported cryptorchidism. Genotyping of the KITLG single nucleotide polymorphism (SNP) rs995030 was performed on the saliva samples of 202 cases and 329 controls. **Results** Testicular cancer was associated with the number of children fathered 5 years before diagnosis (odds ratio (OR) per additional child: 0.78, 95% confidence interval (CI): 0.58-1.04) and sibship size (OR per additional sibling: 0.76, 95% CI: 0.66-0.88). When considering the reproductive history until 1 year before diagnosis, attempting to conceive for at least 12 months or fathering a child using assisted reproduction techniques was not associated with the risk of testicular cancer, nor was age at first attempt to conceive or physician-assigned diagnosis of infertility. The SNP rs995030 was strongly associated with risk of testicular cancer (per allele OR: 1.83; 95%CI: 1.26-2.64), but it did not modify the association between number of children and the risk of testicular cancer. **Conclusion** This study supports the repeatedly reported inverse association between number of children and risk of testicular cancer, but it does not find evidence of an association for other indicators of subfertility.

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PMC Identifier

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Status

Embase

Institution

(Grasso, Zugna, Fiano, Rodriguez, Maule, Gillio-Tos, Merletti, Richiardi) Cancer Epidemiology Unit-CeRMS, Department of Medical Sciences, University of Turin and CPO Piedmont, Turin, Italy (Ciuffreda, Lista) Medical Oncology Division 1, University Hospital Citta Della Salute E Della Scienza, Turin, Italy

(Segnan) Department of Cancer Screening and Unit of Cancer Epidemiology, WHO Collaborative Center for Cancer Early Diagnosis and Screening, CPO Piedmont and University Hospital Citta Della Salute E Della Scienza, Turin, Italy

Publisher

Public Library of Science (E-mail: plos@plos.org)

Year of Publication

2016

530.

Transitional Urology for Male Adolescents: What Adult Urologists Should Know.

Suson K.D.

Embase

Current Urology Reports. 17 (10) (no pagination), 2016. Article Number: 71. Date of Publication: 01 Oct 2016.

[Review]

AN: 611690562

In recent years, there has been increasing interest in transitional urology, or how to best prepare patients with major congenital urologic diseases, such as bladder exstrophy and neuropathic bladder, to manage their own health care with adult urologists. However, common pediatric urologic conditions may be encountered by the adult urologist with more regularity. This review focuses on three relatively common conditions which may be identified in childhood, the consequences from which a patient may seek help from an adult urologist: cryptorchidism, varicocele, and Klinefelter syndrome.

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Institution

(Suson) Children's Hospital of Michigan, 3901 Beaubien, Detroit, MI 48201, United States

Publisher

Current Medicine Group LLC 1 (E-mail: info@phl.cursci.com)

Year of Publication

2016

531.

Testicular prostheses in children: Is earlier better?.

Peycelon M., Rossignol G., Muller C.O., Carricaburu E., Philippe-Chomette P., Paye-Jaouen A., El Ghoneimi A.

Embase

Journal of Pediatric Urology. 12 (4) (pp 237.e1-237.e6), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 610626344

Introduction The absence of a testis occurs for various reasons in children, but testicular prosthesis implantation in children is uncommon. The optimal time for prosthesis placement is still unclear, and its complication rate has been poorly studied in children. **Objective** The aim of this study was to determine the risk factors of complications in cases of testicular prosthesis implantation in children. **Study design** A monocentric, retrospective review was performed of children implanted with a testicular prosthesis between 2008 and 2014. All implantations were performed through an inguinal incision with a standardized procedure. Children were divided into two groups depending on the interval after orchiectomy: (A) early implantation (delay between surgeries <1 year); and (B) delayed surgeries (delay \geq 1 year). **Statistical analysis** was performed with Student and Fisher tests. **Results** Twenty-six patients (A, 15; B, 11) had a total of 38 testicular prostheses placements. Mean follow-up was 36.2 months. First surgery was performed at the mean age of 11.8 years (range 0-17.9) (A, 14.1; B, 8.1; $P = 0.01$) and testicular prosthesis implantation at the mean age of 14.7 years (range 9-18) (A, 14.3; B, 14.6) with a mean delay of 36.1 months (A, 1.3; B, 80.3). Indications were mainly spermatic cord torsion (27%), bilateral anorchia (27%), and testicular atrophy after cryptorchidism surgery (19.2%). **Complications** (10.5%) included two cases of extrusion, one infection and one migration. Patient 1 had a history of acute lymphoblastic leukemia with testicle relapse 2 years after induction therapy. High-dose chemotherapy, total body irradiation and bilateral orchiectomies were performed, and bilateral prostheses were implanted 12 years after the end of chemotherapy. **Complications** happened 85 days after surgery. Patient 2 was followed-up for a proximal hypospadias. The tunica vaginalis flap, which was used during a redo urethroplasty, lead to testicular atrophy. Thirteen years after the last penile surgery, a testicular prosthesis was placed through an inguinal incision, and extrusion occurred 203 days after surgery. Bacterial cultures of the prostheses were sterile and histological review showed no sign of granuloma or graft

rejection. The complication rate was significantly higher if the delay between the two surgeries exceeded 1 year ($P = 0.01$). Indications of orchiectomy, prior scrotal incision, and prosthesis size were not risk factors. Conclusions Testicular prosthesis implantation was relatively safe in a pediatric cohort. The complication rate was significantly higher if the delay between the orchiectomy and the prosthetic placement exceeded 1 year. These results suggest that reducing the delay between orchiectomy and prosthesis implantation may lead to fewer complications.

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Institution

(Peycelon, Rossignol, Muller, Carricaburu, Philippe-Chomette, Paye-Jaouen, El Ghoneimi)

Department of Pediatric Surgery and Urology, Robert-Debre University Hospital, Assistance-Publique Hopitaux de Paris, Universite Paris Diderot, Sorbonne Paris Cite, Paris, France

Publisher

Elsevier Ltd

Year of Publication

2016

532.

Prospective study of histological and endocrine parameters of gonadal function in boys with cryptorchidism.

Verkauskas G., Malcius D., Eidukaite A., Vilimas J., Dasevicius D., Bilius V., Hadziselimovic F.

Embase

Journal of Pediatric Urology. 12 (4) (pp 238.e1-238.e6), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 613343998

Introduction A transient increase in gonadotropins and testosterone during mini-puberty causes gonocytes to differentiate into Ad spermatogonia, which establish male germ cell memory and male-specific DNA methylation pathways. Over half of patients with unilateral cryptorchidism and the majority of patients with bilateral cryptorchidism display an abnormal spermiogram, which

indicates that unilateral cryptorchidism is a bilateral disease; therefore, it represents a serious andrological problem. The aim of this study was to evaluate relationships between hormonal parameters and testicular biopsy findings in boys with cryptorchidism. Method Seventy-one boys (median age 15 months; range 7-65 months) who underwent orchidopexy (24% had bilateral cryptorchidism) were tested for serum LH, FSH, and inhibin B. With ipsilateral testis biopsy histology, we determined the tubular fertility index (TFI), Ad spermatogonia counts, and Ad/tubular index (Ad/T). We compared age groups (<18 vs. >18 months old); groups with and without Ad spermatogonia; groups with unilateral and bilateral cryptorchidism; and extreme groups with high infertility risk (HIR; n = 12; TFI <0.2; Ad/T = 0) and low infertility risk (LIR; n = 9; TFI >0.9; Ad/T>0.02). Results Of the specimens, 38% had no Ad spermatogonia. Age was significantly negatively correlated with TFI and Ad/T, but positively correlated with FSH. Median LH values were significantly higher in LIR than in HIR groups. Unilateral and bilateral cryptorchidism showed similar TFI, Ad/T, and hormone concentrations. The areas under ROC curves for FSH, LH, and inhibin B (0.66, 0.601, and 0.599, respectively) showed low diagnostic value for predicting HIR (no Ad spermatogonia). Conclusion Our observation of lower plasma LH levels in the group with the most pronounced testicular pathology was the opposite of what we would have expected if testicular pathological changes were caused by a primary gonadal defect. Therefore, low plasma LH levels in the HIR group confirmed the notion that this group of patients with cryptorchidism had hypogonadotropic hypogonadism. The estimated incidence of defective mini-puberty in boys with cryptorchidism could be as high as 50%. Testicular biopsies from boys with cryptorchidism lacked Ad spermatogonia. Fertility parameters worsened with age. Significantly lower basal LH in the HIR group indicated hypogonadotropic hypogonadism. Serum hormone levels could not predict histological biopsy findings.

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Status

Embase

Author NameID

Vilimas, Juozas; ORCID: <http://orcid.org/0000-0002-4631-0720>

Institution

(Verkauskas, Vilimas, Bilius) Faculty of Medicine, Vilnius University, Vilnius, Lithuania (Malcius)

Lithuanian University of Health Sciences, Kaunas, Lithuania

(Eidukaite) State Research Institute Centre for Innovative Medicine, Vilnius, Lithuania

(Dasevicius) National Centre of Pathology, Vilnius, Lithuania

(Hadziselimovic) Institute for Cryptorchidism Research, Kindermedizinisches Zentrum, Liestal,

Switzerland

Publisher
Elsevier Ltd
Year of Publication
2016

533.

Acquired cryptorchidism: More harm than thought?.

Promm M., Schroder A., Neissner C., Eder F., Rosch W.H., Schroder J.

Embase

Journal of Pediatric Urology. 12 (4) (pp 236.e1-236.e6), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 613342841

Introduction Acquired cryptorchidism (AC) has been recognized as a subgroup of undescended testes (UDT). There is growing evidence that the compromising effect equals that of congenital UDT (cUDT). This prospective study included an extensive histological examination of biopsies taken from AC patients. Patients and methods From August 2013 to December 2014, 21 boys (3-12 years of age) underwent testicular biopsy during orchiopexy for AC. Patient and family histories were taken. The amount of germ cells (GC) per tubule (T) and the amount of adult dark spermatogonia (Ad-S) per T were determined by resin semi-thin sections examination. The samples were also scanned for signs of malformation. Immunohistochemical stains were performed as markers for atypical germ cells. Results Four (19%) boys were born prematurely, two (9.5%) were small for gestational age (SGA), and nine (43%) had a positive family history of UDT. The median of GC/T was 1.06 in boys <9 years, and 0.60 in boys >=9 years. The median of Ad-S/T was 0.02 in boys <9 years and 0.01 in boys >=9 years. There were no signs for malformation and no atypical cells. The immunohistochemical stains were negative in all specimens. Conclusions Prematurity, SGA, and a positive family history appeared to be predictors for AC. Extensive histopathological examination of AC revealed a significant reduction of germ cell count and fertility markers, comparable with that in cUDT. The alterations were more severe in boys aged >=9 years. It is unclear as to whether or not this was possibly caused by a longer duration of inguinal position, but this finding suggests that routine checks of testicular position throughout childhood are needed, and that there is a cause for continued efforts in

educating parents and primary care physicians regarding AC. Current data support the notion of surgical correction once the diagnosis is made. [Table presented]

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Status

Embase

Author NameID

Promm M.; ORCID: <http://orcid.org/0000-0002-7645-1591>

Institution

(Promm, Schroder, Neissner, Rosch) Department of Paediatric Urology, Klinik St. Hedwig, University Medical Center of Regensburg, Steinmetzstr. 1-3, Regensburg 93049, Germany
(Eder, Schroder) Department of Pathology, University Medical Center of Regensburg, Franz-Josef-Straus-Allee 11, Regensburg 93053, Germany

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2016

534.

Testicular atrophy following paediatric primary orchidopexy: A prospective study.

Durell J., Johal N., Burge D., Wheeler R., Griffiths M., Kitteringham L., Stanton M., Manoharan S., Steinbrecher H., Malone P., Griffin S.J.

Embase

Journal of Pediatric Urology. 12 (4) (pp 243.e1-243.e4), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 613342508

Background With the Nordic consensus statement advocating orchidopexy at an earlier age, the present study sought to investigate the outcomes of primary paediatric orchidopexy at a tertiary UK centre. Objective To prospectively assess testicular atrophy following primary orchidopexy for undescended testes in a paediatric population. Secondary outcomes were complication rates and whether outcomes were dependent on grade of operating surgeon. Study design Prospective

data regarding age at operation, classification of the undescended testis, length of follow-up, and subjective comparison of intraoperative and postoperative testicular volumes compared with the contralateral testis were collected. Testicular atrophy was defined as >50% loss of testicular volume or a postoperative testicular volume <25% of the volume of the contralateral testis. Patients were excluded for incomplete data and follow-up <6 months. Results Data for 234 patients were analysed. Testicular atrophy occurred in 2.6% of cases. There was no reported testicular re-ascent. All secondary acquired cases underwent a previous ipsilateral hernia repair. There was no significant difference in outcomes comparing the grade of surgeon (consultant n = 8, trainee/staff-grade surgeon n = 7-8). There was a trend towards postoperative catch-up growth in approximately one fifth of cases. Discussion Previous studies have reported a testicular atrophy rate of 5%. The present study reported a similar rate of 2.6%. In agreement with a previous publication, it was also found that testicular atrophy was not dependent on the grade of operating surgeon. The mechanism for testicular catch-up growth is not well understood. Animal studies have supported the hypothesis that increased temperature has a detrimental effect on testicular volume. However, follow-up in the present cohort was short (median 6.9 months), making interpretation of this finding difficult. It is acknowledged that clinical palpation alone to determine testicular volume potentially introduces intra-observer and inter-observer error. However, prospective studies using ultrasound to determine testicular volumes following orchidopexy have reported catch-up growth. Conclusion This study represented one of the larger collections of prospective assessments of outcomes following primary orchidopexy. It was acknowledged that subjectively assessing testicular volume is not ideal; however, the data correlated with similar studies. [Table presented]

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Institution

(Durell, Johal, Burge, Wheeler, Griffiths, Kitteringham, Stanton, Manoharan, Steinbrecher, Malone, Griffin) Department of Paediatric Surgery and Urology, Southampton Children's Hospital, University Hospital Southampton, Tremona Road, Southampton, United Kingdom

Publisher

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535.

Single scrotal incision orchiopexy versus the inguinal approach in children with palpable undescended testis: a systematic review and meta-analysis.

Feng S., Yang H., Li X., Yang J., Zhang J., Wang A., Lai X.-H., Qiu Y.

Embase

Pediatric Surgery International. 32 (10) (pp 989-995), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 611645755

Purpose: We performed a systematic review and meta-analysis to compare the efficacy and safety between single-incision, transscrotal orchidopexy, and the traditional inguinal orchidopexy in children.

Method(s): A systematic search of the electronic databases was conducted to identify studies compared the transscrotal orchidopexy (SO) and inguinal orchidopexy (IO) for children.

Parameters, such as operative time, the incidence of patent processus vaginalis, and postoperative complications, including wound infection, testicular atrophy, testicular reascent, hernia, or hydrocele, were pooled and compared by meta-analysis.

Result(s): Among the 1376 children with palpable undescended testes (UDTs) included in the eight studies, 697 had received SO and 679 IO. There were shorter operative times with the SO approach compared with IO. However, no significant difference was found between SO and IO in the incidence of patent processus vaginalis and postoperative complications, including wound infection, testicular atrophy, testicular reascent, and hernia.

Conclusion(s): SO is a safe and effective surgical approach alternative to IO for pediatric UDTs. Compared with IO, SO has the advantage of shorter operative times. Besides, the incidence of postoperative wound infection may be slightly lower in SO. We suggest that SO should be considered as an acceptable option for children with UDTs.

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Embase

Institution

(Feng, Yang, Li, Yang, Zhang, Wang, Qiu) Department of Pediatric Surgery, Hangzhou Children's Hospital, No. 195 Wenhui Rd, Xiacheng district, Hangzhou 310015, China (Lai) Institute of

Inflammation and Diseases, The First Affiliated Hospital of Wenzhou Medical University,
Wenzhou, China

Publisher

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Year of Publication

2016

536.

The undescended testis: Clinical management and scientific advances.

Vikraman J., Hutson J.M., Li R., Thorup J.

Embase

Seminars in Pediatric Surgery. 25 (4) (pp 241-248), 2016. Date of Publication: 01 Aug 2016.

[Article]

AN: 611170961

Undescended testes (UDT), where one or both testes fail to migrate to the base of the scrotum, can be congenital (2-5% of newborn males) or acquired (1-2% of males). The testis may be found in any position along its usual line of descent. Cryptorchidism affects the developing testicular germ cells and increases the risk of infertility and malignancy. Clinical management aims to preserve spermatogenesis and prevent the increased risk of seminoma. Examination to document the testicular position will guide the need for imaging, medical management and the surgical approach to orchidopexy.

