Male Genitourinary Development and Assessment

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Boys are beyond the range of anybody’s sure understanding, at least when they are between the ages of 18 months and 90 years.

—James Thurber

Learning Objectives:
1. Describe the sequence of male puberty and clinical methods to assess genital development
2. Diagnose and manage common male genitourinary conditions and concerns
3. Generate a differential diagnosis and management plan for acute scrotal pain and testicular masses

Primary References:

Author’s Note: The primary reference listed above uses gender binary language. It is important to address gender identity with your patient and use language that aligns with their preferred identity. Where indicated in this chapter, the words male and female indicate assigned sex at birth.

CASE ONE:

Pugh Berty is a 14-year-old boy that presents to your office for well child examination. You go over the agenda for the visit including explaining that a genitourinary examination should be performed. Pugh then asks, “Do you really need to an exam? They just did that last year!” His father nods in agreement.

1. What do you tell Pugh and his father about the genitourinary exam?

The external examination of male genitalia is part of routine examination for all adolescent (and younger age) males and is recommended on an annual basis in a 2012 position paper by the Society for Adolescent Health and Medicine and in the 4th edition of Bright Futures from the AAP. Clinicians should consider three valuable aspects of routine genital examination. First, the exam is intended to find abnormalities, which can arise at any moment and may not be reported by the patient due to discomfort with reporting about “private parts”. Second, the exam allows the clinician to assess for pubertal development and foster additional conversation around this topic. Finally, the fact that the exam is being performed serves to normalize genital health for the child (independent of gender) as they grow. The child may be more likely to report abnormalities and feel less worried about an examination by a healthcare provider in the future.

The upright (standing) position should be used since a condition such as varicocele may be undetected in the supine position. Prior to beginning the examination, it is important to complete a comprehensive HEADSSS/ SSHADES assessment with the patient alone to discuss any physical and emotional changes during puberty, body image, gender and sexual identity, and mood including suicidality and self-harm.

You can let the family and patient know that you will be assessing the penis, foreskin (if present), scrotum, testicles, genital hair, and skin. You will be specifically looking for abnormalities in penile, testicular, and scrotal size and structure; assessing the urethral meatus; and evaluating for any lesions that can be seen with sexually transmitted infections. In addition, you will be able provide reassurance about what is normal during sexual development, such as progression of pubic hair and enlargement of the penis and testes.
Of note, the AAP recommends the use of a chaperone during genital, anorectal, or breast examination. The chaperone can ensure the professionalism of the examination and serve as a witness.

2. What is the Sexual Maturity (Tanner) Rating for male genitalia?

The usual sequencing of puberty in males begins with testicular enlargement, followed by pubic hair growth, and then an increase in height. When examining the male external genitalia, the provider must assess the testes for size, descent, and other abnormalities. Prepubertal testis are <2 cm and <4 ml at stage 1, whereas an adult testis are >5 cm and >15 ml at stage 5. The moderator can refer learners to table 3 in the article by Cavanaugh. A Prader orchidometer can be used to assist with testicular size assessment. At Stage 1, there will be no pubic hair present, but by stage 5, hair will spread to the medial thigh. Consider height, weight, and BMI along with the associated percentiles when assessing Tanner Staging to determine whether there is a deviation in the growth curve that might indicate that puberty is not progressing as expected. If there is any abnormality, further work up may be required. The moderator is referred to the chapter on Normal and Precocious Puberty for more detail on genitourinary development in males.

3. In addition, to assessing pubertal status, what abnormalities in genital development may be evident during the examination?

Hypospadias affects 1/300 live male births per the American Urological Association. During the eighth week of gestation, the genital tubercle is stimulated by testosterone to form the penis. The penile urethra develops when the urethral folds form a seam which fuses in the midline, and hypospadias occurs following deviations in this process. Usually detected in the newborn period, hypospadias may also be accompanied by an abnormal appearing foreskin and ventral curvature of the penis. Surgery is needed and is usually performed at 6 months of age. It is also important to note that circumcision should be avoided at birth for these infants.

