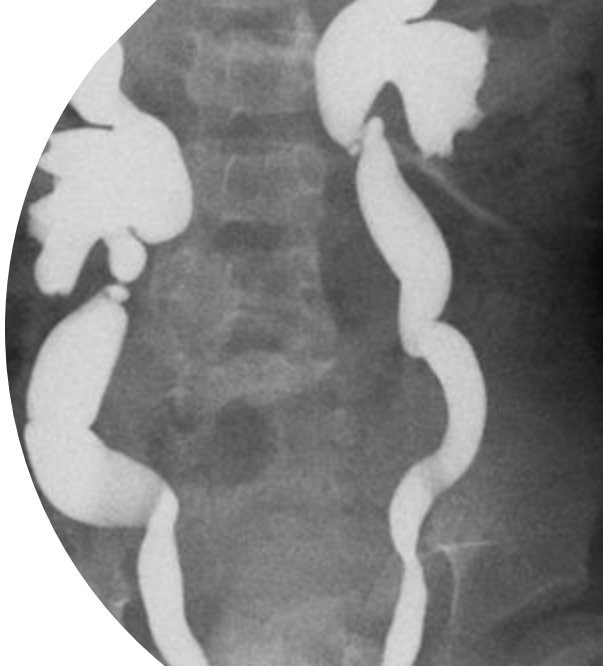
Principles in Pediatric Urology



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Topics in BLUE are the required ones

- Urinary Tract Infection
- Ureteropelvic Junction Obstruction
- Vesico-Ureteric Reflux
- Posterior Urethral Valves
- Circumcision
- Megaureter
- Neurogenic Bladder
- Hypospadias
- Disorders of Sex Development

Urinary Tract Infection

UTI

Urinary Tract Infection (UTI)

- The urinary tract is a common portal of infection in childhood.

- Diagnosis:
  - Suggestive symptoms
  - Urine microscopy
  - Urine quantitative culture of a properly collected sample

Urinary Tract Infection (UTI)

- F >> M (except in the first year of life when M>F).
- 8% of girls and 1–2% of all boys will get UTI during their childhood.

Urinary Tract Infection (UTI)

- Microbiology
  - E. coli (most common (80%)
  - Other pathogens include:
    - Gram –ve (as Citrobacter, Proteus, Pseudomonas, and Serratia)
    - Gram +ve (as Staphylococcus spp., Enterococcus spp., and Hemophilus spp.)

- Route of infection:
  - Ascending (via the urethra)

    or
  - Hematogenous (commoner in infants)

Risk Factors

- Antenatally diagnosed renal anomalies
- Family history of renal disease or VUR
- Bladder outflow obstruction (i.e., urethral valves)
- Neurogenic bladder or dysfunctional voiding
- Urolithiasis
- Constipation
- Uncircumcised males

Clinical Features

- Symptoms (upon age of the child):
  - Older children and adolescents: fever, dysuria, urgency, frequency, and urinary incontinence.
  - Infants: poor feeding, failure to thrive, temperature instability, jaundice, and vomiting.

- PEx:
  - Renal angle, abdominal or suprapubic tenderness
  - Abdominal mass, or a palpable bladder
  - Dribbling, poor stream, or straining to void
  - Hypertension (suggests hydronephrosis or renal parenchyma disease)
  - Dehydration and sepsis (in severe cases)

Urinary Tract Infection (UTI)

- In general, UTI must be suspected and urine must be tested:
  - any child with a fever above 38°C
    and
  - no other cause is evident

Investigations

- Urinalysis
  - Clean-catch sample (adhesive urobags (convenient in infants) are not ideal because of contamination. In the male, retract foreskin to expose the meatus)
  - Catheter specimen or a suprapubic sample (under ultrasonic guidance).

- Urine dipstick:
  - for leukocyte esterase or nitrite positivity (recommended mainly for >3-year-old).

- Urine microscopy:
  - Mainly for <3-year-old) for WBCs (>5 per high-power field), RBCs, bacteria, casts, and skin contamination.
  - Bacteriuria (preferably with pyuria) is considered positive.

- Urine culture:
  - Clean-catch sample:
    - Bacterial counts of $>10^5$ are diagnostic of UTI.
    - Bacterial counts 10^{3-5} → repeat test.
    - Bacterial count $<10^3$ → not significant.
  - Suprapubic sample: any organisms are diagnostic.
  - Multiple organisms:
    - Contamination with perineal flora OR Urinary tract abnormalities

Imaging

- Traditional approach
  - Birth to 1 year: US, DMSA scan, and VCUG
  - 1–5 years: US and DMSA only
  - Five years: US only

- In UK, the National Institute of Clinical Excellence (NICE) has suggested modification to:
  - Birth to 6 months: US only. (DMSA and VCUG if there is abnormality on US, or severe, atypical, or recurrent UTI).
  - 6 months to 3 years: US and DMSA (+/- VCUG if abnormalities on US, family history of VUR, or a poor urinary stream).
  - 3 years: US and DMSA (no need of VCUG in most cases)

Imaging

- Renal and bladder US:
  - for size and shape of kidneys
  - presence or absence of ureter
  - urolithiasis, hydronephrosis, hydroureter, and ureterocele
  - bladder emptying (in toilet-trained children)

- VCUG:
  - to diagnose posterior urethral valves or VUR
  - Bladder anatomy, bladder diverticulae or features of neurogenic bladder
  - [In girls, direct isotope cystogram (DIC) is preferred, as the radiation dose is much smaller]

- Radio-isotope renal studies:
  - DMSA scan:
    - investigation of choice
    - shows the kidney outline and detects renal scars
  - MAG-3 renogram:
    - preferred (to a DMSA scan) if there is hydronephrosis or if the ureters are dilated
    - can pick up VUR in the indirect cystogram phase in toilet-trained children

Management

- Parenteral antibiotics (e.g., ceftriaxone or cefotaxime):
  - All infants (<3 month) OR older patients who are sick
  - For ~10 days (if acute pyelonephritis is present), then the duration of therapy should be.

- Oral antibiotic (e.g., cephalexin, co-amoxiclav, trimethoprim, or nitrofurantoin):
  - started once the patient is afebrile OR lower UTI/cystitis in patients (>3 months) for 3 days only
  - provided patients are not sick AND good response within 48 h
  - up to 1/3 of community acquired UTIs are nowadays resistant to trimethoprim

- Routine antibiotic prophylaxis is not recommended (unless recurrent infections).

- Treat constipation (if present) [as constipation and dysfunctional voiding are more common causes of recurrent UTI than diseases like VUR]

- Asymptomatic bacteriuria:
  - does not require treatment
  - needs careful follow-up (if symptoms appear, urine tested again and treatment started)

Outcome

- Most UTIs are uncomplicated and respond to outpatient antibiotics.

- Complicated UTI needs prolonged follow-up to prevent long-term sequelae, like:
  - Renal parenchymal scarring
  - Hypertension
  - Decreased renal function
  - Renal failure

Ureteropelvic Junction Obstruction

(UPJO)

Ureteropelvic Junction Obstruction (UPJO)

- Hydronephrosis (HN) ~1 in 500 pregnancies (due to UPJ obstruction ~50%)
- M:F 2:1 | Lt:Rt 3:2 | bilateral 10–40%
- Most UPJ obstructions are now detected by antenatal US

Etiology

- **Intrinsic**
  - Intrinsic narrowing (patent but aperistaltic) (↓ muscle fibers, replacement by fibrotic tissue with disruption of spiral orientation).
  - Rarely: mucosal valves, polyps, and true ureteric strictures

- **Extrinsic**
  - Aberrant or supernumerary renal vessels (~30% of UPJ, an artery directly enters the lower pole of the kidney).
  - Kinking as a result from severe vesicoureteral reflux (VUR).

