Diaper Dilemmas:
Evaluating Newborn Genitalia

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Disclosure

• I have no financial interest in or affiliation with any commercial supporter to disclose.
Learning Objectives

1. Describe treatment of phimosis and penile adhesions

1. Recognize differences between undescended vs retractile testes

1. Identify newborn genitalia abnormalities related to disorders of sex development
Topics

• Phimosis: physiologic vs. pathologic
• Penile adhesions vs. skin bridge
• Hypospadias
• Undescended testicle vs. retractile testis
• Urethral prolapse
3 yo boy whose foreskin does not retract...

- Mom states that she is worried she cannot retract her son’s foreskin
- Has noticed a white ball ?pus underneath the skin
- Wants to know if her son needs a circumcision
- No history of UTIs
- No ballooning with urination
- No history of penile skin infections
Physical Exam
Physiologic Phimosis

• Natural adhesions between the glans and the prepuce
• Preputial phimotic ring
• 2 factors contribute to separation of penile skin from glans
  1. Epithelial debris = smegma
  2. Intermittent erections
• Retractability increases with age
  • 90% of 3 yo boy are able to retract their foreskin
  • Less than 1% of boys by 17 years of age are unable to retract
Pathologic Phimosis

Ballooning

Balanitis xerotica obliterans (BXO)

Balanoposthitis
Treatment

• Indications to enhance preputial retractability
  • persistent primary phimosis
  • secondary phimosis
  • balanitis/posthitis
  • BXO
  • UTIs

• Corticosteroid cream 2xday x 4-8 weeks (43-86% effective)
Surgical Options

- Circumcision
- Prepucioplasty
AAP Statement on Circumcision

“Systematic evaluation of English-language peer-reviewed literature from 1995 through 2010 indicates that preventive health benefits of elective circumcision of male newborns outweigh the risks of the procedure.”

“Benefits include significant reductions in the risk of urinary tract infection in the first year of life and, subsequently, in the risk of heterosexual acquisition of HIV and the transmission of other sexually transmitted infections.”
## Circumcision Controversy

<table>
<thead>
<tr>
<th>Pro Circumcision</th>
<th>Neutral</th>
<th>Against Circumcision</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prevention of: penile cancer</td>
<td>Cosmetic</td>
<td>Risks of surgery: bleeding, infection, tissue loss</td>
</tr>
<tr>
<td>UTIs in babies</td>
<td>Sexual pleasure/function</td>
<td>Pain</td>
</tr>
<tr>
<td>STDs</td>
<td>Religious/cultural</td>
<td>Meatal stenosis</td>
</tr>
<tr>
<td>Future morbidity</td>
<td></td>
<td>Buried penis</td>
</tr>
</tbody>
</table>
3 yo boy whose foreskin does not retract...

- Healthy foreskin with no evidence of scarring
- Options: observe, course of corticosteroid cream
- Counsel on no medical need for surgical intervention
Post Circumcision Adhesions vs. Skin Bridge
1 day old boy with a hypospadias

• Born full term
• No medical problems
• No GU family history
• Parents want a circumcision
Physical Exam
Mom asks what is hypospadias?

• Association of three anomalies of the penis:

  • Abnormal *ventral opening* of the urethral meatus that may be located anywhere from the ventral aspect of the glans penis to the perineum

  • Abnormal ventral curvature of the penis (*chordee*)

  • Abnormal *distribution of foreskin* (dorsal hood/ventral deficiency)
Epidemiology

What is the prevalence of HPS?
→ 1 in 250 live male births

What percent of patients with HPS have a father with HPS?
→ 6%-8%

Is there an increased prevalence of HP in twins?
→ 8.5 fold increase in one of monozygotic twins compared to singletons
Etiology

• Multifactorial, polygenic model

• Endocrine disruptor: inadequate androgen/insensitivity of target tissues

• Arrest of development
What are the indications for surgery?

• Provide ability to micturate in standing position

• Effectively inseminate

• Cosmesis
What are the complications of HPS repairs?

- Bleeding/Hematoma
- Meatal Stenosis
- Urethrocutaneous Fistula
- Infection
- Urethral Diverticulum
- Balanitis Xerotica Obliterans
- Recurrent Penile Curvature
- Urethral Stricture
- Repair Breakdown
1 day old boy with a hypospadias

- Circumcision contraindicated
- Normal routine diaper care
- Plan for referral to urologist at 3-4 months of age
- Hypospadias repair between 6-9 months of age
6 mo boy with a right undescended testicle...

