

Male Reproductive System Disorders

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GENERAL APPROACH

- Male reproductive assessment includes genitalia, inguinal region, anus, and rectum
- Genital assessment done annually during physical exam and as needed
- Assess for axillary hair and gynecomastia
- Male Tanner staging (Sexual Maturity Rating) should be noted
- First sign of pubertal change in males is adrenarche (testicular enlargement)
- History should include attention to complaints of dysuria, polyuria, abdominal pain, enuresis (nocturnal, daytime, primary, secondary); testicular or scrotal pain; history of sexual activity, partner preference, types of sex engaged in, number of partners, condom use; complaints of urethral discharge; alterations in erection patterns; history of urinary tract infections (UTIs) and sexually transmitted infections (STIs); and any surgical procedures to the genitourinary system
- Family history for urology disorders and pubertal development should be noted
- Teach and encourage testicular self-examination (TSE)

PHYSICAL EXAMINATION

- Male Sexual Maturity Rating (SMR) or Tanner staging described in terms of pubic hair and genital development (can occur at different rates)

- Male genital stages
 - *Genital Stage 1 (G1)*: prepubertal, phallus child-like
 - *Genital Stage 2 (G2)*: no change in phallus; scrotum reddened, thinner, and longer; testes enlarge
 - *Genital Stage 3 (G3)*: greater enlargement in scrotum, phallus increased length
 - *Genital Stage 4 (G4)*: further enlargement and darkening of scrotum, phallus increased length and circumference
 - *Genital Stage 5 (G5)*: adult phallus and scrotum in size and contour (Finberg & Kleinman, 2002)
- Male pubic hair stages
 - *Pubic Hair Stage 1 (PH1)*: none
 - *Pubic Hair Stage 2 (PH2)*: sparse amount of long, downy hair along penis base
 - *Pubic Hair Stage 3 (PH3)*: moderate amount of curly, coarser hair, extending more laterally
 - *Pubic Hair Stage 4 (PH4)*: hair that resembles adult hair, no extension on medial surface of thighs
 - *Pubic Hair Stage 5 (PH5)*: adult type and quantity, extends to medial surface of thighs

DISORDERS OF THE NEWBORN

HYPOSPADIAS

Description/Epidemiology

- Congenital deformity of penis resulting in the urethral meatus located anywhere along the ventral surface of the penis from the proximal glans to the perineum
- Chordee or a downward curvature of penis may be noted (~35% of cases)
- Approximately 1 in 250 males
- More common in White males and those conceived with fertility medications or in vitro fertilization
- Usually an isolated anomaly but common when there are multiple anomalies; most commonly associated condition is undescended testicles

Etiology

- Unknown

Clinical Manifestations

History

- Uncorrected older child may have spraying or deflection of the urinary stream
- Sits to void
- Needs to direct stream

Physical Exam

- Meatal opening not located at tip of penis but anywhere along ventral surface from glans to perineum
- Ventral foreskin absent; dorsal foreskin appears “hooded”
- Chordee or ventral curvature of penis often present
- Associated findings may include inguinal hernia, undescended testicle(s), or hydrocele.

Risk Factors

- Family history: present in about 8% of fathers, 14% of siblings of the affected child, 21% if two family members
- Twin

Differential Diagnosis

- Intersex abnormalities

Diagnostic Testing

- Renal and bladder ultrasound may be obtained to rule out a concurrent upper-tract anomaly (very uncommon)
- Chromosome studies if question of gender, especially if associated with undescended testes

Management

- Avoid circumcision so the tissue may be used for surgical correction
- Refer to pediatric urology at birth
- Surgical correction by a pediatric urologist
- Referral to pediatric urologist shortly after birth for evaluation
- Hypospadias and concurrent bilateral cryptorchidism warrant evaluation for possible intersex anomaly

Prevention and Screening

- Careful examination of the newborn

CRYPTORCHIDISM**Description/Epidemiology**

- The failure of one or both testes to descend through the inguinal canal and remain in the scrotum. This typically occurs between 7 and 9 months' gestation.
- Most common locations are
 - Prescrotal (below the external inguinal ring but not in the scrotum)
 - Inguinal or canalicular (between the internal and external inguinal ring)
 - Abdominal and impalpable
- Most common genitourinary disorder in boys
- 60%–70% experience spontaneous descent by 3 months
- Approximately 10% are bilateral
- In 3%–5% of infants with impalpable testis, testis will be absent