Clinical Features

- Most are asymptomatic (detected via prenatal screening US)

- Older children (and adults):
  - Episodic flank or abdominal pain (~50%)
  - Palpable flank mass (~50%)
  - Hematuria
  - Recurrent UTIs (~30%)

Investigations

- Antenatal US:
  - Renal pelvis AP diameter >4 mm at GA of <33 weeks
  - Renal pelvis AP diameter >7 mm at GA of >33 weeks

Antenatal US

- Society of Fetal Urology (SFU)
  - Grade 0 – normal kidney
  - Grade 1 – minimal pelvic dilation
  - Grade 2 – greater pelvic dilation without caliectasis
  - Grade 3 – caliectasis without cortical thinning
  - Grade 4 – HN with cortical thinning

Investigations

- Postnatal US (Primary investigation tool for HN)
  - Anechoic or hypoechoic cavity
  - AP diameter of renal pelvis (correlates with likelihood of obstruction but not degree of obstruction which requires complementary radioisotope scans)
  - False-positive, due to either:
    - large extrarenal pelvis
    - peripelvic renal cyst
    - nonobstructive hydronephrosis due to VUR [i.e. hydroureteronephrosis (HUN)]

Investigations

- **Radioisotope Scan**
  - MAG3 (rather than DTPA) is the scan of choice.
    - Drainage curve of an obstructed kidney fails to decline (t½ >20 min) even after the administration of diuretics [IV furosemide (0.5–1.0 mg/kg)].
    - Reduction in differential renal function (<40%) → key sign of UPJ obstruction.

- **Intra-Venous Pyelography (IVP)** (uncommonly used nowadays)
  - Shows dilatation of the renal calyces and pelvis, funneling down to a narrow beak end, with nonvisualization of the ipsilateral ureter.
  - IV furosemide (0.5–1.0 mg/kg) may help in differentiating true UPJ obstruction from nonobstructive hydronephrosis.

Treatment

- Antenatally detected HN
  - Conservative management in group with good renal function (>40%).
  - Pyeloplasty (needed in <50%) for group with functional deterioration and increasing dilatation.

- Conventional open **OR** laparoscopic dismembered pyeloplasty
  - "Anderson–Hynes" excision of the narrowed segment, spatulation, and anastomosis to the most dependent portion of the renal pelvis.

- Endourological pyeloplasty
  - Use of balloon dilatations, percutaneous antegrade endopyelotomy, and retrograde ureteroscopic endopyelotomy.

Megaureter

Megaureter is classified as:

- Obstructed (M:F 4:1 | L > R | Bilateral 20%)
  - Primary: adynamic juxtavesical segment of the ureter.
  - Secondary: ↑ vesical pressures (e.g., PUV or a neurogenic bladder (NB)).
- Refluxing
  - Primary: severe VUR.
  - Syndromic: Megacystitis megaureter syndrome.
  - Secondary: ↑ vesical pressures (e.g., PUV or a neurogenic bladder (NB)).
- Obstructed/refluxing
- Nonobstructed/nonrefluxing

Clinical Features

- Increasing HUN
- Decrease in renal function
- UTI
- Recurrent flank pain

Investigations

- US → HUN
- MAG3 scan → degree of obstruction and differential renal function
- IVU → to see anatomy (uncommonly used)
- VCUG → r/o VUR

Management

- Ureteral reimplantation: when associated with severe VUR or obstruction.
  - Mobilize the megaureter via an intravesical, extravesical, or combined.
  - Reduce ureteral caliber.
  - Antireflux reimplant.

- Nephroureterectomy: when the function of the kidney drained by a megaureter is severely impaired.

Vesico-Ureteric Reflux

(VUR)

Vesico-Ureteric Reflux (VUR)

- Reflux is noted in up to 40% of fetuses
- Female predominant
- Peak incidence at 3 years
- Familial incidence is 2–4% of all cases
- Unilateral or bilateral (60%).

Pathology

- **Primary VUR:**
  - due to a short submucosal tunnel [intravesical “tunnel length : ureteral width” should be about 5:1 to prevent VUR]

- **Secondary VUR:**
  - due to either:
    - PUV
    - Anterior urethral valves
    - Neurogenic bladder (NB)
    - Ureteroceles
    - Bladder diverticula
    - Ectopic ureters associated with duplex system

Clinical Features

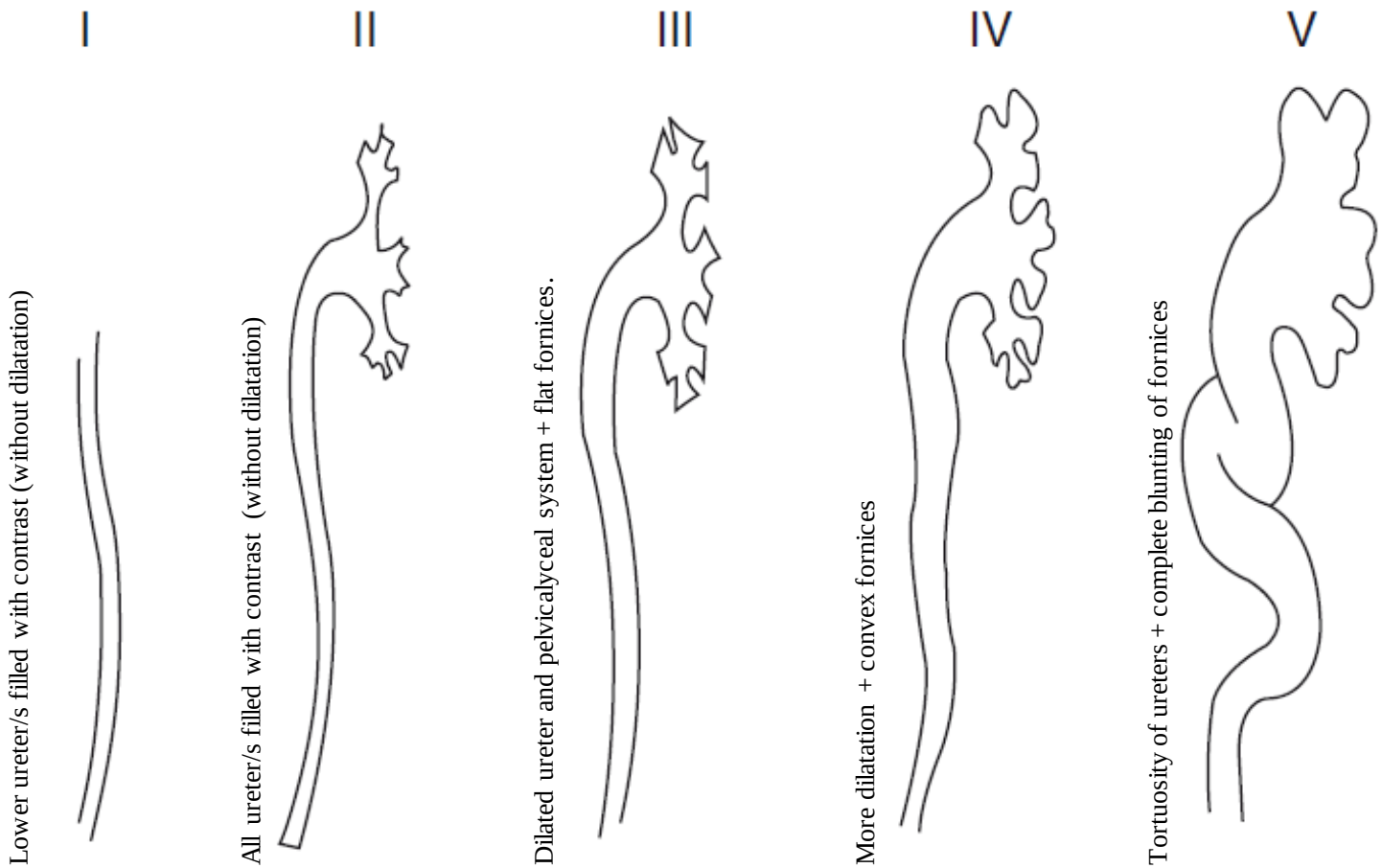
- Symptoms of UTI
- Renal scarring (radiologically demonstrated scarring is almost always due to VUR)
- Renal dysfunction (renal concentrating ability and deterioration of GFR)
- Hypertension
- Reduced somatic growth

Investigations

- Urine analysis → r/o infection
- US → HUN
- MAG3 scan → differential renal function
- DMSA → for renal scars and differential renal function
- VCUG → for degree of VUR
- GFR → for baseline renal function
- Direct isotope cystography (DIC) → for follow-up scans

Vesico-Ureteric Reflux (VUR)

- Grading



Vesico-Ureteric Reflux (VUR)

Management

- Low-grade reflux is more likely to resolve spontaneously with age (in the absence of any malformation).