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Institution

(Vikraman, Hutson, Li) Douglas Stephens Surgical Group, Murdoch Childrens Research Institute, Melbourne, Victoria, Australia (Vikraman, Hutson) Department of Paediatrics, The University of Melbourne, Melbourne, Victoria, Australia

(Hutson) Department of Urology, The Royal Childrens Hospital, 50 Flemington Rd, Parkville, Victoria 3052, Australia

(Thorup) Department of Paediatric Surgery and Surgical Clinic, Rigshospitalet, Copenhagen, Denmark

(Thorup) Faculty of Health and Medical Sciences, Copenhagen University, Copenhagen, Denmark

Publisher

W.B. Saunders

Year of Publication

2016

537.

Genomic copy number variation association study in Caucasian patients with nonsyndromic cryptorchidism.

Wang Y., Li J., Kolon T.F., Olivant Fisher A., Figueroa T.E., Banihani A.H., Hagerty J.A., Gonzalez R., Noh P.H., Chiavacci R.M., Harden K.R., Abrams D.J., Stabley D., Kim C.E., Sol-Church K., Hakonarson H., Devoto M., Barthold J.S.

Embase

BMC Urology. 16 (1) (no pagination), 2016. Article Number: 62. Date of Publication: 21 Oct 2016.

[Article]

AN: 612833541

Background: Copy number variation (CNV) is a potential contributing factor to many genetic diseases. Here we investigated the potential association of CNV with nonsyndromic cryptorchidism, the most common male congenital genitourinary defect, in a Caucasian population.

Method(s): Genome wide genotyping were performed in 559 cases and 1772 controls (Group 1) using Illumina HumanHap550 v1, HumanHap550 v3 or Human610-Quad platforms and in 353 cases and 1149 controls (Group 2) using the Illumina Human OmniExpress 12v1 or Human OmniExpress 12v1-1. Signal intensity data including log R ratio (LRR) and B allele frequency (BAF) for each single nucleotide polymorphism (SNP) were used for CNV detection using PennCNV software. After sample quality control, gene- and CNV-based association tests were performed using cleaned data from Group 1 (493 cases and 1586 controls) and Group 2 (307 cases and 1102 controls) using ParseCNV software. Meta-analysis was performed using gene-based test results as input to identify significant genes, and CNVs in or around significant genes

were identified in CNV-based association test results. Called CNVs passing quality control and signal intensity visualization examination were considered for validation using TaqMan CNV assays and QuantStudio 3D Digital PCR System.

Result(s): The meta-analysis identified 373 genome wide significant ($p < 5 \times 10^{-4}$) genes/loci including 49 genes/loci with deletions and 324 with duplications. Among them, 17 genes with deletion and 1 gene with duplication were identified in CNV-based association results in both Group 1 and Group 2. Only 2 genes (NUCB2 and UPF2) containing deletions passed CNV quality control in both groups and signal intensity visualization examination, but laboratory validation failed to verify these deletions.

Conclusion(s): Our data do not support that structural variation is a major cause of nonsyndromic cryptorchidism.

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Status

Embase

Institution

(Wang, Olivant Fisher, Stabley, Sol-Church, Barthold) Nemours Biomedical Research, Nemours/Alfred I. DuPont Hospital for Children, Wilmington, DE 19803, United States (Figuroa, Banihani, Hagerty, Gonzalez, Noh, Barthold) Division of Urology, Nemours/Alfred I. DuPont Hospital for Children, Wilmington, DE 19803, United States

(Li, Chiavacci, Harden, Abrams, Kim, Hakonarson) Center for Applied Genomics, Children's Hospital of Philadelphia, Philadelphia, PA 19104, United States

(Kolon) Division of Urology, Children's Hospital of Philadelphia, Philadelphia, PA 19104, United States

(Hakonarson, Devoto) Division of Genetics, Children's Hospital of Philadelphia, Philadelphia, PA 19104, United States

(Hakonarson, Devoto) Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, United States

(Devoto) Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, United States

(Devoto) Department of Molecular Medicine, Sapienza University, Rome, Italy

(Gonzalez) Auf der Bult Kinder- und Jugendkrankenhaus, Hannover, Germany

(Noh) Division of Pediatric Urology, Cincinnati Children's Hospital Medical Center, Cincinnati, OH, United States

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Year of Publication

2016

538.

The role of ultrasound in the management of undescended testes before and after orchidopexy -
An update.

Jedrzejewski G., Wieczorek A.P., Osemlak P., Nachulewicz P.

Embase

Medicine (United States). 95 (51) (pp e5731), 2016. Date of Publication: 2016.

[Article]

AN: 613914998

The aim of this study was to evaluate the testicular volume and structure using ultrasound (US) before and up to 3 years after orchidopexy in children with different age. A total of 128 patients underwent orchidopexy for undescended testes. Afterwards, patients were invited for annual follow-up and control scrotal US. The total number of analyzed testes after orchidopexy was 184. Patients were divided according to age at the time of surgery: group I (2-4 years old), group II (5-7), and group III (8-10). In all patients, the testicular volume ratio was calculated as the operated testes volume versus the control testes mean volume. There was an increase in the median ratio in all age groups, from 0.86 to 0.95 in group I, 0.82 to 0.92 in group II and 0.78 to 0.90 in group III. In group of the patients 2 to 4 years old the growth of the ratio 3 years after surgery was statistically significant. Abnormalities in the structure of the testes, which may indicate severe damage to the testis, were seen in approximately 20% of patients on initial exams. On follow-up exams, this type of structure remained in 7% of patients. Testes with an initial ratio >0.25 and inhomogeneous structure did not show any significant growth. Scrotal US can be used for an accurate comparative assessment of the structure and growth of the testes before and after orchidopexy. Abnormalities in the structure of the testes may identify testes requiring more advanced methods of evaluation. Abbreviation: US = ultrasound.

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Embase

Institution

(Jedrzejewski, Wieczorek) Department of Pediatric Radiology, Medical University of Lublin, Gebali 6, Lublin 20-093, Poland (Osemlak, Nachulewicz) Department of Pediatric Surgery, Medical University of Lublin, Lublin, Poland

Publisher

Lippincott Williams and Wilkins (E-mail: kathiest.clai@apta.org)

Year of Publication

2016

539.

Persistent mullerian duct syndrome: A 24-year experience.

Saleem M., Ather U., Mirza B., Iqbal S., Sheikh A., Shaukat M., Sheikh M.T., Ahmad F., Rehan T.

Embase

Journal of Pediatric Surgery. 51 (10) (pp 1721-1724), 2016. Date of Publication: 01 Oct 2016.

[Article]

AN: 611157116

Background Persistence of mullerian duct derivatives in otherwise normal male child is a very rare disorder. This may lead to diagnostic as well as management dilemma. Materials and methods The medical record of 27 cases of persistent mullerian duct syndrome (PMDS) operated in three teaching hospitals more than a period of 24 years is retrieved and analyzed for demography, clinical presentation, investigations, and treatment. Results There were a total of twenty seven male children with PMDS. The age was ranged between 3 months and 19 years. Ten patients presented with isolated bilateral UDT, six patients with bilateral UDT and unilateral inguinal hernia (4 left and 2 right sided inguinal hernia), and eight patients presented with right inguinal hernia and left sided UDT. Eight of 27 patients showed familial trends i.e. four pairs of brothers had PMDS in our series. In 21 patients, the diagnosis was made incidentally while operating for UDT and inguinal hernia. At operation 5 patients had female type of PMDS and 22 patients had male type PMDS. In 6 patients (male type), the PMDS was associated with transverse testicular ectopia. In 18 patients the initial operation was performed through inguinal incision with excision of mullerian remnants in the same settings in 12 patients. In 4 patients, straightforward laparotomy performed (familial cases) to excise mullerian remnants. In 5 patients, the PMDS was diagnosed on laparoscopy; initially biopsy of these remnants and gonads was

done followed by excision of remnants by laparotomy approach. Biopsies taken from gonads in each patient revealed testicular tissue with variable degree of immaturity and dysplasia. The biopsy of mullerian remnants did not reveal any malignancy. All patients were genotypically male. Conclusion Isolated undescended testes, left UDT and right inguinal hernia, bilateral UDT and unilateral inguinal hernia are the main presenting features of PMDS. About 30% of the patients showed familial tendency. Inguinal exploration for UDT or inguinal hernia, and laparoscopy for UDT reveal incidental findings of mullerian remnants. PMDS can be managed as single stage procedure however two stage procedure including gonadal biopsies in first stage followed by mullerian remnants excision and orchidopexy in the second stage can be opted if there is doubt about gonads and genotype.

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Status

Embase

Institution

(Saleem, Mirza, Iqbal, Sheikh) Department of Paediatric Surgery, The Children's Hospital & the Institute of Child Health Lahore, Pakistan (Ather) Department of Gynaecology, Al-Noor Medical Centre, Lahore, Pakistan

(Shaukat, Sheikh) Department of Paediatric Surgery, Mayo Hospital/KEMC, Lahore, Pakistan

(Ahmad, Rehan) Department of General Surgery, BVH, Bahawalpur, Pakistan

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2016

540.

Colour Doppler and ultrasound characteristics of testicular Leydig cell tumours.

Maxwell F., Izard V., Ferlicot S., Rachas A., Correas J.-M., Benoit G., Bellin M.-F., Rocher L.

Embase

British Journal of Radiology. 89 (1062) (no pagination), 2016. Article Number: 20160089. Date of Publication: 2016.

[Article]

AN: 612533124

Objective: To assess the colour Doppler and ultrasound features of testicular Leydig cell tumours (LCTs) in a population of 38 surgically proven lesions.

Method(s): From August 2008 to March 2015, we retrospectively included 38 surgically proven LCTs in 36 patients. Clinical data, scrotal colour Doppler, Bmode ultrasound and videos images were reviewed for each patient. The volume, echotexture of the testis, size, shape, echogenicity and the vascularization pattern of the lesion were evaluated. The tumour margins were categorized as either smooth or lobulated. The vascularization was classified as intense, moderate or without any hypervascularization. We defined the vascularization pattern groups as central, peripheral and mixed (the latter meaning both central and peripheral).

Result(s): 26 patients were referred for infertility [5 patients were subsequently diagnosed with Klinefelter syndrome (KS) and 5 patients with cryptorchidism]. 28 patients underwent testis-sparing surgery, while 8 patients underwent a radical orchiectomy. The LCTs were mostly infracentimetric (68.4%), with a median size of 7.0mm (ranging from 4.0 to 11 mm). 50% of the lesions had lobulated margins, and these were significantly larger than the smooth lesions ($p,0.05$). The content of the lesions was markedly homogeneous and hypoechoic. All lesions had sharp demarcations from the adjacent pulp. 36/38 lesions exhibited moderate-to-intense hypervascularization, with a mixed intrinsic and peripheral rim pattern. Larger lesions were more hypervascularized ($p,0.05$). LCTs in patients with KS had atypical features.

Conclusion(s): Typical sporadic LCTs appeared as isolated hypoechoic, infracentimetric masses, with a clear demarcation from the adjacent pulp. They presented intrinsic and peripheral rim hypervascularization.

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Status

Embase

Institution

(Maxwell, Bellin, Rocher) Department of Radiology, Bicetre University Hospital, Paris, France

(Izard, Benoit) Department of Urology, Bicetre University Hospital, Paris, France

(Ferlicot) Department of Pathology, Bicetre University Hospital, Paris, France

(Rachas) Department of Public Health and Epidemiology, Bicetre University Hospital, Paris, France

(Correas) Departement of Radiology, Necker Hospital, Paris, France

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British Institute of Radiology (E-mail: publications@bir.org.uk)

Year of Publication

2016

541.

Malignancy in disorders of sex development.

Kathrins M., Kolon T.F.

Embase

Translational Andrology and Urology. 5 (5) (pp 794-798), 2016. Date of Publication: 2016.

[Review]

AN: 613625537

Disorders of sex development (DSD) represent a spectrum of conditions in which chromosomal, gonadal, or anatomic sex are atypical and affect 1 in 4,500-5,000 live births. The diagnosis of DSD raises concerns of tumor risk and treatment as well as future fertility preservation. We review the current understanding of the types of gonadal tumors that arise in DSD patients as well as possible markers and treatment. The goal is to inform the members of the DSD team (urologist, endocrinologist, geneticist, psychologist) of the latest findings regarding malignancy in DSD. PubMed and Google Scholar™ literature searches were performed of current and past peer-reviewed literature on DSD (intersex) regarding gonadal development and tumor formation/treatment. Relevant reviews and original research articles were examined, including cited references, and a synopsis of the data was generated. DSD patients are at increased risk for the development of testicular carcinoma in-situ (CIS) and germ cell tumors (GCT), including seminoma, non-seminoma, juvenile granulosa cell, gonadoblastoma, and dysgerminoma. Cancer risk factors include Y-chromosomal material and gonadal position, especially for streak gonads. The 46 XX DSD patients [congenital adrenal hyperplasia (CAH)] with no genetic Y-chromosomal material are not at higher risk of cancer. Post-pubertal complete androgen insensitivity syndrome (AIS) patients remain prone to tumor development if the testes remain in the abdomen. Estimates of the risk of GCT in partial AIS for untreated undescended testes may be as high as 50%. The cancer risk of scrotal testes in partial AIS is unknown. CIS occurs almost exclusively in patients with hypovirilization, most notably in AIS. Persistent Mullerian Duct Syndrome (PMDS) confers the usual cancer risk associated with cryptorchidism, but also a possible tumor risk of the Mullerian remnant. Several markers are under investigation for tumor evaluation in the DSD population beyond hCG and AFP (Oct3/4, TSPY, WT-1). The management of patients with DSD

is complex and evaluation of tumor risk is aided by advances in genotyping for Y-chromosomal material not evident in traditional karyotyping. More complete genetic screening for DSD patients should increasingly become the standard of care. Developments in pathologic diagnosis will further challenge our traditional understanding of the oncologic management and surveillance of these patients. Future studies utilizing more advanced histologic examination of gonads will improve our understanding of the true incidences of malignancy in this diverse population.

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Status

Embase

Institution

(Kathrins, Kolon) Department of Urology (Surgery), Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA, United States (Kolon) Department of Urology (Surgery), Children's Hospital of Philadelphia, Philadelphia, PA, United States

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AME Publishing Company (E-mail: info@amepc.org)

Year of Publication

2016

542.

The incidence of apparent congenital urogenital anomalies in North Indian newborns: A study of 20,432 pregnancies.

Bhat A., Kumar V., Bhat M., Kumar R., Patni M., Mittal R.

Embase

African Journal of Urology. 22 (3) (pp 183-188), 2016. Date of Publication: 01 Sep 2016.

[Article]

AN: 613244818

Introduction and objectives Over the last few decades, congenital anomalies of the urogenital system have increased globally as a consequence of advanced maternal age at pregnancy and developments in assisted reproductive techniques. The aim of this study was to determine the incidence of apparent congenital urogenital anomalies in North Indian newborns and the causative factors. Subjects and methods The data of all newborns delivered at our institute between September 2012 and August 2014 were collected for this prospective study. The

predetermined data format included the newborns' birth weight and gestational age, the maternal age, parity and infertility treatment, if any. Newborns weighing less than 1000 g or born before 32 weeks of gestation were excluded from the study. Results During the study period, 20,432 deliveries were recorded (10,952 male and 9480 female babies). Apparent urogenital congenital anomalies were diagnosed in 799, with an incidence of 39.1 per 1000 newborns. The most common anomaly was cryptorchidism found in 678 newborns, while hypospadias was noted in 61, ambiguous genitalia in 34, congenital hernia/hydrocele in 20 and an exstrophy-epispadias complex in 5 children. Prune belly syndrome was seen in 1 newborn. Newborns weighing less than 2500 g had a higher proportion of anomalies (9.64%) in comparison to those weighing over 2500 g (1.99%) ($p = 0.0001$). A maternal age >30 years, parity >2 and infertility treatment were recorded in 5.40%, 4.93% and 9.80%, respectively, and all were independently associated with an increased risk of urogenital anomalies ($p = 0.0001$). Conclusions The incidence of apparent congenital urogenital anomalies was 3.91%. Infertility treatment, parity >2 and a maternal age >30 years were independently associated with an increased risk of congenital urogenital anomalies.

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Status

Embase

Author NameID

Bhat A.; ORCID: <http://orcid.org/0000-0002-2141-4726>

Institution

(Bhat) Department of urology, Dr. S.N. Medical College, Jodhpur, India (Bhat, Kumar, Bhat, Kumar, Mittal) Department of Urology, S.P. Medical College, Bikaner, Rajasthan 334003, India (Patni) Department of Obstetric and Gynecology, S.P. Medical College, Bikaner, Rajasthan 334003, India

Publisher

Pan African Urological Surgeons Association(PAUSA) (E-mail: sunnydoodu@yahoo.com)

Year of Publication

2016

543.