Etiology

- Testosterone deficiency
- Abnormal or absent testis
- Short spermatic cord
- Narrow inguinal canal
- Adhesions

Clinical Manifestations**History**

- Parent may or may not have seen testicles, often notice “empty” appearance of scrotum during diaper changes or fullness during bath
- Prematurity
- Family history of cryptorchidism

Physical Exam

- Warm hands (to decrease cremasteric reflex) and position the patient if necessary (tailor position or sitting cross-legged, standing, or kneeling) to facilitate examination
- Examine scrotum
 - Unilateral or bilateral
 - Palpable or nonpalpable: may be located in the inguinal canal, intra-abdominal, or absent
 - Determine if retractile or true undescended testicle; retractile is normal variant usually due to a strong cremasteric reflex and may be “milked down” or palpable with positioning
 - Testicular size may be hypertrophied in some cases
 - Assess for hernias and hydroceles (about 75% of boys also have a hernia)
- Examine for hypospadias

Risk Factors

- Prematurity
- Intersex abnormalities
- Low birth weight
- Twin
- (+) Family history
- Firstborn
- Cesarean section
- Toxemia
- Congenital subluxation of the hip
- Endocrine or chromosomal disorder
- Winter conception
- Down syndrome
- Maternal age < 20 or > 35 years
- Other urinary disorders

Differential Diagnosis

- Retractable testis

Diagnostic Testing

- Usually obtained by specialist
- Ultrasound, CT scan, and MRI may be used to locate testis
- Laparoscopic exam may be done to locate abdominal testis
- Chromosome analysis if bilateral or associated with hypospadias

Management**Nonpharmacological Treatment**

- Surgical correction (orchiopexy or orchiectomy) by a pediatric urologist at about 9–12 months of age

Follow-up

- Routine well-child visits important to document presence of testes

Complications

- Malignancy more common in undescended testis; 10% of all testicular cancers occur in patients with an undescended testis
- Cryptorchid testis more prone to torsion
- Risk of infertility about 25%–50% if unilateral and 50%–75% if bilateral
- Fertility about 75%–90% after orchiopexy when performed before age 2 years

Prevention and Screening

- Careful examination during newborn exam and well-child visits; important to document presence and position of testicle(s)

PENILE DISORDERS**BALANITIS****Description/Epidemiology**

- Inflammation of the glans
- Balanoposthitis: inflammation of foreskin and glans penis occurring in phimosis or uncircumcised males
- Approximately 6%

Etiology

- Accumulation of debris under foreskin from poor hygiene that results in irritation/infection of the glans
- Skin flora are usual cause but also may be gram-negative organisms
- If urethral discharge, consider STI or candidiasis
- Allergy or trauma (rare)

Clinical Manifestations**History**

- Crying/fussiness
- Older children typically complain of dysuria and pain in genital area
- Complaints of swelling and exudate on penile shaft
- Fever
- Circumcision history
- History of repeated UTIs
- History of balanitis

Physical Exam

- Exudate
- Erythema and swelling of penis
- Note scar tissue if history of repeated episodes of balanitis
- Debris accumulation under foreskin may be noted

Risk Factors

- Poor hygiene
- Uncircumcised males
- Lack of condom use

Differential Diagnosis

- Cellulitis
- Herpes simplex virus (HSV)
- Dermatitis
- Eczema

Diagnostic Testing

- Culture

Management**Nonpharmacological Treatment**

- Warm baths and soaks
- Area left open to air if feasible

Pharmacological Treatment

- Appropriate oral antibiotic therapy based on history and culture
- Topical:azole for candidiasis
- Hydrocortisone cream topically for pruritus
- Resolution of symptoms expected with topical and systemic antibiotic therapy
- Consultation with dermatology or urology if no improvement with oral antibiotic therapy
- Hospitalization is warranted in the neonatal period for IV antibiotic therapy
- Repeated occurrence may warrant a referral for circumcision consult in the uncircumcised male

Complications

- Paraphimosis
- Scar tissue if repeated episodes of balanitis

Prevention and Screening

- Education regarding correct hygiene
- Education to promote use of barrier method of contraception to avoid STI