- Management options:
  - Antibiotic chemoprophylaxis
    - Long-term prophylactic antibiotics (few months up to 2 years).
  - Submucosal injection of bulking agent
    - Subureteric injection of Deflux® (dextranomer microspheres in sodium hyaluronic solution)
    - Results better for lower grades of reflux (>80% success) (may be repeated to improve success rate)
    - Less successful in children with neurogenic bladder (NB)

Vesico-Ureteric Reflux (VUR)

Surgery

- Indications:
  1. Failure of management with chemoprophylaxis and/or submucosal injection therapy.
     - Therapy given for a maximum period of 2 years
     - Poor compliance to medical management
     - Persistent UTI or breakthrough infections (>2/year)
     - Appearance of new scars or enlargement of pre-existing scars
     - Deterioration of renal function
  2. Secondary VUR due to anatomical anomalies (Hutch diverticulum, ureterocele, duplex ureter, primary megaureters, PUV, and neurogenic bladder).
  3. Higher grades (IV, V) at any age.
  4. Hypertension due to renal cause.
  5. Single kidney with higher grade of VUR.
  6. Decrease in renal growth +/- somatic growth.

Vesico-Ureteric Reflux (VUR)

Surgery

- Reimplantation of ureters
  - Transtrigonal ureteric (Cohen) reimplantation [most common]
  - Intravesical technique (Leadbetter–Politano)
  - Extravesical detrusorraphy technique (Lich and Gregoir)
  - ± Ureteric tapering or plication

Vesico-Ureteric Reflux (VUR)

Postoperative Management

- Extravesical drains and ureteric catheters for 7–10 days.
- Antibiotics for ~3 months.
- Follow-up VCUG or DIC (to confirm absence of reflux).

Complications

- Persistent reflux (usually lower grade and improves spontaneously on continued antibiotics)
- Ureteric obstruction (due to devascularisation, kinking or torsion of the distal ureter)
- Intravesical calculi
- Injury to the bowel, fallopian tubes and/or vas deferens

Vesico-Ureteric Reflux (VUR)

Outcome

- Medical management:
  - successful in low grades of reflux
  - has to be continued for a long period

- Surgical repair:
  - success rate from 93 to 98%
  - corrects VUR but does not reverse scarring nor parenchymal damage

- Deflux® injections have reduced the number of surgical procedures performed in the recent past.

- The main factor determining the outcome is the extent of renal scarring or renal parenchymal damage at the onset.

Neurogenic Bladder

(NB)

Neurogenic Bladder (NB)

- Ideal bladder function
- Bladder should have the ability to fill to capacity while maintaining low pressures.
- Sensation to void when full bladder and be under voluntary control.
- Bladder should be empty at the end of voiding without residue.

Neurogenic Bladder (NB)

- Neurology
- Parasympathetic (predominant)
  - S2–S4 spinal segments, via pelvic nerves, to the detrusor muscle.

- Sympathetic
  - T9–L1 spinal segments, via sympathetic chain and hypogastric plexus, to the bladder neck.

- Somatic innervation
  - S2–S4 segments, via pudendal nerve, to the external sphincter.

Neurogenic Bladder (NB)

- Etiology

Congenital	Acquired
Myleomeningocele	Trauma
Spina bifida occulta (including tethered cord, lipoma of cord)	Tumors
Sacral agenesis	Infarction
	Transverse myelitis

Neurogenic Bladder (NB)

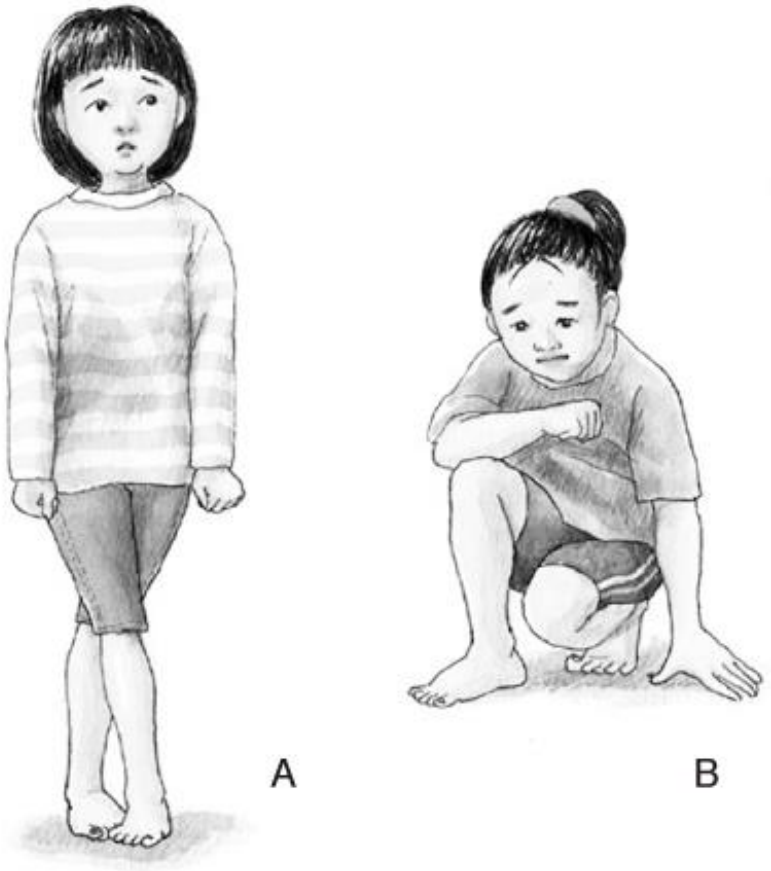
- Etiology

- Almost always due to spinal cord anomalies (congenital and acquired) (myelomeningocele most common).

- Nonneurogenic neuropathic bladder (Hinman–Allen syndrome) → psychological overlay in addition to bladder dysfunction.

Neurogenic Bladder (NB)

- Clinical Features
- Urge incontinence (overactive detrusor)
- Stress incontinence (underactive sphincter)
- Vincent's curtsy postures to inhibit voiding
- (cross-legged "A" or squat with heel pressir
- Assess back for evidence of:
  - spina bifida occulta (hairy patch or lumbosacral li
  - OR
  - sacral agenesis
- Full neurological assessment to elicit the integrity of segments S2–4



Neurogenic Bladder (NB)

- Concept
- “Unsafe” bladder → inability to empty distended bladder
- “Safe” bladder → leaks easily under pressure or usually empty on examination

Neurogenic Bladder (NB)

- Investigations
- US:
  - upper tract dilatation
  - postvoiding bladder residual volume
- Cystogram:
  - r/o VUR, bladder trabeculation or diverticular formation
- Urodynamic Studies [next slide]

Neurogenic Bladder (NB)

- Basic Video Urodynamics
- Normal bladder capacity (mL)
  - Infants capacity = weight (in kg) × 7
  - 1–12 years capacity = age (in years) × 30 + 30
- Detrusor hyperreflexia (significant if pressure is >30–40 cm H2O)
- Compliance (at expected bladder capacity, detrusor pressure should be <30–40 cm H2O)
- Sphincteric incompetence (opens abnormally at low pressures)
- Leak-point pressure (at which external sphincter opens)
  - <40 cm H2O → "safe" bladder
  - >40 cm H2O → potential for upper tracts deterioration
- Detrusor sphincter dyssynergia (detrusor contraction and external sphincter non-relaxation → ↑ intravesical pressures)
- [Intravesical pressure = detrusor pressure + intraabdominal pressure]

Neurogenic Bladder (NB)

- Patterns of Neurogenic Bladder
- Contractile bladder (usually the result of suprasacral cord lesions)
  - Innervation of the detrusor and external urethral sphincter are intact
  - Conus reflexes are positive
  - Voiding usually occurs by detrusor hyperreflexia and is associated with detrusor sphincter dyssynergia
  - Intravesical pressures are usually high (ultimately lead to secondary upper renal tract complications)

- Acontractile bladder
  - Innervation of both detrusor and external sphincter is destroyed
  - Conus reflexes are negative
  - Detrusor contractility is absent but some degree of sphincteric incompetence is always present
  - Voiding occurs either by overflow or by raising intraabdominal pressure
  - A degree of detrusor noncompliance is common

- Intermediate bladder dysfunction
  - Combination of anomalies: various degrees of detrusor noncompliance, detrusor hyperreflexia, and sphincteric incontinence

Neurogenic Bladder (NB)

- Management
- Natural history: progressive deterioration by the age of 3 years (in up to 60% of all children)
- Early institution of CIC can prevent both renal damage and secondary bladder wall changes ➔ improving long-term outcomes
- Both detrusor hyperreflexia and detrusor noncompliance can be treated:
  - medically with anticholinergics (e.g., oxybutanin and tolerodine)
  - +/- surgically (to increase the capacity and outlet resistance)

Neurogenic Bladder (NB)

- Clean intermittent catheterization (CIC) combined with an anticholinergic (oral or intravesical) is the standard therapy for neurogenic bladder.