Current management of non-palpable testes: A literature review and clinical results.

Sepulveda X., Lopez Egana P.-J.

Embase

Translational Pediatrics. 5 (4) (pp 233-239), 2016. Date of Publication: 2016.

[Review]

AN: 613217861

Cryptorchidism is a common pathology that occurs in 3% in full term newborns, and it decreases to 0.8-1.2% at 1 year of age. Nearly a 20% of undescended testes are non-palpable. Various surgical treatments have been described, but its management is still controversial. A literature review was made of non-palpable testes, analysing diagnostic tools, treatment and its results. Additionally we reviewed non-palpable testes cases treated in our centre in the last 20 years. Different techniques are described for the management of non-palpable testes; with or without section of the spermatic vessels and/or in 1 or 2 stages. Nowadays, literature supports the laparoscopic management in two stages. In our experience, we have better results in two-stage Fowler-Stephens than one-stage, with lower rates of testicular atrophy. Non-palpable testes are a common pathology in paediatric urology. Analysing the literature and our experience we recommend a two-stage surgery for intra-Abdominal testes, which has demonstrated good results and lower percentage of atrophic testis.

Status

Embase

Institution

(Sepulveda) Department of Pediatric Surgery, University of Valparaiso, Gustavo Fricke Hospital, Vina del Mar, Chile (Lopez Egana) Pediatric Urology Service, Exequiel Gonzalez Cortes Hospital, Barros Luco 3301, San-Miguel-Santiago, Chile

(Lopez Egana) Department of Pediatrics and Pediatric Surgery, School of Medicine, University of Chile, Santiago, Chile

(Lopez Egana) Clinica Alemana, Santiago, Chile

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2016

Diagnostic accuracy of conventional MRI with diffusion weighted imaging (DWI) in detection of cryptorchidism taking diagnostic laparoscopy as gold standard.

Zahra M., Javed A.M., Noreen A., Bushra H., Saeed U., Amin M.

Embase

Pakistan Journal of Medical and Health Sciences. 10 (2) (pp 471-474), 2016. Date of Publication: April-June 2016.

[Article]

AN: 612541129

Background: Use of imaging studies for accurate preoperative localization of the non-palpable testis is a widely prevalent practice. MRI is noninvasive, does not involve ionizing radiation, and yields multiplanar images. The combination of DWI and conventional MRI was the most sensitive and most accurate technique.

Aim(s): To determine diagnostic accuracy of DW-MRI for accurate preoperative localization and planning of the non-palpable testis.

Method(s): A total of 500 patients of empty scrotum with testis not palpable on physical examination and age ≤ 10 years were included in the study. Patients with h/o previous scrotal surgery, anorectal or renal malformation were excluded. All the patients were then underwent DW-MRI. The DW-MRI findings were recorded as presence or absence of the undescended testis. Diffusion weighted magnetic resonance imaging findings were correlated with laparoscopic findings.

Result(s): Mean age was 7.21 ± 1.43 years. DW-MRI detected the cryptorchidism in 378(80.82%) patients, out of which, 360 (True Positive) had cryptorchidism and 18(False Positive) had no cryptorchidism on laparoscopy. Overall sensitivity, specificity, positive predictive value, negative predictive value and diagnostic accuracy of DW-MRI in detecting cryptorchidism was 93.51%, 84.35%, 95.24%, 79.51% and 91.40% respectively.

Conclusion(s): This study concluded that diffusion weighted magnetic resonance imaging is a highly sensitive and accurate non-invasive modality for detecting cryptorchidism.

Status

Embase

Institution

(Zahra, Javed, Noreen, Bushra, Saeed, Amin) Department of Diagnostic Radiology, Children Hospital, Institute of Child Health, Multan, Pakistan

Publisher

Lahore Medical And Dental College (Tulspura, North Canal Bank, Lahore, Pakistan)

Year of Publication

2016

545.

Current Management of Undescended Testes.

Kurz D., Tasian G.

Embase

Current Treatment Options in Pediatrics. 2 (1) (pp 43-51), 2016. Date of Publication: 01 Mar 2016.

[Review]

AN: 611920127

Undescended testes (UDTs) are a relatively common finding in newborn males, especially in those born prematurely. Upon discovering a non-intrascrotal testis, it is important to determine whether the testis is palpable or non-palpable and whether the finding is unilateral or bilateral. Imaging should not be used in this workup, as no current modality has been shown to be adequately sensitive or specific to aid in management decisions. Patients with UDTs diagnosed after 6 months of age should be referred to a specialist for correction so that surgery may be performed within 1 year thereafter. This allows testes to descend spontaneously if they are to do so while facilitating early intervention to decrease the risk of subfertility and testicular malignancy for those patients in whom spontaneous descent does not occur. The surgical approach is often dependent on the location of the testis on physical exam. Most orchiopexies for palpable testes are performed through an inguinal incision, although a scrotal approach can be safely utilized depending on the testis position. Diagnostic laparoscopy is most often used for non-palpable testes, as it not only allows for the identification of an atrophic or absent testicle, but it also provides an opportunity to perform an orchiopexy simultaneously should a viable testis be found. Hormonal therapy is not recommended for treatment of UDTs due to its low success rate, the incidence of secondary re-ascent, and the possible detrimental effects on spermatogenesis. Finally, patients with bilateral non-palpable UDTs require a more extensive preliminary evaluation to rule out congenital adrenal hyperplasia (CAH) and disorders of sexual development (DSD). This involves serum electrolytes, karyotype analysis, and hormonal testing including a serum mullerian inhibiting substance (MIS), in order to determine if testicular tissue is present and functional.

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Status

Embase

Institution

(Kurz) University of Pennsylvania Perelman School of Medicine, Philadelphia, PA, United States
(Tasian) Division of Urology and Center for Pediatric Clinical Effectiveness, The Children's Hospital of Philadelphia, University of Pennsylvania Perelman School of Medicine, Philadelphia, PA, United States

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546.

Surgical pathologies in children presenting for non-medical. Tibbi endikasyon disi sunnet icin basvuran cocuklarda cerrahi patolojiler <Tibbi endikasyon disi sunnet icin basvuran cocuklarda cerrahi patolojiler.>

Thomas D.T.

Embase

Marmara Medical Journal. 29 (2) (pp 84-87), 2016. Date of Publication: 2016.

[Article]

AN: 611771138

Objectives: Circumcision is the most commonly performed surgical procedure in the world. In this study, we determined the frequency of additional surgical pathologies in patients presenting for non-medical circumcision.

Material(s) and Method(s): Children presenting to a secondary healthcare facility between June 2013 - June 2014 for non-medical circumcision were included in this study. Patients' ages, presenting at outpatient clinics were noted together with the results of physical examinations.

Result(s): A total of 2088 children presented for non-medical circumcision. Their average age was 5.2 years. 56.3% of patients presented to the Pediatric Surgery outpatient clinic, 25.3% presented to the Urology outpatient clinic and 18.3% presented to the General Surgery outpatient clinic.

Additional surgical pathologies were noted in 3.9% of patients and these were: phimosis (n=36), inguinal hernia/hydrocele (n=12), buried penis (n=10), undescended testis (n=9), retractile testis (n=5), hypospadias (n=3), megameatus (n=2), umbilical hernia (n=2) and varicocele (n=2). The surgical plans for 37 (1.8%) patients changed due to the findings at examination.

Conclusion(s): Changes in surgical plans were required for 1.8% of patients. It is therefore important that all patients, including those presenting with a request for circumcision, have a detailed physical examination.

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Status

Embase

Institution

(Thomas) Department of Pediatric Surgery, Faculty of Medicine, Maltepe University, Ataturk Cad. Cam Sok. No 3, Maltepe, Istanbul, Turkey

Publisher

Marmara University

Year of Publication

2016

547.

Growth and descent of the testes in infants with hypogonadotropic hypogonadism receiving subcutaneous gonadotropin infusion.

Lambert A.-S., Bougneres P.

Embase

International Journal of Pediatric Endocrinology. 2016 (1) (no pagination), 2016. Article Number: 13. Date of Publication: 2016.

[Article]

AN: 611276824

Background: One third of infants with congenital hypogonadotropic hypogonadism (CHH) are said to have micropenis and/or bilateral or unilateral cryptorchidism leading many of them to orchiopexy. Our previous study in two patients suggests that prolonged subcutaneous infusion of large doses of gonadotropins might normalize testicular function and growth. Case presentation: To confirm the effects of early and prolonged subcutaneous infusion of large doses of gonadotropins on growth and descent of the testes. Eight boys with CHH, aged 0.25-11 months. Testes were non-palpable in 5 or in high scrotal position in 3. CHH was isolated in 5 infants and part of a syndrome of combined pituitary hormonal deficits in the 3 others. In response to gonadotropin infusion, mean levels of testicular hormones were normalized. Complete testis

descent occurred in 6 patients. Partial descent occurred in 2. Testes re-ascended in 1 patient. Testes and penis gained normal dimensions in all cases.

Conclusion(s): Subcutaneous gonadotropin infusion seems able to induce testis descent in a large proportion of infants with CHH. If confirmed, this may allow patients to avoid testes surgery but studies in larger series are needed to evaluate the benefits of this treatment versus traditional orchiopexy.

Copyright © 2016 The Author(s).

Status

Embase

Institution

(Lambert, Bougneres) Department of Pediatric Endocrinology, Pole I3E, Hopital Bicetre, Assistance Publique des Hopitaux de Paris, Paris Sud University, Le Kremlin-Bicetre, France

Publisher

BioMed Central Ltd. (E-mail: info@biomedcentral.com)

Year of Publication

2016

548.

Pediatric laparoscopic surgery; initial experience from Pakistan; first 100 cases in single center.

Ishaq H., Qazi S.H., Dogar S., Khan Durrani M.Y., Faruque A.V.

Embase

JPMA. The Journal of the Pakistan Medical Association. Suppl. 66 3) (10) (pp S116-S118), 2016.

Date of Publication: 01 Oct 2016.

[Article]

AN: 623574375

In a developing country like Pakistan, laparoscopic surgeries are not considered favourable by many, possibly because of high costs or a lack of expertise. It is an established fact that laparoscopic surgery offers better surgical treatments with a shorter hospital stay and fewer complications. The current retrospective study was conducted at a tertiary care hospital in Karachi and comprised of laparoscopy cases performed by a single surgeon from March 2012 to September of 2014. A total of 100laparoscopic surgeries were performed; mostly appendectomies 49(49%) and undescended testes (UDTs) 34(34%). Overall, there were 70(70%)

male patients. The mean age of the patients was 7.1 years and standard deviation (SD) of 2. Four (4%) patients had cellulitis. Laparoscopy paediatric surgery offered advantages of fewer wound-associated complications, less incisional pain, a shorter recovery time, and improved cosmesis.

PMC Identifier

27895374 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27895374>]

Institution

(Ishaq) Undergraduate Medical Student, Aga Khan Medical College, Karachi, Pakistan (Qazi, Faruque) Section of Pediatric Surgery, Aga Khan University Hospital, Karachi, Pakistan

(Dogar) Section of Pediatric Surgery, Indus Hospital, Karachi, Pakistan

(Khan Durrani) Intern, Aga Khan Medical College, Karachi, Pakistan

Year of Publication

2016

549.

Environmental chemicals impact dog semen quality in vitro and may be associated with a temporal decline in sperm motility and increased cryptorchidism.

Lea R.G., Byers A.S., Sumner R.N., Rhind S.M., Zhang Z., Freeman S.L., Moxon R., Richardson H.M., Green M., Craigon J., England G.C.

Embase

Scientific reports. 6 (pp 31281), 2016. Date of Publication: 09 Aug 2016.

[Article]

AN: 622885311

Adverse temporal trends in human semen quality and cryptorchidism in infants have been associated with exposure to environmental chemicals (ECs) during development. Here we report that a population of breeding dogs exhibit a 26 year (1988-2014) decline in sperm quality and a concurrent increased incidence of cryptorchidism in male offspring (1995-2014). A decline in the number of males born relative to the number of females was also observed. ECs, including diethylhexyl phthalate (DEHP) and polychlorinated biphenyl 153 (PCB153), were detected in adult dog testes and commercial dog foods at concentrations reported to perturb reproductive function in other species. Testicular concentrations of DEHP and PCB153 perturbed sperm viability, motility and DNA integrity in vitro but did not affect LH stimulated testosterone secretion

from adult testis explants. The direct effects of chemicals on sperm may therefore contribute to the decline in canine semen quality that parallels that reported in the human.

PMC Identifier

27503122 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27503122>]

Institution

(Lea, Byers, Sumner, Freeman, Richardson, Green, England) School of Veterinary Medicine and Science, University of Nottingham, UK (Lea) School of Animal Rural and Environmental Sciences, Nottingham Trent University, UK

(Rhind, Zhang) Environmental and Biochemical Sciences, The James Hutton Institute, UK

(Moxon) National Breeding Centre, Guide Dogs for the Blind Association, UK

(Craigon) School of Biosciences, University of Nottingham, UK

Year of Publication

2016

550.

Evaluation of postoperative undescended testicles using point shear wave elastography in children.

Hattapoglu S., Goya C., Arslan S., Alan B., Ekici F., Tekbas G., Yildiz I., Hamidi C.

Embase

Ultrasonics. 72 (pp 191-194), 2016. Date of Publication: 01 Dec 2016.

[Article]

AN: 619829364

PURPOSE: To demonstrate the difference in tissue stiffness by comparing the value of the shear wave velocity (SWV) of postoperative undescended testicles with that of normal testes.

METHODS: This study included 39 patients and 30 healthy controls. US and p-SWE (VTQ) were performed using with a linear probe (4-9MHz). Forty-seven operated undescended testicles comprised "Group A", 27 testes with normal scrotal placement since birth in patient population comprised "Group B". A total of 60 testes in 30 healthy controls were included as "Group C".

Finally, the testes of Group A, B, C were statistically compared in terms of the SWV and volume.

RESULTS: The shear wave values of the 47 testes in Group A were 0.75-2.8 (median, 1.1)m/s, and the SWVs of the 27 testes in Group B were 0.62-1.2 (median, 0.84)m/s. The SWVs of the 60 testes in Group C were 0.65-1 (median, 0.82)m/s. The testicular volumes of Group A ranged from

0.19 to 4.7 (median, 0.15)cm(3), Group B ranged from 0.34 to 8 (median, 0.74)cm(3) and Group C ranged as 0.4-15.5 (median, 0.91)cm(3).

CONCLUSIONS: VTQ method of p-SWE is a new method that may reveal the difference in stiffness between scrotally placed testes and postoperative undescended testicles.

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PMC Identifier

27567037 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27567037>]

Institution

(Hattapoglu, Goya, Alan, Ekici, Tekbas, Hamidi) Department of Radiology, Medical School, Dicle University, Diyarbakir, Turkey (Arslan) Department of Pediatric Surgery, Medical School, Dicle University, Diyarbakir, Turkey

(Yildiz) Department of Biostatistics, Medical School, Dicle University, Diyarbakir, Turkey

Year of Publication

2016

551.

Gene Expression Changes Underlying Idiopathic Central Hypogonadism in Cryptorchidism with Defective Mini-Puberty.

Hadziselimovic F., Gegenschatz-Schmid K., Verkauskas G., Docampo-Garcia M.J., Demougin P., Bilius V., Malcius D., Dasevicius D., Stadler M.B.

Embase

Sexual development : genetics, molecular biology, evolution, endocrinology, embryology, and pathology of sex determination and differentiation. 10 (3) (pp 136-146), 2016. Date of Publication: 2016.

[Article]

AN: 619662316

The whole genome RNA profiling of testicular biopsies by DNA strand-specific RNA sequencing was examined to determine a potential causative role of isolated congenital cryptorchidism in azoospermia and/or infertility in the context of our previously published GeneChip data.

Cryptorchid patients, aged 7 months to 5 years and otherwise healthy, were enrolled in this prospective study. During surgery, testicular tissue biopsies were obtained for histological examination and RNA sequencing. Fifteen patients were selected based on the histological

results and were divided into 2 groups. Seven were classified as belonging to the high infertility risk (HIR) and 8 to the low infertility risk (LIR) group. Cryptorchid boys in the HIR group lacked transformation of gonocytes into Ad spermatogonia due to impaired mini-puberty. This group of patients will be infertile despite successful surgery. The new important finding was a decreased PROK2, CHD7, FGFR1, and SPRY4 gene expression in the HIR group. Furthermore, identification of multiple differences in gene expression between HIR and LIR groups underscores the importance of an intact hypothalamic-pituitary-gonadal axis for fertility development. Our RNA profiling data strongly support the theory that in the HIR group of cryptorchid boys insufficient PROK2/CHD7/FGFR1/SPRY4 gene expression induces deficient LH secretion, resulting in impaired mini-puberty and infertility. We therefore recommend hormonal treatment for this cohort of cryptorchid boys with defective mini-puberty following a seemingly successful orchidopexy.