PHIMOSIS**Description/Epidemiology**

- Foreskin cannot be retracted over the glans penis; either congenital or acquired
- Primary phimosis: physiological until age 6 years
- Secondary phimosis: results when a previously retractible foreskin can no longer be retracted or after puberty
- Actual phimosis (scar tissue) 2%–10% of uncircumcised males

Etiology

- Physiological (normal variant): foreskin adherent to glans in newborn, will naturally separate usually by age 6 years
- Pathological phimosis occurs when the foreskin becomes scarred and does not resolve with time. Theories for the development of scarring include
 - Forcible retraction of the foreskin
 - Irritation, such as from soiled diapers
 - Infection (balanitis), usually from poor hygiene
 - Congenital anomaly, congenital narrowing or tightening of the prepuce

Clinical Manifestations**History**

- Inability to retract foreskin when previously retractable
- Scarred, unretractable foreskin may result in symptoms such as dysuria, hematuria, and poor urinary stream; severe scarring may cause ballooning of the foreskin on urination
- If stenosis worsens, rarely may develop hydronephrosis and renal failure
- May be asymptomatic, but that is more consistent with physiological tightness of the foreskin

Physical Exam

- Foreskin: tight, not able to be retracted to visualize the meatus or, if an older boy, not able to retract over the glans
- Tip of the foreskin may be whitish, scarred, and stenotic (may not be able to visualize the meatus)

Risk Factors

- Forcible retraction of the foreskin
- Poor hygiene practices producing infection and subsequent scarring

Differential Diagnosis

- Balanitis
- Trauma

Management**Nonpharmacological Treatment**

- Teaching parents to gently retract foreskin only as far as it will easily move for cleaning purposes
- Good hygiene in uncircumcised male
- Teach child proper care when age-appropriate

Pharmacological Treatment

- Topical steroids have been used to treat adhesions
- Use of topical antibiotic ointments such as Neosporin or Bacitracin applied to the tip of the foreskin can act as a protective barrier to fecal contamination
- Pathological phimosis should be referred to a pediatric urologist for possible lysis of adhesions or circumcision

Follow-up

- Examination during well-child exams

Prevention and Screening

- Teach parents proper care for the uncircumcised penis
- Emphasize that foreskin retractions should never be forceful
- Examine the foreskin on routine well-child exams

TESTICULAR DISORDERS

VARICOCELE

Description/Epidemiology

- Dilated elongated (varicose) veins of the pampiniform plexus above the testis; can involve the internal spermatic vein
- Likely the result of increased pressure on incompetent venous valves in the internal spermatic vein
- Approximately 20% of males
- > 90% are left-sided
- Can be bilateral

Etiology

- Unknown but possibly due to incompetence of the valves of spermatic vein
- Most are left-sided, rarely involves the right.
- Right-sided varicocele, must rule out vena cava obstruction or obstruction from a tumor or intra-abdominal pathology

Clinical Manifestations**History**

- Painless mass outside testicle; occasionally described as a dull scrotal ache that worsens with activity; may radiate to groin and medial thigh
- Patient may note that size decreases with lying down, especially after standing for long periods
- May progressively worsen

Physical Exam

- Examine standing; then supine. Most palpable only in the standing position because gravity allows retrograde filling of the spermatic venous complex. Lying down usually decompresses the varicocele and relieves symptoms.
- Scrotum feels like a bag of soft worms above and lateral to the testes
- Examine testis, epididymis, and vas deferens and compare size of contralateral testis
- Testis on affected side may be smaller, evaluate with orchidometer
- If right side only and does not change with position, consider abdominal pathology

Differential Diagnosis

- Hydrocele
- Spermatocele
- Epididymal cyst
- Tumor

Diagnostic Testing

- Ultrasound to compare testicular volume

Management**Nonpharmacologic Treatment**

- Monitor testicular growth and size, watching for testicular atrophy
- Follow adolescent to monitor testicular growth; see every 6–12 months until end of puberty
- Painful or large varicoceles, right-sided or bilateral, and varicoceles in prepubertal males require immediate referral
- Testicular atrophy on the affected side
- Any question regarding the need for surgery
- Return if painful or change in the size of the scrotum or testes

Complications

- Decreased testicular size on affected side
- Infertility
- Catch-up growth seen after surgical repair (80% of cases), as well as improved sperm count

Prevention and Screening

- Should examine patient in standing position (may disappear when supine)
- Annual testicular examination