Neurogenic Bladder (NB)

- Augmentation cystoplasty ± catheterizable conduit
- A cystoplasty using ileum is most commonly performed.
- Alternative conduits:
  - Common: ileocecum, sigmoid colon, and stomach
  - Less common: autoaugmentation, detrusorectomy, and ureterocystoplasty

Neurogenic Bladder (NB)

- Augmentation cystoplasty ± catheterizable conduit

- Complications of Cystoplasty
- Mucus production leading to catheter blockage, infection, and bladder stones.
- Metabolic changes (hyperchloremic alkalosis, electrolyte disturbance, systemic alkalosis (gastrocystoplasty).
- Spontaneous perforation.
- Metaplasia/malignancy.
- Bowel problems (diarrhea, vitamin B12 deficiency).
- Dysuria and hematuria (gastrocystoplasty).

Neurogenic Bladder (NB)

- Sphincteric Incompetence
- Medical treatment remains unsatisfactory but marginal improvement may be seen with alpha-adrenergic agonists such as ephedrine.
- Surgical Options:
  - Periurethral bulking agents, e.g., collagen, silicone, and Deflux®.
  - Bladder neck suspension and slings, e.g., Marshal–Marchetti bladder neck suspension and bladder neck slings.
  - Pippi-Salle procedure – urethral lengthening procedure suitable for girls but not for boys.
  - Mitrofanoff procedure – involves closure of the bladder neck and using the appendix as an appendicovesicostomy. The appendix is tunneled between the bladder and the abdominal wall, which is then used as a catheterizable stoma.
  - Artificial urinary sphincter.
  - Monti procedure – when the appendix is unavailable, a segment of ileum can be tubularized and used in a fashion similar to an appendicovesicostomy.
  - ACE procedure – most neurogenic bladders have associated bowel dysfunction due to neurogenic bowel. An appendix or Monti tube can be implanted between the cecum and the anterior abdominal wall as a catheterizable channel for antegrade continent enemas.

Posterior Urethral Valves

(PUV)

Posterior Urethral Valves (PUV)

- Posterior urethral valves remain the most common reasons for renal failure and renal transplantation in children.

Posterior Urethral Valves (PUV)

- Anatomy

- The male urethra is divided into four segments:
- Posterior
  - Prostatic urethra – from the bladder neck to the site of “urogenital diaphragm.”
  - Membranous urethra – “urogenital diaphragm”.

- Anterior
  - Bulbar urethra – from the distal margin of the urogenital diaphragm to penoscrotal junction.
  - Penile urethra – urethra that traverses the penile shaft including the glans.

Posterior Urethral Valves (PUV)

- Congenital Obstructing Posterior Urethral Membrane
  - This is a newer concept whereby the uninstrumented infants urethra looks more like a circumferential obstructing membrane with a small central opening, which following catheterization or instrumentation reverts to the classical “valve” appearance.

- Traditionally, three types of posterior urethral valve has been described.

Posterior Urethral Valves (PUV)

Table 5.5.1 Young's Classification[1]

	Description	Result
Type I (~95%)	Bicuspid valve from posterior edge of the verumontanum extending distally and anteriorly and fusing in the midline	Obstructive
Type II	Prominent longitudinal folds extending from verumontanum toward the bladder neck	Nonobstructive
Type III (5–10%)	Circumferential ring distal to the verumontanum at the level of the membranous urethra	Obstructive

NB, Cobb's collar[2] (or congenital urethral stricture) – distal membrane with a central opening within bulbous urethra

Posterior Urethral Valves (PUV)

- Clinical Features
- Antenatal (66%):
  - dilated posterior urethra and bladder ("key-hole" sign) ± hydroureteronephrosis in male fetus on maternal US
  - renal echogenicity and whether there is evidence of oligohydramnios (possible cause of hypoplastic lungs)

- Postnatal (33%):
  - Poor stream ± palpable bladder
  - UTI
  - Renal failure with poor somatic growth and lethargy
  - Diurnal incontinence

Posterior Urethral Valves (PUV)

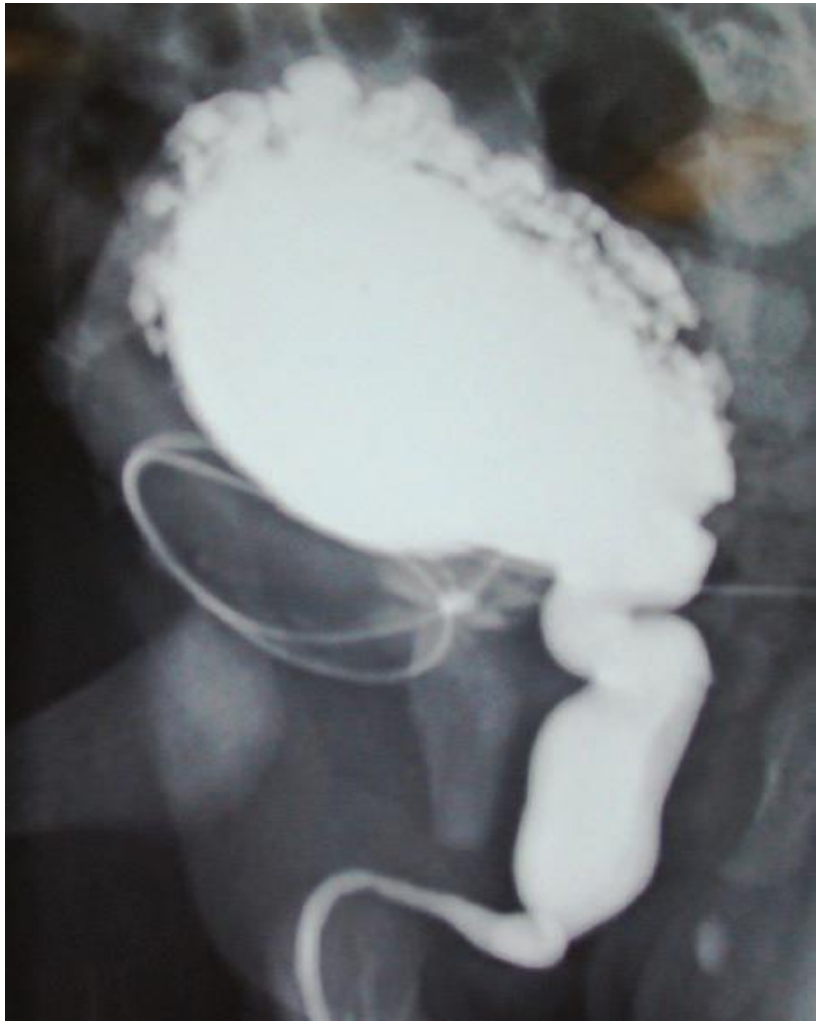
- Investigation

- Micturating Cysto-Urethrogram (MCUG)
  - Wide posterior urethra and prominent bladder neck
  - Partial filling of anterior urethra
  - Posterior urethral bulging forward over the bulbous urethra
  - Valve leaflet lucencies - occasionally.

Posterior Urethral Valves (PUV)

- Worse Prognostic Factors
- Antenatal factors
  - Gestation at detection (<24 weeks).
  - US appearance (renal cystic changes imply renal dysplasia).
  - Oligohydraminos.
  - Fetal urine analysis (usually hypotonic with Na<100 mmol/L and Cl<90 mmol/L.
  - ↑ Na >100, Cl >90, and b-2-microglobulin >40, urine osmolality >210 mOsm, and urine output <2 mL/h

- Postnatal factors
  - Lowest serum creatinine >1 mg/dL at 1 year of age.



MCUG showing posterior urethral valves with trabeculated bladder with pseudo-diverticuli

Posterior Urethral Valves (PUV)

- Management
- Drain the bladder
  - Infant “feeding” tube is ideal (avoid Foley --> stimulate bladder spasms in the already hypertrophic bladder + can cause secondary ureteric obstruction)
  - Suprapubic catheter (if urethral route is unsuccessful)
- Prevent infection (Prophylactic antibiotics)
- Preserve renal function (intensive IV fluids and electrolyte balance)

Posterior Urethral Valves (PUV)

- PUV Ablation

- Transurethral diathermy ablation and division of the valve leaflets at the 5 and 7 o’clock (Nd YAG laser used occasionally).