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PMC Identifier

27561106 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27561106>]

Year of Publication

2016

552.

Nonobliteration of the Processus Vaginalis: Sonography of Related Abnormalities in Children.

Rafailidis V., Varelas S., Apostolopoulou F., Rafailidis D.

Embase

Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine. 35 (4) (pp 805-818), 2016. Date of Publication: 01 Apr 2016.

[Review]

AN: 617448524

The objective of this pictorial essay is to systematically classify processus vaginalis-related disorders in the light of embryology and present illustrative sonograms with corresponding diagrams. Failure of the processus vaginalis to obliterate during gestation results in a wide spectrum of anomalies, including communicating and noncommunicating hydroceles and inguinal and inguinoscrotal hernias, along with other related disorders of the genital system. There are varying classifications in the literature regarding the aforementioned entities. Proper and timely diagnosis of these entities is essential, given the differences in treatment. Although physical

examination can narrow the differential diagnosis, sonography plays an essential role in establishing the diagnosis.

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PMC Identifier

28027612 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=28027612>]

Institution

(Rafailidis) Department of Radiology, AHEPA University General Hospital, Thessaloniki, Greece
(Varelas, Apostolopoulou, Rafailidis) Department of Radiology, G. Gennimatas General Hospital, Thessaloniki, Greece

Year of Publication

2016

553.

Do retractile testes have anatomical anomalies?.

Anderson K.M., Costa S.F., Sampaio F.J., Favorito L.A.

Embase

International braz j urol : official journal of the Brazilian Society of Urology. 42 (4) (pp 803-809), 2016. Date of Publication: 01 Jul 2016.

[Article]

AN: 617370351

OBJECTIVES: To assess the incidence of anatomical anomalies in patients with retractile testis.

MATERIALS AND METHODS: We studied prospectively 20 patients (28 testes) with truly retractile testis and compared them with 25 human fetuses (50 testes) with testis in scrotal position. We analyzed the relations among the testis, epididymis and patency of the processus vaginalis (PV). To analyze the relations between the testis and epididymis, we used a previous classification according to epididymis attachment to the testis and the presence of epididymis atresia. To analyze the structure of the PV, we considered two situations: obliteration of the PV and patency of the PV. We used the Chi-square test for contingency analysis of the populations under study ($p < 0.05$).

RESULTS: The fetuses ranged in age from 26 to 35 weeks post-conception (WPC) and the 20 patients with retractile testis ranged in ages from 1 to 12 years (average of 5.8). Of the 50 fetal testes, we observed complete patency of the PV in 2 cases (4%) and epididymal anomalies (EAs)

in 1 testis (2%). Of the 28 retractile testes, we observed patency of the PV in 6 cases (21.4%) and EA in 4 (14.28%). When we compared the incidence of EAs and PV patency we observed a significantly higher prevalence of these anomalies in retractile testes ($p=0.0116$).

CONCLUSIONS: Retractable testis is not a normal variant with a significant risk of patent processus vaginalis and epididymal anomalies.

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PMC Identifier

27564294 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27564294>]

Institution

(Anderson, Costa, Sampaio, Favorito) Unidade de Pesquisa Urogenital, Universidade Estadual do Rio de Janeiro, RJ, Brasil

Year of Publication

2016

554.

Mixed Gonadal Dysgenesis with an unusual "inverted" Y chromosome.

Makhija D., Shah H., Tiwari C., Jayaswal S., Desale J.

Embase

Developmental period medicine. 20 (3) (pp 178-180), 2016. Date of Publication: 01 Jan 2016.

[Article]

AN: 616855863

Mixed gonadal dysgenesis is a rare disorder of sex development associated with sex chromosome aneuploidy and mosaicism of the Y chromosome. It is characterized by a unilateral non-palpable (usually intra-abdominal) testis, a contralateral streak gonad and persistent mullerian structures. The clinical presentation can vary from a typical male to female phenotype including all degrees of cryptorchidism, labial fusion, clitoromegaly, epispadias and hypospadias. It is the second most common cause of ambiguous genitalia in the neonatal period. We report a case of Mixed Gonadal Dysgenesis with an inverted Y chromosome.

PMC Identifier

27941186 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27941186>]

Institution

(Makhija) Dept of Paediatric Surgery, TNMC & BYL Nair Hospital, Mumbai Central, Mumbai, Maharashtra. India, E-Mail ID: deepee.sweety@gmail.com (Shah, Tiwari, Jayaswal, Desale)
Dept of Paediatric Surgery, TNMC & BYL Nair Hospital, Mumbai Central, Mumbai, Maharashtra.
India

Year of Publication

2016

555.

VARIATIONS IN TIMING OF ELECTIVE ORCHIDOPEXY.

Dobanovackic D., Vuckovic N., Slavkovic A., Lucic Prostran B., Lakic T., Gajic I.

Embase

Medicinski pregled. 69 (3-4) (pp 106-109), 2016. Date of Publication: 01 Mar 2016.

[Article]

AN: 611950774

RESULTS: A total of 637 patients (722 orchidopexies) underwent the elective operative treatment of undescended testis during the observed period. The analysis revealed that only 144 (22.60%) of cryptorchid infants were operated on within their first 18 months of life. In the group of 359 patients from* the urban environment, 101 (28.13%) were operated under the age of 18 months. Among the 278 patients from the rural environment, 43 (15.46%) were 18 months and younger at the time of surgery. **CONCLUSION:** The timing of surgical treatment of undescended testis in the study period was far from the recommended optimal time. It is evidently necessary to plan and provide additional information for pediatricians and parents about the current view on cryptorchidism and consequences of the late treatment.

INTRODUCTION: Undescended testis or cryptorchidism is detected in 3% of full-term male newborns, and in up to 33% of preemies. As the testicular descent may sometimes resolve spontaneously during first months of life, cryptorchidism is found in 1% of boys one year old. According to Consensus of Nordic experts in pediatric urology regarding cryptorchidism the optimal period for surgery is 12-18 months of age. The goal of this study was to identify the age of patients with congenital undescended testis at the time of surgery.

MATERIAL AND METHODS: A retrospective study included all the cases of cryptorchid patients who had undergone orchidopexy in the period from 2007 to 2014. The patients' age and the place of residence were analyzed.

PMC Identifier

27506098 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=27506098>]

Year of Publication

2016

556.

Human anogenital distance: An update on fetal smoke-exposure and integration of the perinatal literature on sex differences.

Fowler P.A., Filis P., Bhattacharya S., Le Bizec B., Antignac J.-P., Morvan M.-L., Drake A.J., Soffientini U., O'Shaughnessy P.J.

Embase

Human Reproduction. 31 (2) (pp 463-472), 2016. Date of Publication: 24 May 2016.

[Article]

AN: 608914350

STUDY QUESTION Do sex and maternal smoking effects on human fetal anogenital distance (AGD) persist in a larger study and how do these data integrate with the wider literature on perinatal human AGD, especially with respect to sex differences? SUMMARY ANSWER Second trimester sex differences in AGD are broadly consistent with neonatal and infant measures of AGD and maternal cigarette smoking is associated with a temporary increase in male AGD in the absence of changes in circulating testosterone. WHAT IS KNOWN ALREADY AGD is a biomarker of fetal androgen exposure, a reduced AGD in males being associated with cryptorchidism, hypospadias and reduced penile length. Normative fetal AGD data remain partial and windows of sensitivity of human fetal AGD to disruption are not known. STUDY DESIGN, SIZE, DURATION The effects of fetal sex and maternal cigarette smoking on the second trimester (11-21 weeks of gestation) human fetal AGD were studied, along with measurement of testosterone and testicular transcripts associated with apoptosis and proliferation. PARTICIPANTS/MATERIALS, SETTING METHODS AGD, measured from the centre of the anus to the posterior/caudal root of penis/clitoris (AGDapp) was determined in 56 female and 70 male morphologically normal fetuses. These data were integrated with current literature on perinatal AGD in humans. MAIN RESULTS AND THE ROLE OF CHANCE At 11-13 weeks of gestation male fetal AGDapp was 61% ($P < 0.001$) longer than in females, increasing to 70% at 17-21 weeks. This sexual dimorphism was independent of growth characteristics (fetal weight, length,

gonad weight). We confirmed that at 14-16 weeks of gestation male fetal AGD was increased 28% ($P < 0.05$) by in utero cigarette smoke exposure. Testosterone levels were not affected by smoking. To develop normative data, our findings have been integrated with available data from in vivo ultrasound scans and neonatal studies. Inter-study variations in male/female AGD differences lead to the conclusion that normalization and standardization approaches should be developed to enable confidence in comparing data from different perinatal AGD studies.

LIMITATIONS, REASONS FOR CAUTION Sex differences, and a smoking-dependent increase in male fetal AGD at 14-16 weeks, identified in a preliminary study, were confirmed with a larger number of fetuses. However, human fetal AGD should be re-assessed once much larger numbers of fetuses have been studied and this should be integrated with more detailed analysis of maternal lifestyle. Direct study of human fetal genital tissues is required for further mechanistic insights. **WIDER IMPLICATIONS OF THE FINDINGS** Fetal exposure to cigarette smoke chemicals is known to lead to reduced fertility in men and women. Integration of our data into the perinatal human AGD literature shows that more work needs to be done to enable reliable inter-study comparisons. **STUDY FUNDING/COMPETING INTEREST(S)** The study was supported by grants from the Chief Scientist Office (Scottish Executive, CZG/1/109 & CZG/4/742), NHS Grampian Endowments (08/02), the European Community's Seventh Framework Programme (FP7/2007-2013) under grant agreement no 212885 and the Medical Research Council, UK (MR/L010011/1). The authors declare they have no competing interests, be it financial, personal or professional.

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PMC Identifier

26732622 [<http://www.ncbi.nlm.nih.gov/pubmed/?term=26732622>]

Status

Embase

Institution

(Fowler, Filis) Institute of Medical Sciences, University of Aberdeen, Foresterhill, Aberdeen AB25 2ZD, United Kingdom (Bhattacharya) Institute of Applied Health Sciences, University of Aberdeen, Foresterhill, Aberdeen AB25 2ZD, United Kingdom

(Le Bizec, Antignac, Morvan) USC INRA 1329, Laboratoire d'Etude des Residus et Contaminants dans les Aliments, LUNAM Université, Oniris, Nantes F-44307, France

(Drake) Endocrinology Unit, Queen's Medical Research Institute, University/BHF Centre for Cardiovascular Science, University of Edinburgh, 47 Little France Crescent, Edinburgh EH16 4TJ, United Kingdom

(Soffientini, O'Shaughnessy) Institute of Biodiversity, Animal Health and Comparative Medicine (IBAHCM), College of Medical, Veterinary and Life Sciences, University of Glasgow, Bearsden Rd, Glasgow G61 1QH, United Kingdom

Publisher

Oxford University Press

Year of Publication

2016

557.

Timing of Orchidopexy for Undescended Testis in Israel: A Quality of Care Study.

Hidas G; Ben Chaim J; Udassin R; Graeb M; Gofrit ON; Zisk-Rony RY; Pode D; Duvdevani M; Yutkin V; Neheman A; Fruman A; Arbel D; Kopuler V; Armon Y; Landau EH.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Israel Medical Association Journal: Imaj. 18(11):697-700, 2016 11.

[Journal Article]

UI: 28466623

BACKGROUND: Strong evidence suggests that in order to prevent irreversible testicular damage surgical correction (orchidopexy) for undescended testis (UDT) should be performed before the age of 1 year.

OBJECTIVES: To evaluate whether orchidopexy is delayed in our medical system, and if so, to explore the pattern of referral for orchidopexy as a possible contributing factor in such delays.

METHODS: We conducted a retrospective chart review of all children who underwent orchidopexy for UDT between 2003 and 2013 in our institution. We collected data on the age at surgery and the child's health insurance plan. We also surveyed pediatricians from around the country regarding their pattern of UDT patient referral to a pediatric urologist or surgeon for surgical correction.

RESULTS: A total of 813 children underwent orchidopexy in our institute during the study period. The median age at surgery was 1.49 years (range 0.5-13). Only 11% of the children underwent surgery under the age of 1 year, and 53% between the ages of 1 and 2 years. These findings were consistent throughout the years, with no difference between the four health insurance plans.

Sixty-three pediatricians who participated in the survey reported that they referred children to surgery at a median age of 1 year (range 0.5-3 years).

CONCLUSIONS: Our results demonstrate delayed orchidopexy in our medical system. There is a need to improve awareness for early specialist consultation in order to facilitate earlier surgery and better care.

Version ID

1

Status

MEDLINE

Authors Full Name

Hidas, Guy; Ben Chaim, Jacob; Udassin, Refael; Graeb, Mary; Gofrit, Ofer N; Zisk-Rony, Rachel Yaffa; Pode, Dov; Duvdevani, Mordechai; Yutkin, Vladimir; Neheman, Amos; Fruman, Amos; Arbel, Dan; Kopuler, Vadim; Armon, Yaron; Landau, Ezekiel H.

Institution

Hidas, Guy. Pediatric Urology Unit, Hadassah-Hebrew University Medical Center, Jerusalem, Israel. Ben Chaim, Jacob. Pediatric Urology Unit, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Udassin, Refael. Department of Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Graeb, Mary. Pediatric Urology Unit, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Gofrit, Ofer N. Department of Urology, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Zisk-Rony, Rachel Yaffa. School of Nursing, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Pode, Dov. Department of Urology, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Duvdevani, Mordechai. Departments of Urology and Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Yutkin, Vladimir. Department of Urology, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Neheman, Amos. Pediatric Urology Unit, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Fruman, Amos. Department of Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Arbel, Dan. Department of Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Kopuler, Vadim. Department of Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Armon, Yaron. Department of Pediatric Surgery, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Landau, Ezekiel H. Pediatric Urology Unit, Hadassah-Hebrew University Medical Center, Jerusalem, Israel.

Year of Publication

2016

558.

One-Stage Laparoscopic Orchiopexy for the Treatment of Intraabdominal Testis.

Jawdat JR; Kocherov S; Chertin B.

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Israel Medical Association Journal: Imaj. 18(11):669-672, 2016 Nov.

[Journal Article]

UI: 28466616

BACKGROUND: Laparoscopy has gradually become the gold standard for the treatment of non-palpable testicles (NPT), with different success and complication rates.

OBJECTIVES: To evaluate outcomes of the one-stage laparoscopic orchiopexy for NPT in our department.

METHODS: We retrospectively evaluated the medical files of patients who underwent laparoscopic orchidopexy with the identical technique. Only patients with at least one year follow-up were included. At follow-up we assessed the age (at surgery), follow-up time, laterality of testes, postoperative complications, testicular size and testicular localization.

RESULTS: Thirty-six consecutive patients, median age 16 months, underwent one-stage laparoscopic orchiopexy. Sixteen patients (44.4%) had peeping testis type, in 13 patients (36.1%) the testicle was located within 2 cm from the internal ring and in the remaining 7 patients (19.4%) it was detected > 2 cm from the internal ring. In six children (16.7%) dividing the spermatic vessels was performed in one stage with laparoscopic orchiopexy. In the remaining 30 patients (83.7%) a laparoscopic one-stage procedure was performed with preservation of the spermatic vessels. Testicular atrophy was observed in 2 cases (5.6%), and 6 patients (16%) had a relatively

small testicle compared to the contralateral normal testicle at follow-up. Two patients (5.6%) presented with testicle positioning at the entrance area into the scrotum. None of the patients demonstrated hernia recurrence at follow-up. There was no difference in surgical outcome in children who had surgery with preservation of the spermatic vessels versus those who underwent orchiopexy with division of the spermatic vessels in one stage.

CONCLUSIONS: Laparoscopic transection of the testicular vessels appeared to be safe in boys with high abdominal testes that did not reach the scrotum after laparoscopic high retroperitoneal dissection.

Version ID

1

Status

MEDLINE

Authors Full Name

Jawdat, Jaber R; Kocherov, Stanislav; Chertin, Boris.

Institution

Jawdat, Jaber R. Department of Pediatric Urology, Shaare Zedek Medical Center, affiliated with Hebrew University-Hadassah Medical School, Jerusalem, Israel. Kocherov, Stanislav.

Department of Pediatric Urology, Shaare Zedek Medical Center, affiliated with Hebrew University-Hadassah Medical School, Jerusalem, Israel.

Chertin, Boris. Department of Pediatric Urology, Shaare Zedek Medical Center, affiliated with Hebrew University-Hadassah Medical School, Jerusalem, Israel.

Year of Publication

2016

559.