TESTICULAR TORSION**Description/Epidemiology**

- Twisting or torsion of spermatic cord, compromising blood supply to testis; a surgical emergency
- Most frequently seen in neonates, preadolescents, and adolescents, but may occur at any age
- 1 in 4,000 males < 25 years of age

Etiology

- Fixation of the testis is abnormal or absent, allowing for the torsion
- Appendages of the testes or epididymis can also be affected
- Many have “bell-clapper” deformity—the tunica vaginalis completely surrounds the testis and epididymis, which hang independently and without fixation (intravaginal torsion)
- In neonates, the testis and the tunica are poorly fixed within the scrotum and the cord structures, testis, and tunica twist (extravaginal torsion); this can occur antenatally

Clinical Manifestations**History**

- Onset: usually acute but may be gradual; ~30% have had one prior episode of pain that resolved (intermittent torsion)
- Pain is severe and may radiate to abdomen, thigh, or flank

- Nausea/vomiting
- Recent trauma, sexual intercourse, or exercise, though torsion may occur while child is at rest
- Epididymis palpated in anatomically incorrect position

Physical Exam

- Acute distress, patient looks ill and very uncomfortable
- Scrotum edematous, warm, erythematous, and skin taut
- Cremasteric reflex absent on affected side
- Testis high-riding in scrotum, enlarged, very tender, can be in transverse position
- Spermatic cord thickened and tender
- Contralateral testis in normal position and nontender
- Usually no voiding problems

Risk Factors

- May occur spontaneously, but is also associated with trauma, even minor trauma, or exercise (~20%)
- Age: uncommon < 10 years of age (except neonatal period)

Differential Diagnosis

- Epididymitis or orchitis
- Incarcerated hernia
- Trauma
- Tumor

Diagnostic Testing

- Doppler ultrasound of scrotum helpful in distinguishing between torsion and epididymitis, but is used when torsion less likely due to time delay
- Urinalysis: not diagnostic; helps with differentials such as pyuria with epididymitis or hematuria with trauma

Management

- All patients with an acute scrotum should be referred immediately to a pediatric urologist
- Surgical exploration, derotation, and orchidopexy (if testis viable, fixed to the tunica vaginalis) and orchidopexy of the contralateral testis performed (increased risk of torsion of contralateral testis)
- If nonviable, orchiectomy is performed
- Salvage rate is usually 100% if treated within 4–6 hours of onset of symptoms; can be < 20% if 12-hour delay and after 24 hours, testis usually necrotic

Follow-up

- Determined by surgeon

Complications

- Loss of affected testis

Prevention and Screening

- Education
- Identification of bell-clapper deformity on physical exam

EPIDIDYMITIS

Description/Epidemiology

- Inflammation of epididymis
- Uncommon in prepubertal males or non–sexually active adolescent males without a history of genitourinary abnormalities

Etiology

- Most common organism is *Chlamydia trachomatis*. Other common organism is *Neisseria gonorrhoeae*
- Adenovirus, mumps, and mononucleosis have all been associated with viral epididymitis
- Other organisms may present dependent upon structural abnormalities of the genitourinary tract anal sexual intercourse practices (*E. coli*) or instrumentation

Clinical Manifestations

History

- Subacute onset of unilateral swelling and pain (acute painful onset with torsion; swelling of epididymis alone more common in epididymitis than torsion)
- Dysuria
- Urethral discharge
- Pyuria
- Fever
- Past medical history of genitourinary surgery or UTIs or dysfunction
- Sexual history
- History noting sports participation and use of scrotal protection or support

Physical Exam

- Epididymal edema and pain
- Urethral discharge
- Fever possible
- Reactive hydrocele present
- (+) Prehn sign: elevation of scrotum relieves pain
- (+) Cremasteric reflex on affected side
- No evidence of trauma

Risk Factors

- Unprotected sexual activity
- Surgery
- Instrumentation
- Congenital or acquired anatomic abnormalities

Differential Diagnosis

- Testicular torsion
- Orchitis
- Mumps orchitis
- Hydrocele
- Varicocele
- Tumor
- Abscess
- Testicular infarction

Diagnostic Testing

- Urinalysis/urine culture as necessary
- Gram stain of urethral swab specimen, noting > 5 polymorphonuclear leukocytes per oil immersion to diagnosis for *N. gonorrhoeae*
- If Gram stain negative, urine culture
- Cultures for chlamydia and gonorrhea
- Serology for syphilis
- Encourage HIV counseling and testing
- Doppler sonogram or scan as warranted (usually would entail consult with urology)