- Temporary Urinary Diversion
  - If ablation not feasible (small infant or lack of scopes)
  - Include vesicostomy, ureterostomy, or pyelostomy

Posterior Urethral Valves (PUV)

- Temporary Urinary Diversion

- Vesicostomy
  - Dome of the bladder bought to the skin midway between the umbilicus and the symphysis pubis.
  - Allows drainage but also cycles urine to the bladder and maintains volume.
  - Closed once renal function has stabilized, upper tracts have diminished, and the child is large enough for a valve ablation.
  - Valve ablation should be performed at the time of vesicostomy closure to avoid urethral stricture developing in a “dry” urethra.

- Higher levels of drainage (bilateral ureterostomy or pyelostomy)

Posterior Urethral Valves (PUV)

- Urinary Ascites (5–10%)
- Usually secondary to urine leak from a renal fornix blowout, renal parenchymal rupture, or bladder perforation.
- Abdominal distension can be marked and cause respiratory compromise.
- Peritoneal absorption of urine can lead to uremia.
- Protect the kidneys from the deleterious effects of constant high back pressure from bladder.

- Vesicoureteric Reflux (50%)
- Bilateral in 50%.
- Reflux subsides with effective valve ablation (~30%).
- Persistent reflux (~30%).
  - Persistent VU reflux can be treated with STING procedure or ureteric reimplantation provided that bladder function is normal on urodynamic evaluation.

Posterior Urethral Valves (PUV)

- VURD Syndrome (Valves Unilateral Reflux Dysplasia)
  - Severe unilateral reflux into a dysplastic kidney.
  - Protects the contralateral kidney from back pressure (“pop-off” mechanism).

- Other “pop-off” mechanisms
  - Large bladder diverticula
  - Urinary ascites
  - Patent urachus

Posterior Urethral Valves (PUV)

- Bladder Dysfunction and the “Valve Bladder”

- Voiding dysfunction is extremely common in children with PUV.
- Secondary to the long-standing obstruction and mural hypertrophy and fibrosis (“valve” bladder).
- Urodynamic patterns can change over time from bladder instability during infancy to myogenic failure in older boys.
- Bladder dysfunction manifesting as incontinence and persistence of upper tract dilatation is being increasingly recognized as one of the factors responsible for eventual renal deterioration.

Posterior Urethral Valves (PUV)

- Bladder Dysfunction and the “Valve Bladder”
- The underlying mechanisms may be:
- Urine concentrating defects
  - Long-standing back pressure leads to the renal tubular dysfunction causing an acquired form of nephrogenic diabetes insipidus.
  - ↑ Urine volume exacerbates incontinence and upper tract dilatation.

- Persistent upper tract dilation
  - ↑ Urine output and hold up at the VUJ – caused by ureter passing through the thick, noncompliant bladder wall.
  - Upper tract pressure studies (Whitaker’s test) have shown that the VUJ obstruction is not constant but increases as the bladder fills.

Posterior Urethral Valves (PUV)

- Bladder Dysfunction and the “Valve Bladder”
  - Treatment is difficult and ADH treatment is not usually successful.
  - Timed voiding in older cooperative children and overnight free drainage in children with a continent catheterisable channel (to ensure lower intravesical pressures) can protect upper tracts.

Posterior Urethral Valves (PUV)

- Urodynamic Patterns and treatment options
- Instability – irregular contractions leading to pain and incontinence.
  - Anticholinergics, e.g., oxybutynin and tolterodine.

- Hypocontractility – leading to overflow incontinence and impedance of upper tract drainage. This so-called "myogenic failure" has been attributed to long-term obstruction or prolonged use of anticholinergics.
  - Clean intermittent catheterization or a catheterisable stoma (e.g., Mitrofanoff).

- Detrusor sphincter dyssynergia – bladder contracts against an unrelaxed sphincter.
  - Historically, bladder neck hypertrophy as seen on the MCUG was thought to play a significant role. Many children went on to have bladder neck incisions. It improved voiding symptoms and upper tracts dilatation in some, but in others it also caused incontinence and retrograde ejaculation.
  - alpha blockers – e.g., doxazocin

- Poor compliance/small volume bladder
  - bladder augmentation

Posterior Urethral Valves (PUV)

- Renal Transplantation
- Up to 30% will ultimately require renal replacement therapy.
- Persistent bladder dysfunction decreases graft survival post transplant.

Posterior Urethral Valves (PUV)

- Fertility Issues

- Diminished fertility possibly due to
  - ↑ Posterior urethral pressure in utero may affect prostate development.
  - ↑ Incidence of UDT.
  - Semen analysis has shown a much thicker ejaculate with decreased sperm motility.
  - Voiding dysfunction and retrograde ejaculation.

Hypospadias

Hypospadias

Hypospadias: is a complex of an abnormal ventral meatus, chordee, and a dorsal hooded foreskin.

- Usually isolated, it can be part of the DSD spectrum.
- One in 300 live-births (but wide variation from 1 in 110 to 1 in 1,250)
- Associated with ↑ parity, ↑ maternal age, and ↓ birth weight
- Associated with inguinal hernia & hydrocele (~10%), and undescended testes (~8%)

Hypospadias

Embryology

- Development of male urethra takes place between 8^{th}-15^{th} week of gestation under the influence of testosterone.

Hypospadias

Etiology **(genetic, endocrine, & environmental)**

1. Genetic Factors:
   - Exact mode of inheritance is unknown.
   - Monozygotic twins: x8 increase in the incidence of hypospadias
   - +ve family history: 8% of fathers and 14% of brothers.

Hypospadias

Etiology

2. Endocrine Factors:

- Deficient androgenic stimulation: defective testosterone production, conversion (to dihydrotestosterone (DHT)), or reduced sensitivity (androgen insensitivity syndrome).

- Increased maternal progesterone exposure: x5 increase in the incidence among boys conceived by IVF (progesterone is commonly administered). Progesterone is a substrate for 5AR and causes competitive inhibition of conversion of testosterone to DHT.

Hypospadias

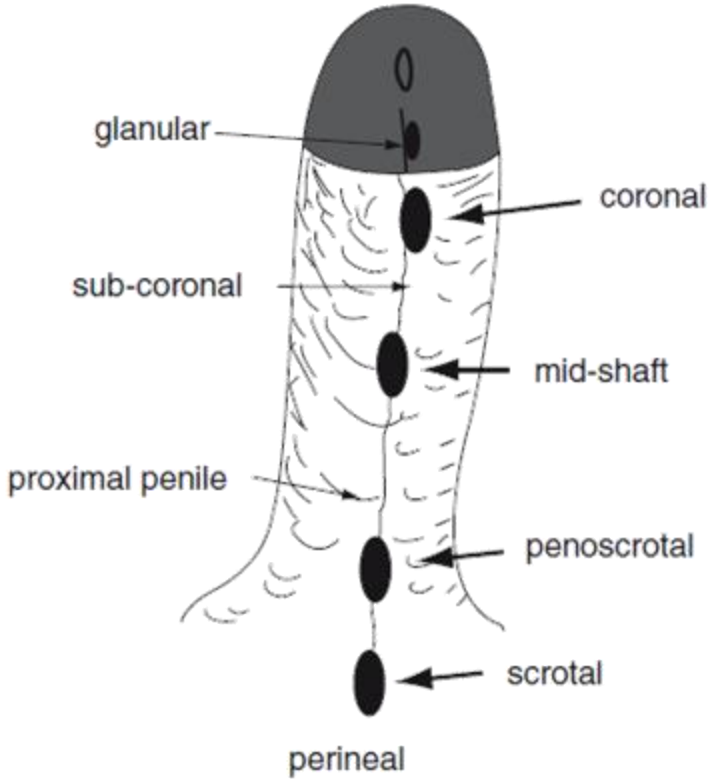
Etiology

3. Environmental Factors:
   - As maternal exposure to estrogenic substances (contained in pesticides, milk, plastic linings of metal cans, and pharmaceuticals).

Hypospadias

Clinical Features

- Classification
  - Distal (glanular, coronal, and subcoronal) (50%)
  - Middle (distal penile, midshaft, and proximal penile) (30%)
  - Proximal (penoscrotal, scrotal, and perineal) (20%)



- True location of the meatus should be ascertained after correction of the penile curvature.