Familial forms of disorders of sex development may be common if infertility is considered a comorbidity.

Brauner R; Picard-Dieval F; Lottmann H; Rouget S; Bignon-Topalovic J; Bashamboo A; McElreavey K.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

BMC Pediatrics. 16(1):195, 2016 11 29.

[Journal Article]

UI: 27899089

BACKGROUND: Families with 46,XY Disorders of Sex Development (DSD) have been reported, but they are considered to be exceptionally rare, with the exception of the familial forms of disorders affecting androgen synthesis or action. The families of some patients with anorchia may include individuals with 46,XY gonadal dysgenesis. We therefore analysed a large series of patients with 46,XY DSD or anorchia for the occurrence in their family of one of these phenotypes and/or ovarian insufficiency and/or infertility and/or cryptorchidism.

METHODS: A retrospective study chart review was performed for 114 patients with 46,XY DSD and 26 patients with 46,XY bilateral anorchia examined at a single institution over a 33 year period.

RESULTS: Of the 140 patients, 25 probands with DSD belonged to 21 families and 7 with anorchia belonged to 7 families. Familial forms represent 22% (25/114) of the 46,XY DSD and 27% (7/26) of the anorchia cases. No case had disorders affecting androgen synthesis or action or 5 alpha-reductase deficiency. The presenting symptom was genital ambiguity (n = 12), hypospadias (n = 11) or discordance between 46,XY karyotyping performed in utero to exclude trisomy and female external genitalia (n = 2) or anorchia (n = 7). Other familial affected individuals presented with DSD and/or premature menopause (4 families) or male infertility (4 families) and/or cryptorchidism. In four families mutations were identified in the genes SRY, NR5A1, GATA4 and FOG2/ZFPM2. Surgery discovered dysgerminoma or gonadoblastoma in two cases with gonadal dysgenesis.

CONCLUSIONS: This study reveals a surprisingly high frequency of familial forms of 46,XY DSD and anorchia when premature menopause or male factor infertility are included. It also demonstrates the variability of the expression of the phenotype within the families. It highlights the need to the physician to take a full family history including fertility status. This could be important to identify familial cases, understand modes of transmission of the phenotype and eventually understand the genetic factors that are involved.

Version ID

1

Status

MEDLINE

Authors Full Name

Brauner, Raja; Picard-Dieval, Flavia; Lottmann, Henri; Rouget, Sebastien; Bignon-Topalovic, Joelle; Bashamboo, Anu; McElreavey, Ken.

Institution

Brauner, Raja. Fondation Ophtalmologique Adolphe de Rothschild and Universite Paris Descartes, Paris, France. raja.brauner@wanadoo.fr. Picard-Dieval, Flavia. Fondation Ophtalmologique Adolphe de Rothschild and Universite Paris Descartes, Paris, France. Lottmann, Henri. Assistance Publique-Hopitaux de Paris, Hopital Necker-Enfants Malades, Service de chirurgie viscerale pediatrique, Paris, France. Rouget, Sebastien. Fondation Ophtalmologique Adolphe de Rothschild and Universite Paris Descartes, Paris, France. Bignon-Topalovic, Joelle. Human Developmental Genetics, Institut Pasteur, Paris, France. Bashamboo, Anu. Human Developmental Genetics, Institut Pasteur, Paris, France. McElreavey, Ken. Human Developmental Genetics, Institut Pasteur, Paris, France.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5129225>

Year of Publication

2016

560.

Commentary to "Management of undescended testes: European Association of Urology/European Society for Paediatric Urology Guidelines".

Kolon TF.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of pediatric urology. 12(6):344, 2016 Dec.

[Journal Article]

UI: 27866870

Version ID

1

Status

MEDLINE

Authors Full Name

Kolon, Thomas F.

Institution

Kolon, Thomas F. Children's Hospital of Philadelphia, Department of Surgery, Division of Urology, 34th St. and Civic Center Blvd., Wood Building, 3rd Floor - Urology, Philadelphia, PA 19104, USA. Electronic address: kolon@email.chop.edu.

Year of Publication

2016

561.

The Undescended Testicles of West Virginia: A Single Center Experience.

Dibianco JM; Point D; Morley C; Zaslau S; Al-Omar O.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

West Virginia Medical Journal. 112(4):24-7, 2016 Jul-Aug.

[Journal Article]

UI: 27491098

OBJECTIVE: To determine the age of pediatric patients who underwent surgical intervention for undescended testicles (UDT) at our institution.

METHODS: We retrospectively reviewed all pediatric patients who underwent orchiopexy and/or diagnostic laparoscopy for undescended or non-palpable testicles with our pediatric urologist from January 2013-March 2014. Patients were separated into those undergoing surgical intervention at 6-12 months, 13-24 months, 25-48 months, and >48 months of age.

RESULTS: 70 patients underwent surgical intervention. Only 15 patients (21.4%) underwent surgical intervention within the recommended time period of 6-12 months. Orchiopexy was performed on 21 patients (30.0%) from 13-24 months, 12 patients (17.1%) from 25-48 months, and 22 patients (31.5%) after 48 months of age.

CONCLUSIONS: Current American Urologic Association (AUA) recommendations advocate orchiopexy between 6-12 months of age. Improved parent and primary care education and access to pediatric urological evaluation of UDT will hopefully improve the timeliness of intervention within our state.

Version ID

1

Status

MEDLINE

Authors Full Name

Dibianco, John Michael; Point, Dana; Morley, Chad; Zaslau, Stanley; Al-Omar, Osama.

Comments

Comment in (CIN)

Year of Publication

2016

562.

A allele of SNP12 in estrogen receptor 1 was a risk factor for cryptorchidism in Asians: a systematic review with meta-analysis and trial sequential analysis. [Review]

Deng C; Dai R; Li X; Liu F.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Pediatric Surgery International. 32(10):981-7, 2016 Oct.

[Journal Article. Meta-Analysis. Review. Systematic Review]

UI: 27376826

OBJECTIVE: Some studies have been carried out to evaluate the association between SNP12 in estrogen receptor 1 and cryptorchidism, but the results remain inconsistent. We carried out a meta-analysis to explore the association between this polymorphism and cryptorchidism risk.

METHODS: All eligible studies were searched in PubMed, Web of Science, Embase and Cochrane Library. Pooled odds ratios, with 95 % confidence intervals, were assessed for the association using fixed- and random-effects models.

RESULTS: Overall, four case-control studies (363 cases, 415 controls) were included in the meta-analysis. No significant publication bias (P Begg = 0.308, P Egger = 0.288) was found. A allele of SNP12 in estrogen receptor 1 was protective factor to cryptorchidism in allele model, dominant genetic model and heterozygote comparison in Caucasians, but the result was turned out to be false positive by trial sequential analysis. However, A in allele model was risk factor to cryptorchidism in Asians (odds ratio 2.02, 95 % confidence interval 1.03-3.01, p = 0.946 for heterogeneity) and the result was turned out to be true positive by trial sequential analysis, even though there were merely two original studies.

CONCLUSIONS: The results of this meta-analysis suggest that A allele of SNP12 in estrogen receptor 1 may increase the risk of cryptorchidism in Asians. Meanwhile, further well-designed

studies with large sample sizes are required to confirm the present findings, especially in Caucasians.

Version ID

1

Status

MEDLINE

Authors Full Name

Deng, Changkai; Dai, Rong; Li, Xuliang; Liu, Feng.

Institution

Deng, Changkai. Department of Pediatric Surgery, Chengdu Women's and Children's Central Hospital, Chengdu, China. Deng, Changkai. Department of Urology Surgery, Children's Hospital of Chongqing Medical University, Ministry of Education Key Laboratory of Child Development and Disorder, Key Laboratory of Pediatrics in Chongqing (CSTC2009CA5002), Chongqing International Science and Technology Cooperation Center for Child Development and Disorders, No.136, Zhongshan 2 RD, Yuzhong District, Chongqing, 400014, China.

Dai, Rong. Chengdu Center for Disease Control and Prevention, Chengdu, China.

Li, Xuliang. Department of Urology Surgery, Children's Hospital of Chongqing Medical University, Ministry of Education Key Laboratory of Child Development and Disorder, Key Laboratory of Pediatrics in Chongqing (CSTC2009CA5002), Chongqing International Science and Technology Cooperation Center for Child Development and Disorders, No.136, Zhongshan 2 RD, Yuzhong District, Chongqing, 400014, China.

Liu, Feng. Department of Urology Surgery, Children's Hospital of Chongqing Medical University, Ministry of Education Key Laboratory of Child Development and Disorder, Key Laboratory of Pediatrics in Chongqing (CSTC2009CA5002), Chongqing International Science and Technology Cooperation Center for Child Development and Disorders, No.136, Zhongshan 2 RD, Yuzhong District, Chongqing, 400014, China. 295969893@qq.com.

Year of Publication

2016

563.

Commentary to 'Prospective study of histological and endocrine parameters of gonadal function in boys with cryptorchidism'.

Rosch WH; Schroder A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of pediatric urology. 12(4):239, 2016 Aug.

[Journal Article]

UI: 27346070

Version ID

1

Status

MEDLINE

Authors Full Name

Rosch, Wolfgang H; Schroder, Annette.

Year of Publication

2016

564.

Re. "Prospective study of histological and endocrine parameters of gonadal function in boys with
cryptorchidism".

Kolon TF.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of pediatric urology. 12(4):240, 2016 Aug.

[Journal Article]

UI: 27296788

Version ID

1

Status

MEDLINE

Authors Full Name

Kolon, Thomas F.

Institution

Kolon, Thomas F. Children's Hospital of Philadelphia, Department of Surgery, Division of Urology, Philadelphia, PA, USA. Electronic address: kolon@email.chop.edu.

Year of Publication

2016

565.

Pediatric Urology for the General Surgeon. [Review]

Chalmers DJ; Vemulakonda VM.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Surgical Clinics of North America. 96(3):545-65, 2016 Jun.

[Journal Article. Review]

UI: 27261794

Pediatric urology spans the neonatal period through the transition into early adulthood. There are a variety of common pediatric urologic conditions that overlap significantly with pediatric surgery. This article reviews the pertinent pathophysiology of a few key disease processes, including the pediatric inguinal hernia and/or hydrocele, cryptorchidism, and circumcision. General surgeons may find themselves in the position of managing these problems primarily, particularly in rural areas that may lack pediatric subspecialization. An understanding of the fundamentals can guide appropriate initial management. Additional focus is devoted to the management of genitourinary trauma to guide the general surgeon in more acute, emergent settings.

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Version ID

1

Status

MEDLINE

Authors Full Name

Chalmers, David J; Vemulakonda, Vijaya M.

Institution

Chalmers, David J. Division of Urology, Maine Medical Center, Tufts University School of Medicine, 22 Bramhall Street, Portland, ME 04102, USA. Electronic address:

dchalmers@mmc.org. Vemulakonda, Vijaya M. Department of Pediatric Urology, Children's Hospital Colorado, 12123 East 16th Avenue, Aurora, CO 80045, USA.

Year of Publication

2016

566.

The histologic features of intratubular germ cell neoplasia and its correlation with tumor behavior.

Basiri A; Movahhed S; Parvin M; Salimi M; Rezaeetalab GH.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Investigative And Clinical Urology. 57(3):191-5, 2016 05.

[Journal Article]

UI: 27195317

PURPOSE: To assess the prevalence of intratubular germ cell neoplasia (ITGCN) in patients with concurrent testis tumor and its correlation with histologic features and serum tumor markers.

MATERIALS AND METHODS: From 2003 to 2015, 179 patients underwent radical orchiectomy due to testicular mass. Tissue specimens were evaluated by an expert uro-pathologist using immunohistochemistry (IHC) staining, in addition to light microscopy, to identify presence of ITGCN. Patients' demographic characteristics, histologic subtypes, pathologic stage of tumor and serum tumor markers were gathered and analyzed.

RESULTS: Eighty-five out of 179 patients (47.5%) had concomitant ITGCN according to IHC staining. There was not statistically significant difference in histologic type, histologic components, cryptorchidism, and lymphovascular invasion between the 2 groups ($p=0.151$, $p=0.11$, $p=0.233$, $p=0.413$, and $p=0.14$, respectively). The prevalence of ITGCN was significantly higher in patients with stage T2 and T3 of tumor than those with stage T1. Elevated serum alpha fetoprotein level is much common in patients with ITGCN ($p<0.001$).

CONCLUSIONS: The prevalence of concurrent ITGCN in our region is lower than previous data from western countries. ITGCN is more common in higher tumor stages and is accompanied with elevated serum alpha fetoprotein levels before surgery. Presence of ITGCN in adjacent tissue may suggest a negative cancer behavior.

Version ID

1

Status

MEDLINE

Authors Full Name

Basiri, Abbas; Movahhed, Saeed; Parvin, Mahmood; Salimi, Maziar; Rezaeetalab, Gholam Hossein.

Institution

Basiri, Abbas. Department of Urology, Shahid Beheshti University of Medical Sciences, Tehran, Iran.; Urology and Nephrology Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran. Movahhed, Saeed. Department of Urology, Shahid Beheshti University of Medical Sciences, Tehran, Iran.

Parvin, Mahmood. Department of Pathology, Shahid Beheshti University of Medical Sciences, Tehran, Iran.

Salimi, Maziar. Department of Urology, Shahid Beheshti University of Medical Sciences, Tehran, Iran.

Rezaeetalab, Gholam Hossein. Department of Urology, Shahid Beheshti University of Medical Sciences, Tehran, Iran.; Urology and Nephrology Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4869572>

Year of Publication

2016

567.

NNT mutations: a cause of primary adrenal insufficiency, oxidative stress and extra-adrenal defects.

Roucher-Boulez F; Mallet-Motak D; Samara-Boustani D; Jilani H; Ladjouze A; Souchon PF; Simon D; Nivot S; Heinrichs C; Ronze M; Bertagna X; Groisne L; Leheup B; Naud-Saudreau C; Blondin G; Lefevre C; Lemarchand L; Morel Y.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

European Journal of Endocrinology. 175(1):73-84, 2016 Jul.

[Journal Article]

UI: 27129361

OBJECTIVE: Nicotinamide nucleotide transhydrogenase (NNT), one of the several genes recently discovered in familial glucocorticoid deficiencies (FGD), is involved in reactive oxygen species detoxification, suggesting that extra-adrenal manifestations may occur, due to the sensitivity to oxidative stress of other organs rich in mitochondria. Here, we sought to identify NNT mutations in a large cohort of patients with primary congenital adrenal insufficiency without molecular etiology and evaluate the degree of adrenal insufficiency and onset of extra-adrenal damages.

METHODS: Sanger or massive parallel sequencing of NNT and patient monitoring.

RESULTS: Homozygous or compound heterozygous NNT mutations occurred frequently (26%, 13 unrelated families, 18 patients) in our cohort. Seven new mutations were identified: p.Met337Val, p.Ala863Glu, c.3G>A (p.Met1?), p.Arg129*, p.Arg379*, p.Val665Profs*29 and p.Ala704Serfs*19. The most frequent mutation, p.Arg129*, was found recurrently in patients from Algeria. Most patients were diagnosed belatedly (8-18 months) after presenting severe hypoglycemia; others experiencing stress conditions were diagnosed earlier. Five patients also had mineralocorticoid deficiency at onset. One patient had congenital hypothyroidism and two cryptorchidism. In follow-up, we noticed gonadotropic and genitalia impairments (precocious puberty, testicular inclusions, interstitial Leydig cell adenoma, azoospermia), hypothyroidism and hypertrophic cardiomyopathy. Intrafamilial phenotype heterogeneity was also observed.

CONCLUSIONS: NNT should be sequenced, not only in FGD, but also in all primary adrenal insufficiencies for which the most frequent etiologies have been ruled out. As NNT is involved in oxidative stress, careful follow-up is needed to evaluate mineralocorticoid biosynthesis extent, and gonadal, heart and thyroid function.

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Version ID

1

Status

MEDLINE

Authors Full Name

Roucher-Boulez, Florence; Mallet-Motak, Delphine; Samara-Boustani, Dinane; Jilani, Houweyda; Ladjouze, Asmahane; Souchon, Pierre-Francois; Simon, Dominique; Nivot, Sylvie; Heinrichs, Claudine; Ronze, Maryline; Bertagna, Xavier; Groisne, Laure; Leheup, Bruno; Naud-Saudreau, Catherine; Blondin, Gilles; Lefevre, Christine; Lemarchand, Laetitia; Morel, Yves.

Institution

Roucher-Boulez, Florence. Molecular Endocrinology and Rare DiseasesLyon University Hospital, Bron, France Claude Bernard Lyon 1 UniversityLyon, France florence.roucher@chu-lyon.fr.