Management**Nonpharmacological Treatment**

- Scrotal support
- Bedrest in acute phase
- Sexual activity and intercourse should be ceased until inflammation resolved and antibiotic therapy completed (10 days to prevent transmission to sexual partner)
- Partner notification (dependent upon organism)
- In absence of urethral discharge, leukocytes on gram stain, or pyuria, urgent urology consult warranted
- Immediate referral to urology if testicular torsion or testicular infarction suspected
- Lack of resolution of symptoms requires urology referral
- Hospitalization warranted for testicular torsion, testicular infarction, or abscess
- Preservation of testicular function requires early surgical intervention

Pharmacological Treatment

- Recommended antibiotic regimen (likely gonorrhea and/or chlamydia)
 - Ceftriaxone 250 mg IM single dose PLUS doxycycline 100 mg orally b.i.d. × 10 days
- Recommended antibiotic regimen (likely enteric organisms or allergic to cephalosporins or tetracycline)
 - Ofloxacin 300 mg orally b.i.d. × 10 days OR levofloxacin 500 mg orally q.d. × 10 days
- Analgesics for fever and pain at appropriate dosage
- Resolution anticipated with compliance with antibiotic therapy
- If no improvement within 3 days of antibiotic therapy, reevaluate or refer to urology

Complications

- Chronic epididymitis
- Infertility if not treated

Prevention and Screening

- Education regarding use of condoms
- Partner identification and treatment as a means of infection control and prevention of complications

TESTICULAR CANCER**Description/Epidemiology**

- Testicular or paratesticular mass: hard, painless, variable size
- Incidence is 2.3–10/100,000 males

Etiology

- Most germ cell in origin and malignant
- Nonseminomas (embryonal cell, choriocarcinoma, teratoma, yolk sac, and mixed origin) most common in 15- to 30-year-olds

Clinical Manifestations**History**

- Circumscribed, painless area of induration, does not transilluminate
- Fullness or heaviness in scrotum
- Edema (73%)
- Pain (hemorrhage or necrosis)
- Systemic symptoms from dissemination: supraclavicular mass, hemoptysis, bone pain, abdominal mass

Physical Exam

- Testis enlarged or lump or irregularity felt on testis
- Check for hydrocele; onset of hydrocele in adolescence may indicate an underlying tumor
- Gynecomastia may occur and be the presenting complaint
- Premature growth of body hair
- Precocious puberty may occur, especially with Leydig tumors
- Abdominal exam for any masses or organomegaly
- Lymphadenopathy may occur
- Chest: breath sounds normal or decreased (may metastasize to lungs)

Risk Factors

- Family history of testicular cancer
- Cryptorchidism
- Mothers exposed to DES (diethylstilbestrol), estrogen, or estrogen-progestin in first 2 months of gestation

Differential Diagnosis

- Hydrocele
- Spermatocele
- Torsion of testicular appendage

Diagnostic Testing

- Testicular/scrotal ultrasound
- Tumor markers: elevations of AFP (alpha-fetoprotein), less consistently beta human chorionic gonadotropin (beta HCG) and LDH (lactic acid dehydrogenase) are supportive of diagnosis

Management**Nonpharmacological Treatment**

- All testicular tumors or masses referred to a pediatric urologist and oncologist immediately
- Surgical management by pediatric urologist

Pharmacological Treatment

- Managed by an oncologist

Follow-up

- Determined by specialists

Complications

- Dependent on type of cancer, clinical course, and therapy

Prevention and Screening

- Teach teens to do monthly testicular self-examinations and to call if abnormal findings
- Evaluate annually at physical examination

CASE STUDIES

Case 1: You are seeing a 17-year-old high school senior for his sports physical. He presents with no complaints, and a review of systems is benign. He is Tanner 5. You are performing his testicular exam when you note that his scrotum is enlarged. When you question him, he states that he “thought it was getting bigger in the last few days” but thought it might be due to an injury that occurred during football practice last week. He reports he was tackled and “took a helmet” in his groin. He was “sore” for a few minutes but was able to return to play within 10 minutes and finished practice. He denies any residual pain at this time.

1. What is your differential diagnosis?
2. What additional history should be obtained?
3. What other findings on exam must be noted?
4. What diagnostic tests will you order?

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