Hypospadias

Clinical Features

- Although usually isolated, it is important to identify a disorder of sexual differentiation (DSD) – suggested by impalpable testes for instance.
- DSD occurs in ~15% of cases if gonad is palpable but ~50% of cases if impalpable.
- Evaluate using US, genitography, biochemical, and chromosomal analysis before making final decisions regarding definitive management.

Hypospadias

Clinical Features

- In terms of choice of surgical options, assess hypospadias by:
  - location of the meatus
  - degree of chordee
  - penile size
  - quality of ventral and proximal shaft skin
  - quality of distal urethral plate
  - depth of glanular groove

Hypospadias

Surgery

- A hypospadiac penis looks abnormal, may interfere with normal voiding in the standing position, and possibly affect fertility by precluding effective insemination.

- **Goal of surgery:** is to create a cosmetically acceptable penis, which allows normal voiding with a forward stream and normal vaginal penetration.

- Some cases of glanular hypospadias can be left entirely alone.

Hypospadias

Preoperative

- The current recommendation is to complete the repair of hypospadias before 18 months of age (minimizes psychological impact of genital surgery).

- Hormone manipulation: penile size can be increased by weekly intramuscular injections of testosterone or hCG or topical application of testosterone or DHT for 4–6 weeks before surgery.

Hypospadias

Orthoplasty (correction of chordee)

- Straightening may be achieved by complete degloving of the penis, excision of a fibrous corpus spongiosum, and dissection of the urethral plate.

- Dorsal plication of tunica albuginea and/or ventral application of a dermal graft may sometimes be required.

Hypospadias

Urethroplasty

- Advancement techniques (e.g., MAGPI)
- Tubularization techniques (e.g., Snodgrass)
- Local tissue flaps (e.g., Mathieu's perimeatal-based flap)
- Two stages using local or extragenital free grafts (e.g., preputial skin, buccal mucosa, and postauricular skin)

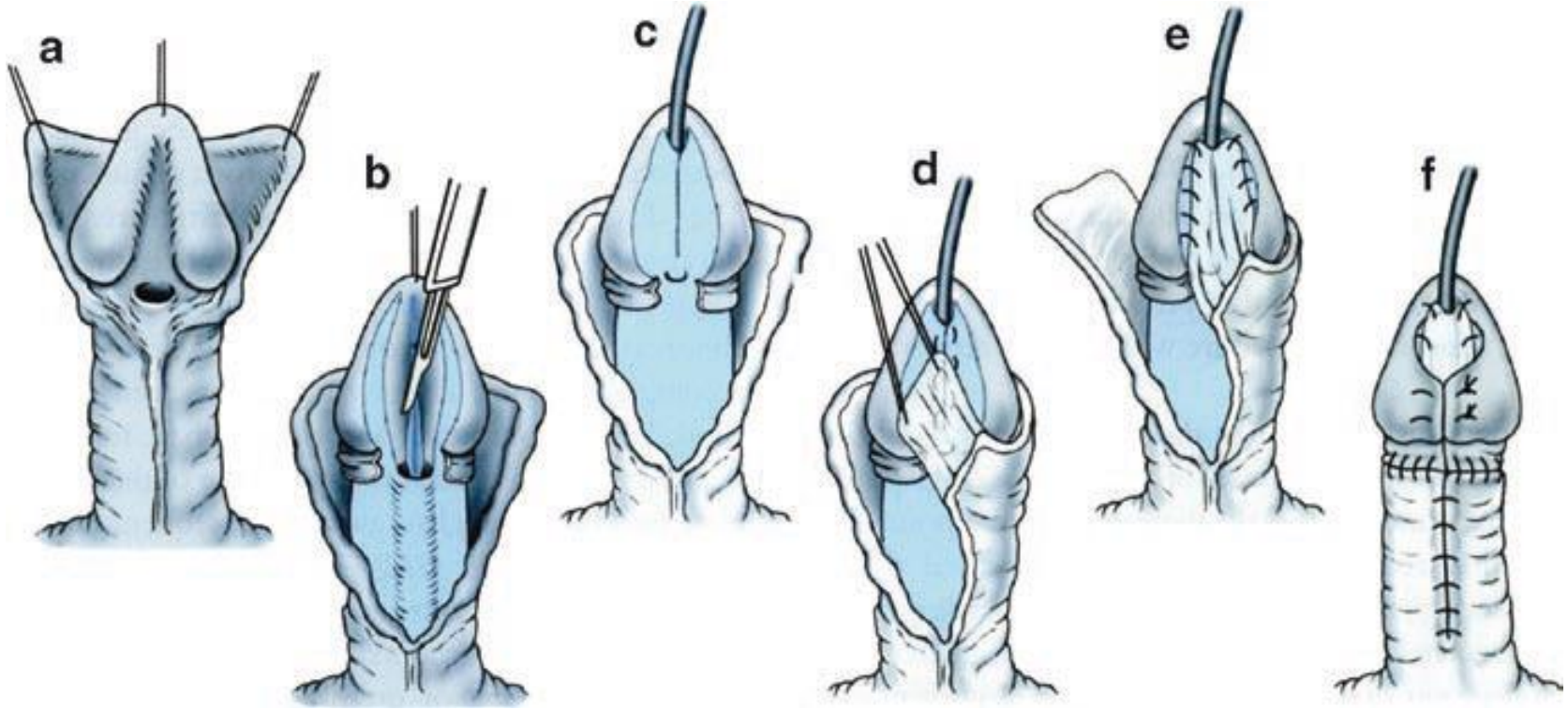
Hypospadias

Meatoplasty and glanuloplasty

- The common goal is to create a slit-like meatus and cover the distal neourethra with flaps of glans epithelium.

Skin coverage

- Usually achieved by ventral transfer of preputial skin with any redundant skin trimmed away to resemble that of a circumcision.

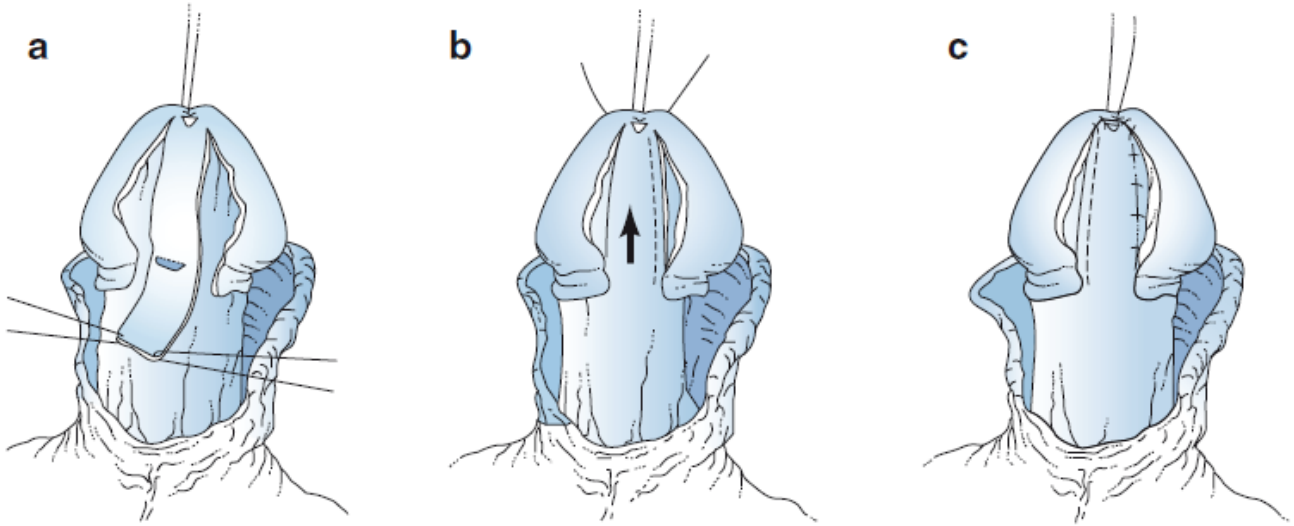


Snodgrass repair

- Adapted from Belman et al. (Belman AB, King LR, Kramer SA (eds) (2002) Clinical pediatric urology, 4th edn. Martin Dunitz, London, p 1077)

Mathieu (meatal based flap)

- Belman AB, King LR, Kramer SA (eds) (2002) Clinical pediatric urology, 4th edn. Martin Dunitz, London, p 1076



Postoperative Phase

- The bladder is drained for 1 week with a “dripping stent,” simple analgesics, and an antibiotic.