Mallet-Motak, Delphine. Molecular Endocrinology and Rare DiseasesLyon University Hospital, Bron, France.

Samara-Boustani, Dinane. Pediatric Endocrinology, Gynecology and DiabetologyNecker University Hospital, Paris, France.

Jilani, Houweyda. Molecular Endocrinology and Rare DiseasesLyon University Hospital, Bron, France.

Ladjouze, Asmahane. Pediatric DepartmentBab El Oued University Hospital, Alger, Algeria.

Souchon, Pierre-Francois. Pediatric Endocrinology and DiabetologyAmerican Memorial Hospital, Reims, France.

Simon, Dominique. Pediatric EndocrinologyRobert Debre Hospital, Paris, France.

Nivot, Sylvie. Department of PediatricsRennes Teaching Hospital, Rennes, France.

Heinrichs, Claudine. Pediatric EndocrinologyQueen Fabiola Children's University Hospital, Brussels, Belgium.

Ronze, Maryline. Endocrinology DepartmentL.-Hussel Hospital, Vienne, France.

Bertagna, Xavier. Endocrinology DepartmentCochin University Hospital, Paris, France.

Groisne, Laure. Endocrinology DepartmentLyon University Hospital, Bron-Lyon, France.

Leheup, Bruno. Paediatric and Clinical Genetic DepartmentNancy University Hospital, Vandoeuvre les Nancy, France.

Naud-Saudreau, Catherine. Pediatric Endocrinology and DiabetologyBretagne Sud Hospital Center, Lorient, France.

Blondin, Gilles. Pediatric Endocrinology and DiabetologyBretagne Sud Hospital Center, Lorient, France.

Lefevre, Christine. Pediatric EndocrinologyJeanne de Flandre Hospital, Lille, France.

Lemarchand, Laetitia. Pediatric DepartmentLa Rochelle-Re-Aunis Hospital Group, La Rochelle, France.

Morel, Yves. Molecular Endocrinology and Rare DiseasesLyon University Hospital, Bron, France
Claude Bernard Lyon 1 UniversityLyon, France.

Year of Publication

2016

568.

Testicular microlithiasis and testicular cancer: review of the literature. [Review]

Pedersen MR; Rafaelsen SR; Moller H; Vedsted P; Osther PJ.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

International Urology & Nephrology. 48(7):1079-86, 2016 Jul.

[Journal Article. Review. Systematic Review]

UI: 27007613

PURPOSE: To perform a systematic literature review to assess whether the occurrence of testicular microlithiasis (TML) in conjunction with other risk factors is associated with testicular cancer.

METHODS: A systematic literature search was performed of original articles in English published 1998 to 2015. Relevant studies were selected by reading the title and abstract by two of the authors. Studies were included if TML was diagnosed by ultrasonography and a risk condition was reported. Studies were only eligible if the particular risk condition was reported in more than one article.

RESULTS: In total, 282 abstracts in were identified. Based on title and abstract the eligibility was assessed and 31 studies were included. Five conditions in relation to TML and testicular cancer emerged: Down syndrome, McCune-Albright syndrome, cryptorchidism, infertility and familial disposition of testicular cancer.

CONCLUSION: Data support the conclusion that TML is not an independent risk factor for testicular cancer but associated with testicular cancer through other conditions. In male infertility, TML appears to be related to an increased risk of testicular cancer possibly as part of a testicular dysgenesis syndrome.

Version ID

1

Status

MEDLINE

Author NameID

Pedersen, Malene Roland; ORCID: <http://orcid.org/0000-0001-5167-7968>

Authors Full Name

Pedersen, Malene Roland; Rafaelsen, Soren Rafael; Moller, Henrik; Vedsted, Peter; Osther, Palle Jorn.

Institution

Pedersen, Malene Roland. Department of Radiology, Vejle Hospital, Part of Lillebaelt Hospital, Kabbeltoft 25, 7100, Vejle, Denmark. malene.roland.vils.pedersen@rsyd.dk. Pedersen, Malene Roland. Department of Urology, Urological Research Center, Lillebaelt Hospital, Fredericia, Denmark. malene.roland.vils.pedersen@rsyd.dk.

Pedersen, Malene Roland. Institute of Regional Health Research, University of Southern Denmark, Odense, Denmark. malene.roland.vils.pedersen@rsyd.dk.

Rafaelsen, Soren Rafael. Department of Radiology, Vejle Hospital, Part of Lillebaelt Hospital, Kappeltoft 25, 7100, Vejle, Denmark.

Rafaelsen, Soren Rafael. Institute of Regional Health Research, University of Southern Denmark, Odense, Denmark.

Moller, Henrik. Cancer Epidemiology and Population Health, King's College London, London, UK.

Moller, Henrik. Research Unit for General Practice, Department of Public Health, Aarhus University, Aarhus, Denmark.

Vedsted, Peter. Research Unit for General Practice, Department of Public Health, Aarhus University, Aarhus, Denmark.

Osther, Palle Jorn. Department of Urology, Urological Research Center, Lillebaelt Hospital, Fredericia, Denmark.

Osther, Palle Jorn. Institute of Regional Health Research, University of Southern Denmark, Odense, Denmark.

Year of Publication
2016

569.

Genetic Association Between IL-21 Polymorphisms and Cryptorchidism in a Chinese Han Population.

Zhang D; Ma M; Pu Y; Li L; Cao G; Bai Y; Zhou B.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Genetic Testing & Molecular Biomarkers. 20(5):261-4, 2016 May.

[Journal Article]

UI: 26990619

AIMS: Increasing evidence suggests an association between interleukin gene polymorphisms and cryptorchidism. To investigate the relationship between IL-21 gene polymorphisms and cryptorchidism susceptibility, a comprehensive genetic association study in a Chinese Han population was conducted.

METHODS AND RESULTS: A total of 328 men were enrolled in a case-control study (116 cases and 212 healthy controls). Three SNPs (rs907715, rs2055979, and rs12508721) of the IL-21 gene were genotyped using the TaqMan(R) genotyping assay. For the rs2055979 C>A polymorphism, A allele carriers had significantly decreased cryptorchidism susceptibility ($p = 0.021$, odds ratios [OR] = 0.58, 95% confidence intervals [CI] = 0.37-0.93) in a dominant model (AA+CA vs. CC), a codominant model (AC vs. CC; $p = 0.029$, OR = 0.52, 95% CI = 0.31-0.85), and an overdominant model (AC vs. CC/AA; $p = 0.0087$, OR = 0.53, 95% CI = 0.33-0.86). However, no associations were identified for the rs907715 C>T and rs12508721 C>A polymorphisms. In the haplotype analyses, the C-C-T haplotype of rs907715-rs2055979-rs12508721 significantly increased the risk of cryptorchidism ($p < 0.0001$, OR = 5.251, 95% CI = 2.528-10.906).

CONCLUSIONS: The rs2055979 C>A polymorphism of the IL-21 gene is associated with cryptorchidism susceptibility, and the C allele increases the risk of cryptorchidism in a Chinese Han population.

Version ID

1

Status

MEDLINE

Authors Full Name

Zhang, Danyan; Ma, Mingfu; Pu, Yan; Li, Lianbing; Cao, Gang; Bai, Yun; Zhou, Bin.

Institution

Zhang, Danyan. 1 Department of Medical Genetics, College of Basic Medical Science, Third Military Medical University, Chongqing, China . Zhang, Danyan. 2 Key Laboratory of Birth Defects and Reproductive Health of the National Health and Family Planning Commission (Chongqing Population and Family Planning Science and Technology Research Institute), Chongqing, China .

Ma, Mingfu. 2 Key Laboratory of Birth Defects and Reproductive Health of the National Health and Family Planning Commission (Chongqing Population and Family Planning Science and Technology Research Institute), Chongqing, China .

Pu, Yan. 3 Laboratory of Molecular Translational Medicine, West China Institute of Women and Children's Health, Key Laboratory of Obstetric & Gynecologic and Pediatric Diseases and Birth Defects of Ministry of Education, West China Second University Hospital, Sichuan University, Chengdu, China .

Li, Lianbing. 2 Key Laboratory of Birth Defects and Reproductive Health of the National Health and Family Planning Commission (Chongqing Population and Family Planning Science and Technology Research Institute), Chongqing, China .

Cao, Gang. 4 Criminal Science & Technology Institute of Karamay Public Security Bureau, Karamay, Singkiang Uighur, China .

Bai, Yun. 1 Department of Medical Genetics, College of Basic Medical Science, Third Military Medical University, Chongqing, China .

Zhou, Bin. 3 Laboratory of Molecular Translational Medicine, West China Institute of Women and Children's Health, Key Laboratory of Obstetric & Gynecologic and Pediatric Diseases and Birth Defects of Ministry of Education, West China Second University Hospital, Sichuan University, Chengdu, China .

Year of Publication

2016

570.

Staged laparoscopic traction-orchiopexy for intraabdominal testis (Shehata technique): Stretching the limits for preservation of testicular vasculature.

Shehata S; Shalaby R; Ismail M; Abouheba M; Elrouby A.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of Pediatric Surgery. 51(2):211-5, 2016 Feb.

[Clinical Trial. Journal Article. Lecture. Multicenter Study]

UI: 26655212

BACKGROUND: We present the midterm results of the novel technique of two-stage laparoscopic traction orchiopexy (SLTO) for the high intraabdominal testis (IAT) for elongation of the testicular vessels without division.

PATIENTS AND METHODS: Boys with IAT proven by laparoscopy were selected for the technique between September 2009 and April 2013 in 2 Egyptian pediatric surgery units. Boys <6months or >9years were excluded. The technique entails fixation of the testis to a point one inch above and medial to the contralateral anterior superior iliac spine for 12weeks. Subdartos orchiopexy is then done at the second stage. Both stages are laparoscopically assisted.

RESULTS: SLTO was used in 124 boys (140 testis units) with IAT. Mean follow-up period was 16 (range 6-36) months. Scrotal testes were achieved in 105 (84%) of 125 followed cases. Success was correlated with patient age at operation and with distance of IAT from the internal inguinal ring with higher success in younger patients and in testes nearer to internal inguinal ring.

CONCLUSION: The novel technique of two-stage laparoscopic traction-orchiopexy is useful in IAT not amenable to one-stage laparoscopic-assisted orchiopexy; it resulted in significant elongation of testicular vessels without atrophy. It is a safe and valid alternative to two-stage laparoscopic Fowler-Stephens technique, which entails division of the main testicular vessels.

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Version ID

1

Status

MEDLINE

Authors Full Name

Shehata, Sameh; Shalaby, Rafik; Ismail, Maged; Abouheba, Mohamed; Elrouby, Ahmed.

Institution

Shehata, Sameh. Pediatric Surgery Department, Alexandria University, Egypt. Electronic address: drsamehs@yahoo.com. Shalaby, Rafik. Pediatric Surgery Department, Al-Azhar University, Egypt.

Ismail, Maged. Pediatric Surgery Department, Al-Azhar University, Egypt.

Abouheba, Mohamed. Pediatric Surgery Department, Alexandria University, Egypt.

Elrouby, Ahmed. Pediatric Surgery Department, Alexandria University, Egypt.

Comments

Comment in (CIN)

Year of Publication

2016

571.

Complete Idiopathic Hypogonadotropic Hypogonadism due to Homozygous GNRH1 Mutations in the Mutational Hot Spots in the Region Encoding the Decapeptide.

Mengen E; Tunc S; Kotan LD; Nalbantoglu O; Demir K; Gurbuz F; Turan I; Seker G; Yuksel B; Topaloglu AK.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Hormone research in paediatrics. 85(2):107-11, 2016.

[Classical Article. Journal Article. Research Support, Non-U.S. Gov't]

UI: 26595427

INTRODUCTION: Mutations of the human GNRH1 gene are an extremely rare cause of normosmic idiopathic hypogonadotropic hypogonadism (nIHH), with only 6 mutations so far described.

PATIENTS: As part of a larger study, families with IHH were screened for mutations in genes known to be associated with IHH. In family 1, a 15-year and 9-month-old boy first presented during infancy with micropenis and bilateral cryptorchidism. His pubic and axillary hair is at stage 4 and 2, respectively. His testes are 1 ml bilaterally, and his stretched penile length is 3.6 cm. In family 2, a 19-year and 2-month-old man was referred because of absence of secondary sexual characteristics. His 13-year and 8-month-old sister did not have any breast development.

RESULTS: In 3 patients from 2 independent families we identified GNRH1 mutations. In the proband from family 1, a homozygous 1-base deletion (c.87delA) leading to a frameshift mutation (p.G29GfsX12) was identified. In family 2, the affected siblings had a novel homozygous mutation of c.G92A leading to p.R31H.

CONCLUSION: Both mutations in these families are located in the region encoding the decapeptide and in the loci where the mutations have been described before. Therefore, these areas can be considered as mutational hot spots, indicating priority for routine diagnostic gene mutation analysis.

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1

Status

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Authors Full Name

Mengen, Eda; Tunc, Selma; Kotan, L Damla; Nalbantoglu, Ozlem; Demir, Korcan; Gurbuz, Fatih; Turan, Ihsan; Seker, Gul; Yuksel, Bilgin; Topaloglu, A Kemal.

Institution

Mengen, Eda. Division of Pediatric Endocrinology, Faculty of Medicine, Cukurova University, Adana, Turkey.

Year of Publication

2016

Factors Predicting Testicular Atrophy after Testicular Salvage following Torsion.

Lian BS; Ong CC; Chiang LW; Rai R; Nah SA.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

European Journal of Pediatric Surgery. 26(1):17-21, 2016 Feb.

[Journal Article]

UI: 26509312

INTRODUCTION: Testicular atrophy (TA) is a significant complication in patients who undergo salvage procedures for testicular torsion. Studies on outcome focus on factors predicting testicular viability during scrotal exploration but few assess factors predicting TA in patients who undergo salvage procedures. We assess the incidence of TA after salvage and identify associated factors.

MATERIALS AND METHODS: With ethical approval, we reviewed patients who underwent salvage for testicular torsion in our institution from 2001 to 2013. Data was collected on patient demographics, duration of pain, sonographic findings, postoperative complications, and follow-up assessment of TA (defined as difference in testicular volume > 50% compared with the contralateral testis, based on measurement by Prader orchidometer or by ultrasound). We excluded patients with torted undescended testis, those under 1 month, and those with follow-up < 6 months. Chi-square or Mann-Whitney U tests were used as appropriate with significance level < 0.05.

RESULTS: Of 85 patients who had scrotal exploration for testicular torsion, 53 had testicular salvage. Overall, 16 patients defaulted or had < 6 months follow-up, leaving 37 patients who were studied, median age 12 years (range, 0.5-16.0 years) at presentation. Median follow-up was 12.5 months (range, 6-88 months). A total of 20 patients (54%) developed TA. Median duration to TA was 12.5 months (range, 2-88 months). All had clinical evidence of atrophy by 14 months, except two who initially defaulted follow-up, but were diagnosed with TA at 35 and 88 months postoperatively when presenting with unrelated complaints. Factors associated with TA were duration of pain > 1 day ($p = 0.004$) and heterogeneous echogenicity on ultrasound ($p = 0.001$). Sonographic evidence of reduced vascularity was not predictive. Of 11 that had pain > 1 day, 10 (91%) had TA. No testes survived when pain \geq 3 days.

CONCLUSION: Half of patients with testicular torsion undergoing salvage surgery will develop testicular atrophy, even when intraoperatively assessed as viable, and should be counseled accordingly. Duration of pain > 1 day and sonographic heterogeneous echogenicity are predictive. Salvage rates are dismal when duration of symptoms exceeds 1 day.

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1

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Authors Full Name

Lian, Bertrand S Y; Ong, Caroline C P; Chiang, Li Wei; Rai, Rambha; Nah, Shireen Anne.

Institution

Lian, Bertrand S Y. Yong Loo Lin School of Medicine, National University of Singapore, Singapore, Singapore. Ong, Caroline C P. Department of Paediatric Surgery, KK Women's and Children's Hospital, Singapore, Singapore.

Chiang, Li Wei. Department of Paediatric Surgery, KK Women's and Children's Hospital, Singapore, Singapore.

Rai, Rambha. Department of Paediatric Surgery, KK Women's and Children's Hospital, Singapore, Singapore.

Nah, Shireen Anne. Department of Paediatric Surgery, KK Women's and Children's Hospital, Singapore, Singapore.

Year of Publication

2016

573.

Polygenic inheritance of cryptorchidism susceptibility in the LE/orl rat.

Barthold JS; Pugarelli J; MacDonald ML; Ren J; Adetunji MO; Polson SW; Mateson A; Wang Y; Sol-Church K; McCahan SM; Akins RE Jr; Devoto M; Robbins AK.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Molecular Human Reproduction. 22(1):18-34, 2016 Jan.

[Journal Article]

UI: 26502805

STUDY HYPOTHESIS: Susceptibility to inherited cryptorchidism in the LE/orl rat may be associated with genetic loci that influence developmental patterning of the gubernaculum by the fetal testis.

STUDY FINDING: Cryptorchidism in the LE/orl rat is associated with a unique combination of homozygous minor alleles at multiple loci, and the encoded proteins are co-localized with androgen receptor (AR) and Leydig cells in fetal gubernaculum and testis, respectively.

WHAT IS KNOWN ALREADY: Prior studies have shown aberrant perinatal gubernacular migration, muscle patterning defects and reduced fetal testicular testosterone in the LE/orl strain. In addition, altered expression of androgen-responsive, cytoskeletal and muscle-related transcripts in the LE/orl fetal gubernaculum suggest a role for defective AR signaling in cryptorchidism susceptibility.

STUDY DESIGN, SAMPLES/MATERIALS, METHODS: The long-term LE/orl colony and short-term colonies of outbred CrI:LE and CrI:SD, and inbred WKY/NcrI rats were maintained for studies. Animals were intercrossed (LE/orl X WKY/NcrI), and obligate heterozygotes were reciprocally backcrossed to LE/orl rats to generate 54 F2 males used for genotyping and/or linkage analysis. At least five fetuses per gestational time point from two or more litters were used for quantitative real-time RT-PCR (qRT-PCR) and freshly harvested embryonic (E) day 17 gubernaculum was used to generate conditionally immortalized cell lines. We completed genotyping and gene expression analyses using genome-wide microsatellite markers and single nucleotide polymorphism (SNP) arrays, PCR amplification, direct sequencing, restriction enzyme digest with fragment analysis, whole genome sequencing (WGS), and qRT-PCR. Linkage analysis was performed in Haploview with multiple testing correction, and qRT-PCR data were analyzed using ANOVA after log transformation. Imaging was performed using custom and commercial antibodies directed at candidate proteins in gubernaculum and testis tissues, and gubernaculum cell lines.

MAIN RESULTS AND THE ROLE OF CHANCE: LE/orl rats showed reduced fertility and fecundity, and higher risk of perinatal death as compared with CrI:LE rats, but there were no differences in breeding outcomes between normal and unilaterally cryptorchid males. Linkage analysis identified multiple peaks, and with selective breeding of outbred CrI:LE and CrI:SD strains for alleles within two of the most significant ($P < 0.003$) peaks on chromosomes 6 and 16, we were able to generate a non-LE/orl cryptorchid rat. Associated loci contain potentially functional minor alleles (0.25-0.36 in tested rat strains) including an exonic deletion in *Syne2*, a large intronic insertion in *Ncoa4* (an AR coactivator) and potentially deleterious variants in *Solh/Capn15*, *Ankrd28*, and *Hsd17b2*. Existing WGS data indicate that homozygosity for these combined alleles does not occur in any other sequenced rat strain. We observed a modifying effect of the *Syne2*(del) allele on expression of other candidate genes, particularly *Ncoa4*, and for muscle and hormone-responsive transcripts. The selected candidate genes/proteins are highly expressed, androgen-responsive and/or co-localized with developing muscle and AR in fetal gubernaculum, and co-localized with Leydig cells in fetal testis.

LIMITATIONS, REASONS FOR CAUTION: The present study identified multiple cryptorchidism-associated linkage peaks in the LE/orl rat, containing potentially causal alleles. These are strong candidate susceptibility loci, but further studies are needed to demonstrate functional relevance to the phenotype.

WIDER IMPLICATIONS OF THE FINDINGS: Association data from both human and rat models of spontaneous, nonsyndromic cryptorchidism support a polygenic etiology of the disease. Both the present study and a human genome-wide association study suggest that common variants with weak effects contribute to susceptibility, and may exist in genes encoding proteins that participate in AR signaling in the developing gubernaculum. These findings have potential implications for the gene-environment interaction in the etiology of cryptorchidism.

LARGE SCALE DATA: Sequences were deposited in the Rat Genome Database (RGD, <http://rgd.mcw.edu/>).

STUDY FUNDING AND COMPETING INTERESTS: This work was supported by: R01HD060769 from the Eunice Kennedy Shriver National Institute for Child Health and Human Development (NICHD), 2P20GM103446 and P20GM103464 from the National Institute of General Medical Sciences (NIGMS), and Nemours Biomedical Research. The authors have no competing interests to declare.

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Authors Full Name

Barthold, Julia Spencer; Pugarelli, Joan; MacDonald, Madolyn L; Ren, Jia; Adetunji, Modupeore O; Polson, Shawn W; Mateson, Abigail; Wang, Yanping; Sol-Church, Katia; McCahan, Suzanne M; Akins, Robert E Jr; Devoto, Marcella; Robbins, Alan K.

Institution

Barthold, Julia Spencer. Pediatric Urology Research Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA

julia.barthold@nemours.org. Pugarelli, Joan. Pediatric Urology Research Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

MacDonald, Madolyn L. Center for Bioinformatics and Computational Biology, Delaware Biotechnology Institute, University of Delaware, Newark, DE, USA.

Ren, Jia. Center for Bioinformatics and Computational Biology, Delaware Biotechnology Institute, University of Delaware, Newark, DE, USA.

Adetunji, Modupeore O. Center for Bioinformatics and Computational Biology, Delaware Biotechnology Institute, University of Delaware, Newark, DE, USA.

Polson, Shawn W. Center for Bioinformatics and Computational Biology, Delaware Biotechnology Institute, University of Delaware, Newark, DE, USA.

Mateson, Abigail. Pediatric Urology Research Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

Wang, Yanping. Pediatric Urology Research Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

Sol-Church, Katia. Biomolecular Core Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

McCahan, Suzanne M. Bioinformatics Core, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

Akins, Robert E Jr. Tissue Engineering and Regenerative Medicine Research Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

Devoto, Marcella. Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA Department of Biostatistics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA Department of Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA Department of Molecular Medicine, Sapienza University, Rome, Italy.

Robbins, Alan K. Pediatric Urology Research Laboratory, Nemours Biomedical Research/Alfred I. duPont Hospital for Children, Wilmington, DE, USA.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4694052>

Year of Publication

2016

574.

Our Experience in Transcrotal Orchidopexy in Children Affected by Palpable Undescended Testis.

Arena S; Impellizzeri P; Perrone P; Scalfari G; Centorrino A; Turiaco N; Russo T; Antonuccio P; Romeo C.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid
MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present
European Journal of Pediatric Surgery. 26(1):13-6, 2016 Feb.

[Evaluation Study. Journal Article]

UI: 26394369

INTRODUCTION: Classically, surgical approach for palpable undescended testis (pUDT) consists is an inguinal orchidopexy. In fact, a double incision allows an adequate mobilization of the spermatic cord and an easy dissection of a patent processus vaginalis and also to perform a subdartos pouch. For reduce potential mobility of the inguinal approach, in 1989 Bianchi and Squire proposed a transcrotal orchidopexy, using a high scrotal incision. We report our experiences and retrospectively evaluate the feasibility and postoperative success of the transcrotal approach for treatment of pUDT.

PATIENTS AND METHODS: From January 2012 to June 2014, 217 patients, affected by pUDT were treated at our Institution, for a total of 231 orchidopexies (203 monolateral pUDT, 14 bilateral pUDT). Patients, in whom, under anesthesia, the testis could be moved to the neck of the scrotum, have been treated with a transcrotal approach using a high scrotal incision. All patients have been clinically followed up at 1 week and at 1, 2, 3, and 6 months, at 1 year, and then annually and using scrotal sonography with color-Doppler at 2, 3, and 6 months.

RESULTS: A total of 205 pUDT (88.7%) were considered eligible for transcrotal. Eight (3.9%) pUDT, that were first approached transcrotally, have been converted to inguinal approach. At follow-up, two moderate scrotal hematomas and one inguinal hernia were noted. No recurrence or testicular atrophy was showed in transcrotal approached testes, while 2 recurrences out of 26 procedures (7.7%) were displayed after inguinal orchidopexy.

CONCLUSIONS: In our experience, transcrotal approach is possible in almost 90% of pUDT. No major complication, such as recurrence or testicular atrophy, has been complained. Just 3 out of the 205 cases (1.5%) reported minor complications and 1 of which required a successful day-case procedure. Our data confirm that transcrotal orchidopexy can be considered effective, safe, and with a success rate being equivalent or better to classical inguinal approach.

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Authors Full Name

Arena, Salvatore; Impellizzeri, Pietro; Perrone, Patrizia; Scalfari, Gianfranco; Centorrino, Antonio; Turiaco, Nunzio; Russo, Tiziana; Antonuccio, Pietro; Romeo, Carmelo.

Institution

Arena, Salvatore. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Impellizzeri, Pietro. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Perrone, Patrizia. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Scalfari, Gianfranco. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Centorrino, Antonio. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Turiaco, Nunzio. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Russo, Tiziana. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Antonuccio, Pietro. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy. Romeo, Carmelo. Department of Paediatric, Gynaecological, Microbiological and Biomedical Sciences, University of Messina, Messina, Italy.

Year of Publication

2016

575.

OUTCOMES OF POSTERIOR SAGITTAL ANORECTOPLASTY FOR HIGH ANORECTAL MALFORMATION IN BENIN CITY, NIGERIA.

Osagie TO; Aisien E; Osifo OD.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Journal of the West African Colleges of Surgeons. 6(1):16-30, 2016 Jan-Mar.

[Journal Article]

UI: 28344935

BACKGROUND: Surgical treatment for high anorectal malformations has evolved over the years with introduction of posterior sagittal anorectoplasty in the early 80s. Posterior sagittal

anorectoplasty is being performed in many centres which necessitates a review of its outcomes in Benin City, Nigeria.

AIM: To report a 10-year outcome and experience gained with posterior sagittal anorectoplasty for children diagnosed with high anorectal malformation.

METHODS: A retrospective analyses of the records of all children with high anorectal malformation and managed with posterior sagittal anorectoplasty between April 2006 and March 2016 at the University of Benin Teaching Hospital.

RESULTS: A total of 96 children were managed for anorectal malformation during the period. High anorectal malformation accounted for 33 (34.4%) cases, the intermediate 15 (15.6%) and the low types were 48 (50%). The 33 radiologically confirmed high type were 19 males and 14 females with a male/female ratio of 1.3: 1. They were aged between 2 days and 4 years with a mean of 6.8 +/- 3 months. A child each had additional prune belly syndrome, multiple limbs anomalies and unilateral undescended testis. Recto-bladder neck/recto-prostatic and recto-vaginal fistulae were recorded in 31 (94%) children. Five (15%) clinically stable neonates had primary posterior sagittal anorectoplasty without colostomy which was well tolerated. The majority, 28 (85%), had conventional posterior sagittal anorectoplasty that involves initial colostomy. Minor postoperative morbidities recorded in 10 (30.3%) children included superficial wound infection in 3 (9%), anal stenosis in 3 (9%) and fecal incontinence in 2 (6%) children which resolved on conservative treatment while 2 (6%) with rectal mucosal prolapse required refashioning. The functional clinical anal outcomes of posterior sagittal anorectoplasty recorded showed that the majority 18 (54.5%) of children were continent while 4 (12.1%) had voluntary bowel controls corresponding with their ages. Anal stenosis in the 3 and incontinence in the 2 children were the common anal dysfunctions recorded. The child with prune belly syndrome had breakdown of colostomy closure which resulted in the one (3%) death recorded.

CONCLUSION: Anorectal malformation was common in this setting during this study with a large proportion of the children diagnosed with the high type and were managed with good outcome using posterior sagittal anorectoplasty.

Version ID

1

Status

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Authors Full Name

Osagie, T O; Aisien, E; Osifo, O D.

Institution

Osagie, T O. DEPARTMENT OF SURGERY, UNIVERSITY OF BENIN TEACHING HOSPITAL, BENIN CITY, NIGERIA. Aisien, E. DEPARTMENT OF SURGERY, UNIVERSITY OF BENIN TEACHING HOSPITAL, BENIN CITY, NIGERIA.

Osifo, O D. DEPARTMENT OF SURGERY, UNIVERSITY OF BENIN TEACHING HOSPITAL,
BENIN CITY, NIGERIA.

PMC Identifier

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Year of Publication

2016

576.

Gonadal response after a single-dose stimulation test with recombinant human chorionic gonadotropin (rhCG) in patients with isolated prepubertal cryptorchidism.

Oliveira LR; Homma TK; Woloszynek RR; Brito VN; Longui CA.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Basic & Clinical Andrology. 26:13, 2016.

[Journal Article]

UI: 27800162

BACKGROUND: The evaluation of prepubertal gonadal Leydig cells secretion requires gonadotropin stimulation. Urinary hCG (human chorionic gonadotropin) is currently unavailable in many countries, however, recombinant hCG (rhCG) can be used. Our aim was to evaluate rhCG-stimulated testicular hormones in a group of patients with cryptorchidism.

METHODS: We evaluated 31 prepubertal boys (age range, 0.75-9.0 years) presenting with unilateral (n = 24) or bilateral (n = 7) cryptorchidism. Patients with other genital abnormalities, previous use of hCG or testosterone or previous surgeries were excluded. Blood samples were obtained at baseline and 7 days after a single subcutaneous dose of rhCG (Ovidrel R 250 mcg) to measure the testosterone, DHT (dihydrotestosterone), AMH (anti-Mullerian hormone), and inhibin B levels.

RESULTS: rhCG stimulation significantly increased testosterone levels from 10 ng/dl to 247.8 +/- 135.8 ng/dl, increased DHT levels from 4.6 +/- 0.8 to 32.3 +/- 18.0 ng/dl, and increased the T/DHT ratio from 2.2 +/- 0.4 to 8.0 +/- 3.5. There was also a significant increase in inhibin B (from 105.8 +/- 65.2 to 132.4 +/- 56.1 pg/ml; p < 0.05) and AMH levels (from 109.4 +/- 52.6 to 152.9 +/- 65.2 ng/ml; p < 0.01) after the rhCG stimulation.

CONCLUSIONS: In this cohort, hormonal responses can be elicited after the rhCG stimulation test, suggesting that rhCG is a promising stimulation test to replace the urinary hCG test during the evaluation of gonadal Leydig cells function. The clinical applicability and adequate performance of rhCG testing must be investigated in future studies.

Version ID

1

Status

PubMed-not-MEDLINE

Authors Full Name

Oliveira, Leticia Ribeiro; Homma, Thais Kataoka; Woloszynek, Renata Reis; Brito, Vinicius Nahime; Longui, Carlos Alberto.

Institution

Oliveira, Leticia Ribeiro. Pediatric Endocrinology Unit, Pediatrics Department, Irmandade da Santa Casa de Misericordia de Sao Paulo, and Santa Casa de Sao Paulo School of Medical Sciences, Rua Dr. Cesario Mota Jr, 112, Vila Buarque, Sao Paulo, CEP 01221-020 Brazil.

Homma, Thais Kataoka. Pediatric Endocrinology Unit, Pediatrics Department, Irmandade da Santa Casa de Misericordia de Sao Paulo, and Santa Casa de Sao Paulo School of Medical Sciences, Rua Dr. Cesario Mota Jr, 112, Vila Buarque, Sao Paulo, CEP 01221-020 Brazil.

Woloszynek, Renata Reis. Developmental Endocrinology Unit, University of Sao Paulo Faculty of Medicine Clinics Hospital - USP, Sao Paulo, Brazil.

Brito, Vinicius Nahime. Developmental Endocrinology Unit, University of Sao Paulo Faculty of Medicine Clinics Hospital - USP, Sao Paulo, Brazil.

Longui, Carlos Alberto. Pediatric Endocrinology Unit, Pediatrics Department, Irmandade da Santa Casa de Misericordia de Sao Paulo, and Santa Casa de Sao Paulo School of Medical Sciences, Rua Dr. Cesario Mota Jr, 112, Vila Buarque, Sao Paulo, CEP 01221-020 Brazil.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5084384>

Other Abstract

Publisher: L'évaluation de la sécrétion des cellules gonadiques de Leydig prépubères nécessite une stimulation par les gonadotrophines. La gonadotrophine chorionique humaine (hCG) urinaire est actuellement indisponible dans de nombreux pays; toutefois, l'hCG recombinante (rhCG) peut être utilisée. Notre objectif était d'évaluer les hormones testiculaires sous stimulation par rhCG dans un groupe de patients qui présentaient une cryptorchidie.; Language: French

Year of Publication

2016

577.

Anti-Mullerian hormone as a marker of steroid and gonadotropin action in the testis of children and adolescents with disorders of the gonadal axis. [Review]

Edelstein NY; Grinspon RP; Schteingart HF; Rey RA.

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International Journal of Pediatric Endocrinology. 2016:20, 2016.

[Review. Journal Article]

UI: 27799946

In pediatric patients, basal testosterone and gonadotropin levels may be uninformative in the assessment of testicular function. Measurement of serum anti-Mullerian hormone (AMH) has become increasingly widespread since it provides information about the activity of the male gonad without the need for dynamic tests, and also reflects the action of FSH and androgens within the testis. AMH is secreted in high amounts by Sertoli cells from fetal life until the onset of puberty. Basal AMH expression is not dependent on gonadotropins or sex steroids; however, FSH further increases and testosterone inhibits AMH production. During puberty, testosterone induces Sertoli cell maturation, and prevails over FSH on AMH regulation. Therefore, AMH production decreases. Serum AMH is undetectable in patients with congenital or acquired anorchidism, or with complete gonadal dysgenesis. Low circulating levels of AMH may reflect primary testicular dysfunction, e.g. in certain patients with cryptorchidism, monorchidism, partial gonadal dysgenesis, or central hypogonadism. AMH is low in boys with precocious puberty, but it increases to prepubertal levels after successful treatment. Conversely, serum AMH remains at high, prepubertal levels in boys with constitutional delay of puberty. Serum AMH measurements are useful, together with testosterone determination, in the diagnosis of patients with ambiguous genitalia: both are low in patients with gonadal dysgenesis, including ovotesticular disorders of sex development, testosterone is low but AMH is in the normal male range or higher in patients with disorders of androgen synthesis, and both hormones are normal or high in patients with androgen insensitivity. Finally, elevation of serum AMH above normal male prepubertal levels may be indicative of rare cases of sex-cord stromal tumors or Sertoli cell-limited disturbance in the McCune Albright syndrome.

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1

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Authors Full Name

Edelsztein, Nadia Y; Grinspon, Romina P; Schteingart, Helena F; Rey, Rodolfo A.

Institution

Edelsztein, Nadia Y. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), CONICET - FEI - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina ; Departamento de Ecologia, Genetica y Evolucion, Facultad de Ciencias Exactas y Naturales, Universidad de Buenos Aires, Buenos Aires, Argentina. Grinspon, Romina P. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), CONICET - FEI - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina.

Schteingart, Helena F. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), CONICET - FEI - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina.

Rey, Rodolfo A. Centro de Investigaciones Endocrinologicas "Dr. Cesar Bergada" (CEDIE), CONICET - FEI - Division de Endocrinologia, Hospital de Ninos Ricardo Gutierrez, Buenos Aires, Argentina ; Departamento de Biologia Celular, Histologia, Embriologia y Genetica, Facultad de Medicina, Universidad de Buenos Aires, Buenos Aires, Argentina.

PMC Identifier

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2016

578.

A Unique Case of Intraabdominal Polyorchidism: A Case Study.

Otero J; Ben-Yakar N; Alemayehu B; Kozusko SD; Borao F; Vates Iii TS.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

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Case Reports Urology. 2016:2729614, 2016.

[Journal Article]

UI: 27722006

Background. Polyorchidism, alternatively supernumerary testes (SNT), is a condition where an individual is born with more than two testicles. This congenital anomaly is quite rare and the

literature has described various presentations. Questions/Purposes. To our knowledge, this presentation of polyorchidism has yet to be described in the literature. The goal of this case study is to add to the pediatric, general, and urologic surgery's body of knowledge of the subject matter. Case Study. A nine-month-old boy was admitted for an impalpable right testis and phimosis. At the time of surgical exploration, there appeared to be polyorchid testis on the right-hand side, with three masses that potentially appeared to be undescended testes. Discussion. Proponents of a conservative approach argue that infertility is common in patients with polyorchidism and, by preserving a potentially functional SNT, there may be improved spermatogenesis. When performing definitive surgical treatment, meticulous intra-abdominal and intrainguinal exploration must be undertaken. Orchiopexy should be performed to reduce the chances of torsion, malignancy, and infertility. Conclusion. Our case is important to the literature as it is the first known case of polyorchidism with 3 SNT on the right side, located intra-abdominally, and in a patient less than 1 year of age.

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1

Status

PubMed-not-MEDLINE

Author NameID

Otero, Javier; ORCID: <https://orcid.org/0000-0003-2436-5138> Ben-Yakar, Natalie; ORCID: <https://orcid.org/0000-0003-3585-6354>

Authors Full Name

Otero, Javier; Ben-Yakar, Natalie; Alemayehu, Biruk; Kozusko, Steven D; Borao, Frank; Vates Iii, Thomas S.

Institution

Otero, Javier. Department of Surgery, Monmouth Medical Center, Barnabas Health, Long Branch, NJ 07740, USA. Ben-Yakar, Natalie. Drexel University College of Medicine, Philadelphia, PA 19129, USA.

Alemayehu, Biruk. St. George's University School of Medicine, St. George's, Grenada.

Kozusko, Steven D. Department of Surgery, Monmouth Medical Center, Barnabas Health, Long Branch, NJ 07740, USA.

Borao, Frank. Department of Surgery, Monmouth Medical Center, Barnabas Health, Long Branch, NJ 07740, USA.

Vates Iii, Thomas S. Department of Urology, Monmouth Medical Center, Barnabas Health, Long Branch, NJ 07740, USA.

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2016

579.

A Case of Cryptorchidism with Ipsilateral Congenital Unilateral Absence of the Vas Deferens and Contralateral Renal Agenesis.

Yu YD; Hong YK.

OVID Medline Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Case Reports Urology. 2016:2379793, 2016.

[Journal Article]

UI: 27597925

Introduction and Aims. Congenital absence of the vas deferens is an uncommon anomaly and this clinical condition is responsible for up to 1-2% of male infertility. It can be either unilateral or bilateral and the associated anomalies include cryptorchidism, seminal vesicles and ejaculatory ducts anomalies, and renal anomalies such as renal agenesis. We hereby present a case of congenital unilateral absence of vas deferens, which was found incidentally during an evaluation of undescended testis in a patient with ipsilateral renal agenesis. Case Presentation. A 10-month-old boy was referred to the urology clinic with an undescended right testis. Preoperative abdominal ultrasonography showed agenesis of the right kidney and the absence of right vas deferens and epididymis was confirmed during laparoscopic orchiectomy performed due to short right spermatic cord. There were no other concomitant anomalies of the genitourinary system observed in evaluation. Conclusion . Congenital unilateral absence of the vas deferens with cryptorchidism and renal agenesis is a rare diagnostic entity. Cryptorchidism or absent vas deferens found incidentally should lead the physician to evaluate the status of the contralateral vas deferens and conduct a renal tract ultrasound study.

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1

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Author NameID

Hong, Young Kwon; ORCID: <https://orcid.org/0000-0002-4344-6788>

Authors Full Name

Yu, Young Dong; Hong, Young Kwon.

Institution

Yu, Young Dong. Department of Urology, CHA Bundang Medical Center, CHA University, Seongnam 13497, Republic of Korea. Hong, Young Kwon. Department of Urology, CHA Bundang Medical Center, CHA University, Seongnam 13497, Republic of Korea.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4997067>

Year of Publication

2016

580.

Congenital Erythropoietic Porphyria with Undescended Testis.

Arora S; Harith AK; Sodhi N.

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MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present

Indian Journal of Dermatology. 61(4):467, 2016 Jul-Aug.

[Journal Article]

UI: 27512208

Hereditary porphyrias are a group of metabolic disorders of heme biosynthesis pathway that are characterized by acute neurovisceral symptoms, skin lesions, or both. Congenital erythropoietic porphyria (CEP) is an extremely rare disease with a mutation in the gene that codes for uroporphyrinogen III synthase leading to accumulation of porphyrin in different tissues and marked cutaneous photosensitivity. We report a case of CEP with infancy onset blistering, photosensitivity, red colored urine, and teeth along with scarring. Examination revealed an undescended testis of the left side. Mutation analysis revealed mutation in the uroporphyrinogen III synthase gene (UROS) resulting in c. 56 A > G (Tyr19Cys). The presence of undescended testis with a rare mutation in a case of CEP which itself is an extremely rare condition make the case interesting.

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1

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Authors Full Name

Arora, Sandeep; Harith, Arun Kumar; Sodhi, Neha.

Institution

Arora, Sandeep. Department of Dermatology, Army College of Medical Sciences, New Delhi, India. Harith, Arun Kumar. Department of Biochemistry, Army College of Medical Sciences, New Delhi, India.

Sodhi, Neha. Department of Dermatology, Army College of Medical Sciences, New Delhi, India.

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Year of Publication

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581.

For Better Orchiopexy, Processus Vaginalis Should Be Dissected and a High Ligation Should Be Performed.

Sonmez K; Karabulut R; Turkyilmaz Z; Kaya C; Pehlivan Y; Basaklar AC.

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Rambam Maimonides Medical Journal. 7(3), 2016 Jul 28.

[Journal Article]

UI: 27487307

OBJECTIVE: Data on the prevalence of patent processus vaginalis (PPV) and hernia in patients with cryptorchidism are controversial. While some pediatric surgeons do not dissect the processus vaginalis (PV), most prefer to do so to prevent hernia formation and to achieve an effective orchiopexy outcome. This study was performed to evaluate the importance of dissection and high ligation of the PV during treatment of undescended testis (UT).

METHODS: The clinical findings and surgical procedures of 55 patients with UT were retrospectively investigated.

RESULTS: The mean patient age was 2.5 (range 1.0-12.0) years. Non-palpable testis (NPT) was located on the right and left side in 39 and 16 patients, respectively. Ultrasonography revealed no testis in 10 patients and an atrophic testis in 7 patients. Seven patients had a parent with an inguinal hernia, and the silk sign or a PPV was detected during inguinoscrotal examination in 22

patients. Undescended testis repair was performed by an inguinal approach in all patients. The inguinal canal was opened in all patients; 42 patients had a wider-than-normal internal ring (>2.5 cm), and the posterior wall of the inguinal canal was consequently weakened. Two-stage orchiopexy was performed in 2 patients, and 15 underwent the Prentiss maneuver. In the remaining patients, the dissection was easily done, and the orchiopexy was performed without any difficulty. Scrotal edema and wound infection occurred in five and two patients, respectively. One patient presented with an atrophic testis, and three had recurrent UT. Inguinal hernia was not observed in any of the patients during the study period, and all procedures were performed on an outpatient basis.

CONCLUSION: High ligation of the PV is an effective method for successful orchiopexy and prevention of inguinal hernia in patients with NPT and UT.

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Authors Full Name

Sonmez, Kaan; Karabulut, Ramazan; Turkyilmaz, Zafer; Kaya, Cem; Pehlivan, Yildiz; Basaklar, A Can.

Institution

Sonmez, Kaan. Department of Pediatric Surgery, Medical Faculty of Gazi University, Ankara, Turkey. Karabulut, Ramazan. Department of Pediatric Surgery, Medical Faculty of Gazi University, Ankara, Turkey. Turkyilmaz, Zafer. Department of Pediatric Surgery, Medical Faculty of Gazi University, Ankara, Turkey. Kaya, Cem. Department of Pediatric Surgery, Medical Faculty of Gazi University, Ankara, Turkey. Pehlivan, Yildiz. Department of Pediatric Surgery, Medical Faculty of Gazi University, Ankara, Turkey. Basaklar, A Can. Department of Pediatric Surgery, Medical Faculty of Gazi University, Ankara, Turkey.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5001792>

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2016

582.

A novel PAX3 mutation in a Japanese boy with Waardenburg syndrome type 1.

Yoshida Y; Doi R; Adachi K; Nanba E; Kodani I; Ryoike K.

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Human Genome Variation. 3:16005, 2016.

[Journal Article]

UI: 27081571

Waardenburg syndrome type 1 (WS1) is a rare autosomal dominant disorder characterized by hair hypopigmentation, abnormal iris pigmentation, and congenital hearing loss. WS1 is caused by mutations in paired box gene 3 (PAX3). We identified a novel PAX3 mutation (c.1107 C>G, p.Ser369Arg) in a Japanese WS1 patient showing abnormal right iris pigmentation, right-sided congenital hearing loss, synophrys, incomplete left cleft lip, and cryptorchidism.

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Authors Full Name

Yoshida, Yu; Doi, Rieko; Adachi, Kaori; Nanba, Eiji; Kodani, Isamu; Ryoike, Kazuo.

Institution

Yoshida, Yu. Division of Oral and Maxillofacial Biopathological Surgery, Department of Medicine of Sensory and Motor Organs, School of Medicine, Tottori University Faculty of Medicine, Yonago, Japan. Doi, Rieko. Division of Oral and Maxillofacial Biopathological Surgery, Department of Medicine of Sensory and Motor Organs, School of Medicine, Tottori University Faculty of Medicine, Yonago, Japan.

Adachi, Kaori. Division of Functional Genomics, Research Center for Bioscience and Technology, Tottori University, Yonago, Japan.

Nanba, Eiji. Division of Functional Genomics, Research Center for Bioscience and Technology, Tottori University, Yonago, Japan.

Kodani, Isamu. Division of Oral and Maxillofacial Biopathological Surgery, Department of Medicine of Sensory and Motor Organs, School of Medicine, Tottori University Faculty of Medicine, Yonago, Japan.

Ryoike, Kazuo. Division of Oral and Maxillofacial Biopathological Surgery, Department of Medicine of Sensory and Motor Organs, School of Medicine, Tottori University Faculty of Medicine, Yonago, Japan.

PMC Identifier

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4775770>

Year of Publication

2016

583.

The use of ultrasound guidance for perioperative neuraxial and peripheral nerve blocks in children

Guay, Joanne. Suresh, Santhanam. Kopp, Sandra. Institution Joanne Guay .TI The use of ultrasound guidance for perioperative neuraxial and peripheral nerve blocks in children.

EBM Reviews - Cochrane Database of Systematic Reviews

Cochrane Database of Systematic Reviews. 2, 2019.

[Systematic Review]

AN: 00075320-100000000-09840

Background

The use of ultrasound guidance for regional anaesthesia has become popular over the past two decades. However, it is not recognized by all experts as an essential tool, perhaps because it is unclear whether ultrasound reduces the risk of severe neurological complications, and the cost of an ultrasound machine (USD 22,000) is substantially higher than the cost of other tools. This review was published in 2016 and updated in 2019.

Objectives

To determine whether ultrasound guidance offers any clinical advantage when neuraxial and peripheral nerve blocks are performed in children in terms of decreasing failure rate or the rate of complications.

Search methods

We searched CENTRAL, MEDLINE, Embase, and two trial registers up to March 2018 together with reference checking to identify additional studies and contacted study authors to obtain additional trial information.

Selection criteria

We included all parallel randomized controlled trials that evaluated the effects of ultrasound guidance used when a regional blockade technique was performed in children. We included studies performed in children (\leq 18 years of age) undergoing any type of surgical procedure

(open or laparoscopic), for which a neuraxial (spinal, epidural, caudal, or combined spinal and epidural) or peripheral nerve block (any peripheral nerve block including fascial (fascia iliaca, transversus abdominis plane, rectus sheath blocks) or perivascular blocks), for surgical anaesthesia (alone or in combination with general anaesthesia) or for postoperative analgesia, was performed with ultrasound guidance. We excluded studies in which regional blockade was used to treat chronic pain.

Data collection and analysis

We used the standard methodological procedures expected by Cochrane. Our primary outcomes were failed blocks, pain scores at one hour after surgery, and block duration. Secondary outcomes included time to perform the block, number of needle passes, and minor and major complications. We used GRADE to assess the quality of evidence for each outcome.

Main results

We included 33 trials with a total of 2293 participants from 0.9 to 12 (mean or median) years of age. Most trials were at low risk of selection, detection, attrition, and reporting bias, however the lack of blinding of participants and personnel caring for participants resulted in 25 trials being judged as at high or unclear risk of bias. We identified five ongoing trials.

Authors' conclusions

Ultrasound guidance for regional blockade in children probably decreases the risk of failed block. It increases the duration of the block and probably decreases pain scores at one hour after surgery. There may be little or no difference in the risks of some minor complications. The five ongoing studies may alter the conclusions of the review once published and assessed.

584.

Allopurinol for women in pregnancy for neuroprotection of the fetus

Martinello, Kathryn A. Shepherd, Emily. Middleton, Philippa. Crowther, Caroline A. Institution

Kathryn A Martinello .TI Allopurinol for women in pregnancy for neuroprotection of the fetus.

EBM Reviews - Cochrane Database of Systematic Reviews

Cochrane Database of Systematic Reviews. 12, 2017.

[Protocol]

AN: 00075320-100000000-11300

This is a protocol for a Cochrane Review (Intervention). The objectives are as follows:

To assess the effectiveness and safety of maternal allopurinol when used for neuroprotection of the fetus.