An additional problem detectable on examination is micropenis, which is defined as a stretched penile length less than 2.5 standard deviations below the mean for age (see Table 1 in the article by Wu and Gitlin). A normal size penis can be confused as being micropenis in the cases of buried penis or penoscrotal fusion. Buried penis is acquired secondary to a large suprapubic fat pad or penile adhesions. Penoscrotal fusion causes a webbed penis, attached down to the scrotum. Work up for true micropenis includes karyotype, hormone levels (LH, FSH, testosterone, and anti-Mullerian), and referral to endocrinology for consideration of hormone replacement.

Phimosis is a condition in which the uncircumcised foreskin cannot be retracted to expose the glans penis. Although resolution commonly occurs by ages 5-7, persistence through seventh grade was seen in 7% of Taiwanese boys in one study. It is important to let the patient and family know to not forcefully retract the foreskin of any child as this can cause trauma. Complications of phimosis can include recurrent balanitis, balanoposthitis, balanitis xerotica obliterans (lichen sclerosis), and scarring. Thus, phimosis in older children can be an indication for treatment with a topical corticosteroid, usually with betamethasone 0.05% or triamcinolone 0.1% cream applied directly to preputial skin for 6-8 weeks.

In assessing size of the testes, the provider should assess for testicular asymmetry. Testicular asymmetry could indicate varicocele, tumor, torsion, infection, hydrocele, or cryptorchidism. Next steps will be determined by exam findings as discussed later in this chapter. If bilateral testicular hypoplasia is suspected; the provider should consider genetic abnormalities such as Klinefelter Syndrome as a cause. However, if there is concern for bilateral macro-orchidism, the provider should consider Fragile X-syndrome in the differential.

Cryptorchidism is a condition occurring in about 3% of full-term infants, in which a testicle is not found in the scrotum and is usually palpated in the inguinal canal. Congenital cryptorchidism should be distinguished from retractile testis, in which the testis can be brought into the scrotum without tension and temporarily remain in the scrotum. This should be monitored annually by the primary care clinician and does not have an increased risk for cancer as the testicles can be manipulated into the scrotum. For true cryptorchidism, patients should be referred to urology at 6 months of age if spontaneous descent has not occurred. In later years, it is important to recognize that a history of cryptorchidism increases risk of testicular cancer.

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Varicocele is an abnormal dilation of the pampiniform plexus in the spermatic cord, usually occurring in the left hemiscrotum as the spermatic vein drains into the left renal vein at a right angle while on the right, the spermatic veins drains directly into the inferior vena cava. Varicoceles can be bilateral but, if there is an isolated varicocele on the right, this raises concern for a tumor that is compressing the IVC. The prevalence increases during adolescence with approximately 8% of 11-14 year old males and 14% of 15-19 year old males having a varicocele. Diagnosis is typically made via examination with the patient standing and when doing a Valsalva maneuver, where the dilated spermatic veins will be palpable and have the appearance and the feel of a “bag of worms.” When the patient lays supine, the varicocele should resolve. If the varicocele is still present in the supine position or if there is an isolated right-sided varicocele, another etiology, such as an underlying mass should be ruled out with imaging studies.

Varicoceles can impact fertility; in adults, 40% percent of men with primary infertility were found to have a varicocele as reported by Macey, et al. While up to 80-85% of men with varicoceles are not infertile, treatment for infertility is ultimately needed in one-fifth to one-third of individuals with varicoceles. Surgical intervention is indicated if there is pain or inadequate testicular growth, or in young adults when abnormal semen parameters are noted. If semen analysis is abnormal, it should be repeated every 1-2 years. The benefits of surgical intervention include increases in testicular volume and improvement in semen analysis.

Other conditions that can be detected in the genitourinary examination include masses such as hydrocele, spermatocele, hernia, and tumors.