- Early complications:
  - Bleeding
  - Hematoma
  - Infection
  - Breakdown of repair

- Late complications:
  - Persistent chordee
  - Meatal stenosis
  - Urethrocutaneous fistula (UCF)
  - Urethral stricture
  - Urethral diverticulum

Circumcision

Natural History of Foreskin Separation

- At birth, the foreskin is naturally adherent to the glans.
- From about 2–4 years of age, there is a dissolution of the adhesions allowing the foreskin to retract.
- From about 5 years, most boys should have the ability of full unimpeded foreskin retraction.

Phimosis

- A condition where the foreskin is unable to be retracted to expose the glans.

Types:

- Physiological phimosis – the normal state in the first years of life
- Pathological phimosis:
  - **Primary**: true congenital phimosis with pin-hole meatus.
  - **Secondary**:
    - Secondary to bacterial infection that may lead to:
      - **balanitis** (inflammation of the glans)
      - **posthitis** (inflammation of the foreskin).
    - Balanitis xerotica obliterans (BXO) [long-standing chronic inflammation, with a dead-white appearance of glans and foreskin, and typically a pin-hole meatus].

Management

- **Conservative**:
  - Reassure, gentle self-retraction
  - Topical steroids (e.g., betamethasone 0.1%)

- **Surgery**:
  - Preputial “stretch”
  - Or Preputioplasty
  - Or Circumcision

Paraphimosis

- The condition where the foreskin is able to be retracted but becomes stuck in that position resulting in distal congestion and edema of glans.

Management

- A surgical emergency

- Manoeuvres can be done in ER:
  - Compresses with ice or sugar (to reduce the swelling and allow protraction)
  - Multiple needle punctures (again to allow fluid to be squeezed out)

- if any of the above failed → send to OR → dorsal slit of the tight band +/- circumcision (under GA)

Circumcision

History

- It is an act of faith in both Jewish and Muslim religions for males to be circumcised:
  - Jewish: have a very precise chronology (eighth day of life) and liturgy (Bris milah)
  - Muslim: actual timing is less important, and it tends to be done at 1st 1-2 weeks of life.

- Although in general, it is not a necessary part of most Christian faiths, various West African churches still encourage it.

Circumcision

Surgical Freehand

- Complete foreskin retraction and division of adhesions.
- Dorsal slit, then circumferential incision around outer skin.
- Hemostasis with bipolar (not monopolar) cautery [attention to frenular artery].
- Similar incision on inner preputial skin leaving 3–7 mm cuff at corona.
- Reconstruction (absorbable suture)

Surgical using specific clamps

- Plastibell®
- Gomko clamp®
- Winkelmann clamp®
- Mogen clamp

Circumcision

Contraindications

- Absolute:
  - Family history of bleeding disorders
  - The Newborn has known bleeding tendency/disorder, or pathological jaundice
- Relative:
  - Hypospadias (as foreskin helps in the surgical repair of hypospadias)

Circumcision

Complications

- Bleeding
- Infection
- Meatal stenosis
- Insufficient/excessive foreskin removed
- Adhesions, skin bridges, or inclusion cysts
- Entrapped penis or secondary phimosis (when circumcision is performed on a boy with buried penis)
- Urethral injury (iatrogenic hypospadias)
- Necrosis of the penis (injudicious use of electrocautery to control bleeding)
- Amputation of the glans (partial or complete)
- Death (mainly due to unnoticed bleeding)

Disorders of Sex Development

(DSD)

Disorders of Sex Development (DSD)

- Rarely, genital appearance at birth may be ambiguous or even contrary to genetic sex.
- Should be regarded as a medical emergency to achieve the best gender outcome for the baby.

Disorders of Sex Development (DSD)

- Ince 2006, the phrase disorders of sexual development (DSD) has been advocated, and appears preferable to the older terms:
    - ambiguous genitalia
    - Hermaphroditism
    - Pseudohermaphroditism
    - intersex

Disorders of Sex Development (DSD)

- Embryology

- From 6 weeks, early bipotential gonads develop within the genital ridge from primordial germ cells.

Disorders of Sex Development (DSD)

- Clinical Features
- The presentation tends to be either that of an under-virilized male, or an over-virilized female (there are exceptions).
- The birth of an infant with ambiguous genitalia will be a cause of much anxiety for the parents and in some cases (congenital adrenal hyperplasia), the sequelae may be life-threatening.
- This event should, therefore, be regarded as medical emergency and subsequent management should proceed in a logical and timely manner in a specialized center with multidisciplinary input.
- It is important to avoid guessing the sex and using pronouns such as “he” or “she.” Phrases such as “your baby” are more appropriate.

Disorders of Sex Development (DSD)

- **Clinical Features**
- Particular points to be noted when examining include:
  - Presence of a palpable gonad on either side.
  - Appearance of phallus (length, width, and chordee).
  - Appearance of scrotum/labioscrotal folds, degree of skin rugosity/pigmentation.
  - Location of external urethral opening and number of orifices present on the perineum.
- A proportion of children with DSD will only present later in life, such as those with Swyer syndrome who look unequivocally female (XY, complete gonadal dysgenesis), but who are unable to develop secondary sexual characteristics or menstruate.

Disorders of Sex Development (DSD)

- **Symmetrical** genital appearance imply a biochemical etiology (e.g., congenital adrenal hyperplasia (CAH))
- **Asymmetrical** appearance implies a chromosomal abnormality (e.g., mixed gonadal dysgenesis)