• Mom has not noticed testicle down
• No scrotal swelling or groin swelling
• Born at 36 weeks
• No medical problems
• Father has history of undescended testicle as child
Physical Exam

• GU
  • Circumcised penis, normal meatus
  • Left testicle down in scrotum, normal size and consistency
  • Right testicle just below external inguinal ring, unable to milk testicle down into scrotum
Definitions

- **Cryptorchidism**
  - Kryptos = hidden
  - Orchis = testis
  - Anywhere between the abdominal cavity and outside the scrotum
  - Along normal path of descent vs. ectopic

- **Retractile Testis**
  - Testis can be milked down into scrotum and remains there temporarily
  - Overactive cremasteric reflex
  - Risk of ascent
Definitions

• Vanishing testicle
  • Diagnosed by blind ending vessels and vas deferens
  • Torsion or compromising vascular event

• Testicular nubbin
  • Also thought to be due to some perinatal compromising vascular event
  • Usually associated with hypoplastic/atretic vessels through internal ring
  • 13% have viable residual testicular elements
    • Residual tubules
    • Germ cells
    • CIS has been reported, ? true risk of malignancy
Incidence

• Isolated UDT – 3% of full-term newborn boys
• Premature infants – 30%
• Bilateral UDT – 1.6-1.9% newborn boys
Natural History

- 70-77% of UDT will spontaneously descend, usually by 3 months
  - Only 6-9% of UDT descend spontaneously beyond 6 months of age (Berkowitz et al. 1993)

- Factors predicting descent
  - Low birth weight
  - Bilateral UDT
  - Normal scrotal size
  - Testes closer to normal scrotal position
Risk Factors

• Low birth weight – independent of gestational age
• Prematurity
• Genetic susceptibility
• Congenital disorders
Genetic Susceptibility

- Cryptorchidism concordance based on family relationships
  - 1.8% in unrelated males
  - 4.3% in half-brothers
  - 7.5% in full brothers
  - 16.7% in dizygotic twins
  - 26.7% in monozygotic twins

- INSL3, its receptor RXFP2, ESR1

Jensen et al. 2010
Consequences of UDT

• Neoplasia
• Infertility
• Hernia
• Testicular torsion
Consequences of UDT

• Neoplasia

• Infertility

• Hernia

• Testicular torsion
# UDT – Relative Risk of Testis Cancer

## Cryptorchidism and Testicular Cancer: Separating Fact From Fiction

**Table 2. Testis cancer in UDT and contralateral normally descended testis, and association between orchiopexy and testis cancer risk**

<table>
<thead>
<tr>
<th>References</th>
<th>Country</th>
<th>Period</th>
<th>Type</th>
<th>No. Pts</th>
<th>Calculated Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pinczowski et al.</td>
<td>Sweden</td>
<td>1965–1983</td>
<td>Prospective database</td>
<td>2,918</td>
<td>RR 7.4</td>
</tr>
<tr>
<td>Moller et al.</td>
<td>Denmark</td>
<td>1986–1988*</td>
<td>Prospective case-control</td>
<td>803</td>
<td>RR 3.61</td>
</tr>
<tr>
<td>Prener et al.</td>
<td>Denmark</td>
<td>1941–1957</td>
<td>Retrospective case-control</td>
<td>549</td>
<td>Adjusted RR 5.2</td>
</tr>
<tr>
<td>Swedlow et al.</td>
<td>United Kingdom</td>
<td>1951–1964</td>
<td>Prospective database</td>
<td>1,124</td>
<td>RR 7.5</td>
</tr>
<tr>
<td>Coupland et al.</td>
<td>United Kingdom</td>
<td>1984–1986</td>
<td>Retrospective case-control</td>
<td>794</td>
<td>OR 3.82</td>
</tr>
<tr>
<td>Kanto et al.</td>
<td>Japan</td>
<td>1975–2002</td>
<td>Retrospective database</td>
<td>240</td>
<td>RR 5.4</td>
</tr>
<tr>
<td>Cortes et al.</td>
<td>Denmark</td>
<td>1971–2004</td>
<td>Prospective database</td>
<td>1,466</td>
<td>RR 4</td>
</tr>
<tr>
<td>Pettersson et al.</td>
<td>Sweden</td>
<td>1965–2000</td>
<td>Prospective database</td>
<td>16,983</td>
<td>IR 2.75</td>
</tr>
</tbody>
</table>

*RR 2.75 - 8*

- RR 2.75: Relative Risk of Testis Cancer
- RR 3.7: Age at correction-risk
- RR 3.61: Age at correction-risk
- RR 5.2: Adjusted RR
- OR 3.82: Not provided
- RR 5.4: Not provided, trend toward more seminoma in late (after age 15 yrs) or uncorrected UDT
- RR 1: Age at orchiopexy not discussed
- IR 2.75: Not provided
- RR 4: Not provided
- RR 2.14: None
Infertility