Hydrocele presents as a scrotal mass that transilluminates with a light source, and is caused by peritoneal fluid collecting between the visceral and parietal layers of the tunica vaginalis, surrounding the testis and spermatic cord. It is commonly caused by a lack of closure of the processus vaginalis after testicular descent (communicating, or open hydrocele), but frequently the processus vaginalis will close with additional fluid present (non-communicating, or closed hydrocele). In newborns, they can be differentiated by noting that fluid in the hydrocele will wax and wane over hours to days if communicating, but will not do so if non-communicating. Non-communicating hydroceles in newborns typically resolve spontaneously by age 2 and can be followed clinically in the absence of other abnormalities. Outside the newborn period, hydroceles can be due to inflammation from epididymitis, masses, or torsion so obtaining a scrotal ultrasound should be considered to determine the cause. An open hydrocele to the peritoneal cavity requires surgical intervention to prevent incarcerated hernia thus all patients with communicating hydroceles or hernias should be referred. A closed hydrocele does not need repair if asymptomatic. Surgical intervention should be considered depending on the etiology of the hydrocele, presence of pain, and integrity of the scrotal skin.

A spermatocele is an epidydimal cyst that is larger than 2 cm. These generally do not require any intervention unless the patient has chronic or recurrent pain.

Testicular cancer is one of the most common cancers amongst adolescents and young adults, with germ cell tumor being the most common. Other types of testicular cancers include sex cord stromal tumors, mixed germ cell and sex cord stromal tumors, and paratesticular cancer including testicular lymphoma and leukemia. However, testicular cancer is uncommon overall and the USPSTF recommends against routine clinician screening or self-examination amongst asymptomatic young men who do not have a history of cryptorchidism. This recommendation does not mean that testicular exam should not be performed for other reasons (e.g., pubertal staging) or when it is clinically indicated based on symptoms. If your patient presents with a concern of a mass, prolonged testicular pain, or asymmetry, cancer should be on the differential. Work up, in conjunction with or by an oncologist, should include an ultrasound as well as alpha-fetoprotein and human chorionic gonadotropin levels if the patient is older than 6 months.

CASE continued:

A few weeks later, you note a message in your inbox from Pugh that he went to the emergency department due to the acute onset of genital pain. The message does not clarify where Pugh is feeling the pain.
He reports feeling much better now but wants to schedule an appointment for follow up.

What is the differential diagnosis of acute penile pain in males?

The differential for acute genitalic pain in males is broad, however it can be helpful to separate symptoms anatomically.

First, considering penile pain, the differential includes penile trauma and as previously mentioned, phimosis and paraphimosis. Penile trauma is overall quite rare in the pediatric population, with the most common injury occurring via toilet seat related accidents. Any penile laceration requires surgical repair. It also very important to consider sexual abuse as the cause of penile trauma, making a confidential discussion with the patient as to the cause of injury key. Paraphimosis is a condition in which the retracted foreskin cannot be returned to its normal position. Patients present with acute severe pain and diagnosis is based upon on examination. Management consists of reduction of the foreskin over the glans penis and pain control (both prior to the reduction and thereafter). Urology should be consulted if there is urinary obstruction or necrosis, or if manual reduction cannot be completed.

When symptomatic, urethritis can also present as penile pain with signs of urethral inflammation including dysuria, pruritis, and penile discharge. *Chlamydia trachomatis* and *Neisseria gonorrhoeae* are well known causes for urethritis. *Trichomonas vaginalis* and *Mycoplasma genitalium* are also common causative agents with the latter being the most common cause of persistent or recurrent nongonococcal urethritis (NGU). Uncommonly urethritis can be caused by coliform bacteria, which may be associated with insertive anal intercourse. The diagnosis of urethritis is made by finding one of the following: 1) Gram stain with intracellular gram-negative diplococci and/or >2-5 WBCs/ HPF in high to lower prevalence setting respectively, 2) mucoid or purulent discharge, 3) positive leukocyte esterase on first-void urine. The presence of intracellular gram negative diplococci on a urethral smear is diagnostic for gonorrhea infection in males. A first void urine sample (the first 30-60 ml of urine after the patient has not voided for at least one hour) should be sent for a nucleic acid amplification test (NAAT) to identify a causative organism.