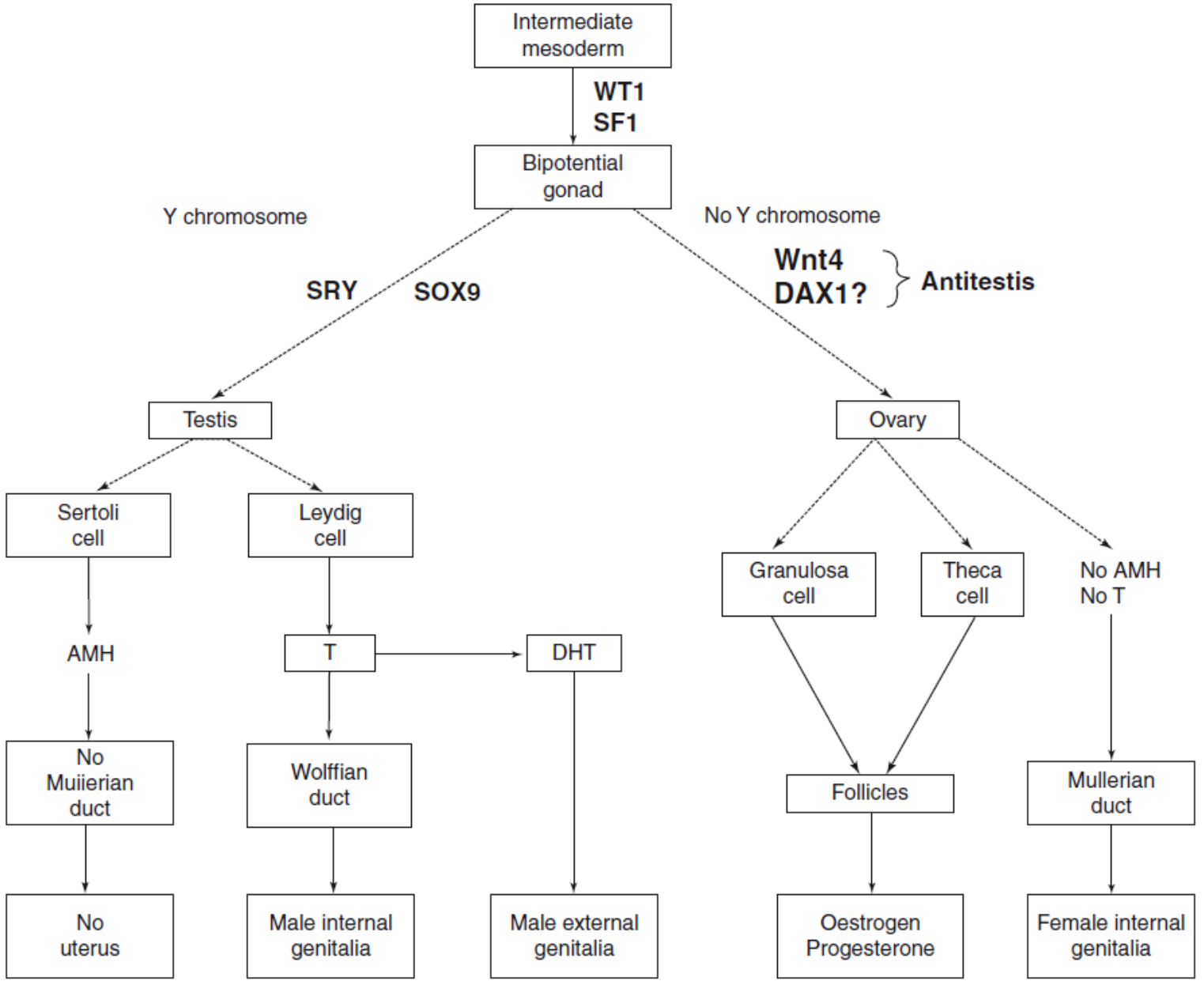


Fig. 5.8.1 Embryology of sexual development

Embryology of sexual development

Table 5.8.1 Classification of DSD (after Lee PA, Houk CP, Hughes IA, Ahmed SF, Houk C, et al. Consensus statement on management of intersex disorders. Pediatrics 2006; 118:e488-e500)

	Variant	Notes
46XY DSD (undervirilized male)	Severe hypospadias	
	Androgen insensitivity syndromes (complete and partial)	Female phenotype, *AR* mutations
	5 α-Reductase deficiency	Failure to produce more active DHT
	Complete gonadal dysgenesis (Swyer syndrome (Swyer GI (1955) Male pseudohermaphroditism: a hitherto undescribed form. Br Med J 2:709–712); 46XY sex reversal)	Female phenotype, *SRY* mutations (~20%), usually detected at puberty
	Leydig cell hypoplasia	Autosomal recessive
	Disorders of anti-Mullerian hormone (AMH) production and receptor	*AMR* and *AMHR-II* mutations
46XX DSD (overvirilized female)	Congenital adrenal hyperplasia	*Vide infra*
	Fetoplacental aromatase deficiency	
	Exogenous androgen exposure	
	Ovotesticular DSD	
Sex chromosome DSD	45XO (Turner[2] syndrome)	Short, webbed neck, female phenotype, and aortic coarctation (60%)
	45XO/46XY (mixed gonadal dysgenesis)	
	46XX/46XY (chimeric ovotesticular DSD)	

Disorders of Sex Development (DSD)

- **Investigations**
  - Genetic – karyotype and specific gene arrays
  - Endocrine – blood and urine biochemistry, hormone assays
  - Imaging – ultrasound (renal/pelvic), contrast studies (cystogram/genitogram), and MRI
  - Surgical – cystovaginoscopy, laparoscopy, and skin/gonadal biopsies
- It is important that the birth is not registered until the final decision has been made.

Disorders of Sex Development (DSD)

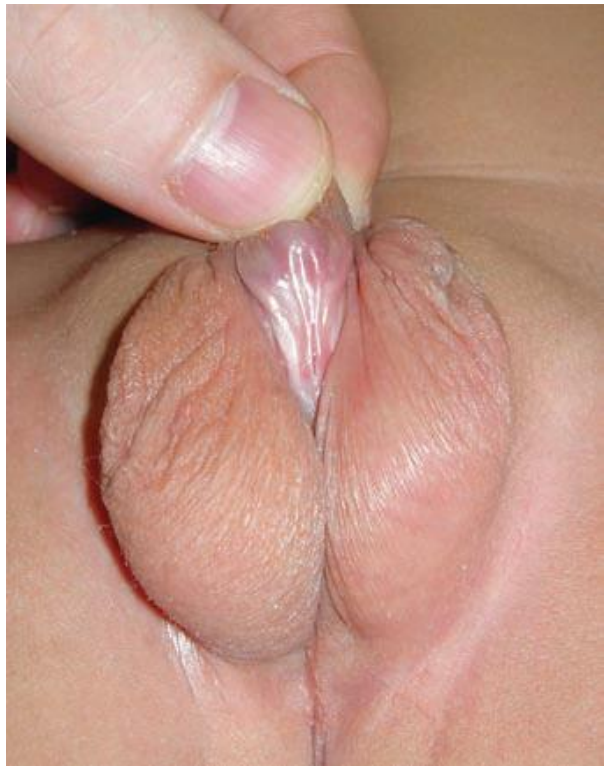
- Wherever possible, gender is assigned according to karyotype, rather than being reassigned to suit the initial appearances of the external genitalia, although this may not always be possible.

Disorders of Sex Development (DSD)

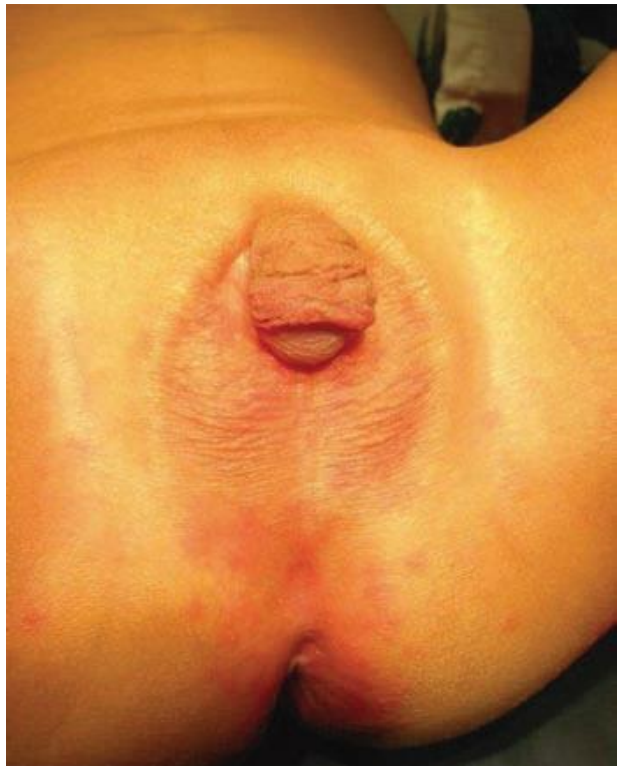
- **Principles**
  - Once a karyotype has been obtained, subsequent management should focus on:
    - Hormone replacement and electrolyte balance if indicated
    - Delineation of anatomy (US, cystovaginoscopy, MRI, and laparoscopy)
    - Need for and timing of reconstruction
    - Management of gonads
      - ↑ Risk of malignancy, streak ovaries → gonadoblastoma
    - Genetic counseling regarding future pregnancies
    - Psychological support for family and patient

Disorders of Sex Development (DSD)

- **Congenital Adrenal Hyperplasia**
  - Usually 21-Hydroxylase Deficiency
  - Appear as infants with ambiguous genitalia and bilateral impalpable gonads.
  - Can become life-threatening due to salt-losing nature (75%) with ↓ aldosterone.
  - Workup:
    - Urgent karyotype – XX.
    - Steroid profile – ↑ 17-hydroxy progesterone ↑ androstenedione levels.



46,XY DSD (severe hypospadias)



46,XX DSD (congenital adrenal hyperplasia)

Disorders of Sex Development (DSD)

- Surgery

- 46,XY DSD (Undervirilized Male)
- The main issues for this group include:
    - Hypospadias – cosmetic/functional aspects
    - Undescended testes/impalpable gonads (?malignant potential).
    - Streak gonads or intra-abdominal gonads at high risk of malignancy (e.g., mixed gonadal dysgenesis) should be removed.
    - Assessment and management of persistent Müllerian structures. Often asymptomatic and if so do not require excision. Recurrent infection or postmicturition dribbling may necessitate their removal.

Disorders of Sex Development (DSD)

- Surgery
- 46,XX DSD (Overvirilized Female)
- The main issues are to improve the external cosmetic appearance, preserve clitoral function, and separation of the urethra and vagina.
- Feminizing genitoplasty may include the following:
  - Clitoroplasty including recession, concealment, or reduction.
  - Vaginoplasty – to separate vagina and urethra.
  - Total or partial urogenital mobilization, vaginal pull-through, or replacement.
  - Introitoplasty to produce a more feminized appearance.

Thank You