Lee & Coughlin, 2001

- Cryptorchid men who had undergone orchiopexy between 1955 - 1975 at the Children's Hospital of Pittsburgh
- Control group of men who were age-matched who underwent unrelated surgery at the same institution
- Review of medical records and by completion of a questionnaire

<table>
<thead>
<tr>
<th>Group</th>
<th>Total (n)</th>
<th>Attempted Paternity (%)</th>
<th>Successful (%)</th>
<th>Unsuccessful (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bilateral</td>
<td>88</td>
<td>49 (55.7)</td>
<td>32 (65.3)*</td>
<td>17 (34.7)</td>
</tr>
<tr>
<td>Unilateral</td>
<td>609</td>
<td>359 (59.0)</td>
<td>322 (89.7)</td>
<td>37 (10.3)</td>
</tr>
<tr>
<td>Control</td>
<td>708</td>
<td>443 (62.6)</td>
<td>413 (93.2)</td>
<td>30 (6.8)</td>
</tr>
<tr>
<td></td>
<td>1405</td>
<td>851 (60.6)</td>
<td>767</td>
<td>84</td>
</tr>
</tbody>
</table>

* P <0.001 vs. unilateral and control groups; chi-square test.

Paternity rates following orchiopexy
- Bilateral UDT – 65%
- Unilateral UDT – 90%
- Control: 93%
Physical Examination

- Ideally, the patient should be relaxed
- Observe before examining
  - Features of the scrotum and its content
- Is the contralateral testicle hypertrophic
- Frog leg position can be useful
- Palpate from iliac crest down to scrotum
  - Lubrication or soap may help
- Milk testicle down, fatigue cremasteric fibers
- If findings are equivocal, consider repeat exam
Retractile Testicle

Courtesy of Manish Bhalla
Testicular Ascent

- Prior documentation of testes in the dependent scrotum should not preclude continued GU exams
- A testis can ascend
- Acquired cryptorchid testes are at risk for developing the same adverse histologic changes
When to Refer?

• Providers should refer infants with a history of cryptorchidism who do not have spontaneous descent by six months (corrected for gestational age)
Imaging for an Undescended Testicle

- Imaging rarely assists in decision-making
  - Ultrasound
    - Sensitivity 45%, specificity 78% (Tasian and Copp, 2011)
  - MRI
    - 60-80% accuracy for non-palpable testes
    - False negative rates: 8-14%
    - False positive rate: up to 20%
      - (Kanemoto et al. 2005, Kantarci et al. 2010, Desireddi et al. 2008)
- There is no radiologic test that can conclude that a testicle is absent
- Surgical exploration/diagnostic laparoscopy is the gold standard
UDT and Ambiguous Genitalia

• Kaefer et al. 1999
  • 79 patients between 1982-1996 presenting with UDT and hypospadias
  • DSD diagnosis in 30% of unilateral and 32% in bilateral UDT patients
  • The DSD diagnosis was more common in patients with non-palpable testes
  • Hypospadias severity (posterior, mid, anterior) has a strong positive correlation with a disorder of sex differentiation
Ambiguous Genitalia
3 month old FTT
Pelvic Ultrasound
Genitogram
Two out of Three Rule
3 yo girl with blood spotting on underwear...

• A healthy 3 year-old girl presented to the office with complaints of blood spotting in underwear x 2 days

• No significant past medical history
Physical Exam

• Healthy appearing
• RRR
• CTAB/L
• Abdomen soft, ND, no masses
• GU exam...
What is the diagnosis?
Physical Exam

- Frog leg position or knees to chest
- Light
- Labial separation and gentle outward traction
  - Clitoris
  - Urethral meatus
  - Hymen
  - Introitus
Urethral Prolapse

• Circumferential eversion of the urethral mucosa
• Prepubertal black girls and postmenopausal white women
• Exact pathophysiological process unknown
  • Hypoestrogenism
  • Abnormal connections between the inner longitudinal and outer circular muscle layers of the distal urethra
  • Episodic increases in intra-abdominal pressure
• How would you treat this?
  • Observation, steroid cream, sitz baths, surgical excision

• Is this likely to recur?
  • Recurrence rate is up to 67%
Treatment

• Observation

• Topical steroid cream & sitz baths

• Surgical excision
  • Circumferential excision of the redundant mucosa and suturing of normal urethra to the vestibule
Question #1

Risk factors for having an undescended testicle include:

a) Low birth weight
b) Prematurity
c) Family history
d) All of the above
Question #2

Post circumcision skin bridges can be treated with betamethasone?

a) True  
b) False
Thank you!