The recommended regimen for treatment of NGU is doxycycline 100 mg PO BID for 7 days. Of note, the CDC indicates that doxycycline does have some activity against *M. genitalium*, but is commonly insufficient to cure the infection. Antimicrobial resistance testing for *M. genitalium*, which could indicate additional treatment options, is not presently available in the United States. In cases of persistent and recurrent urethritis, testing for *T. vaginalis* should be sent followed by presumptive treatment with single dose metronidazole in men who have sex with women. If *M. genitalium* is confirmed on NAAT testing, one week of moxifloxacin should follow the one-week course of doxycycline to eradicate this organism.

4. What is the differential diagnosis of acute scrotal pain in males?

When assessing acute scrotal pain, one must consider testicular torsion. While the etiology of nontraumatic testicular torsion is unclear, the “bell-clapper” deformity is often considered a risk factor. In this condition, the testis lies horizontally, is unanchored posteriorly, and is suspended within the tunica vaginalis solely by the spermatic cord. It is important to consider testicular torsion on the differential diagnosis for lower quadrant abdominal pain, even in the absence of scrotal pain, given the embryologic descent of the testes. Examination of a patient with suspected torsion can show absent cremasteric reflex, swelling of the hemiscrotum, and lower abdominal tenderness. Diagnosis via ultrasound with doppler is key to viability of the affected testis, enabling prompt surgical repair which should happen within 6 hours to maximize success. However, the affected testicle may still atrophy even if detorsion occurs within this time frame.

Other etiologies of scrotal pain include orchitis and epididymitis, which are not always caused by bacterial infections. A common cause of non-infectious testicular pain in prepubertal boys is inflammation caused by a twisted appendix epididymis or appendix testis, which are remnants of the Mullerian and Wolffian components from fetal development and may present with swelling of the hemiscrotum. Physical examination can show the “blue dot” sign (specific but not sensitive) and preserved cremasteric reflex and Doppler ultrasonography can distinguish torsion of these embryologic
remnants from testicular torsion, a surgical condition. For torsed appendices, treatment includes supportive care including scrotal elevation, ice packs, and NSAIDs. Resolution with autoinfarction should occur in 5-7 days. Orchitis can be caused by viral infections secondary to mumps, parvovirus, echorvirus, rubella, coxsackie, and lymphocytic choriomeningitis or bacterial infections such as brucellosis or resulting from instrumentation or anatomical abnormalities.

For patients who are sexually active, *N. gonorrhoeae* and *C. trachomatis* are the most common causes of inflammation of the testis and epididymis. The clinical presentation includes unilateral testicular pain and swelling, tender epididymis, and possibly a hydrocele. Additionally, epididymitis is usually associated with urethritis. However, since this presents similarly to testicular torsion, an ultrasound should be completed in those with any suspicion of torsion. Treatment for epididymitis due to gonorrhea or chlamydia includes a one-time dose of 500 mg ceftriaxone PLUS doxycycline 100 mg twice a day for 10 days.

In males who engage in insertive anal sex, treatment is a one-time dose ceftriaxone 500 mg IM and levofloxacin 500 mg orally for 10 days instead of doxycycline to cover enteric organisms. If only enteric organisms are likely, use levofloxacin as monotherapy.

Testicular trauma can be divided into blunt or penetrating. When considering blunt trauma that may lead to testicular rupture, it is important to assess with ultrasound as examination can be limited due to swelling and tenderness. Emergent consultation of urology is pertinent to help manage blunt or penetrating trauma to the scrotum.

**Additional References:**

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Resources: