

# UROLOGY

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## HISTORY

- ❑ pain: location (CVA, genitals, suprapubic), onset, quality (colicky, burning), severity, radiation
- ❑ associated symptoms: fever, chills, weight loss, nausea, vomiting
- ❑ irritative symptoms: frequency, nocturia, dysuria, urgency
- ❑ obstructive symptoms: hesitancy, straining, intermittency, decreased force or caliber of stream, prolonged voiding, post-void dribble, incomplete emptying
- ❑ incontinence: stress incontinence, urge incontinence, overflow incontinence, history of neurological problems, past pregnancies and method of delivery, past abdominal-pelvic operations
- ❑ urine: hematuria, pneumaturia, foul smell, colour (cloudy, white, orange)
- ❑ urethral discharge (colour, amount, smell), sexual history, UTIs, external skin lesions, lymphadenopathy
- ❑ others: renal calculi, infertility, erectile dysfunction, congenital disorders, family history of urological disease

## PHYSICAL

- ❑ inspection
  - abdomen: masses, scars from previous operations, suprapubic distension, hair distribution
  - penis: circumcision, phimosis/paraphimosis, epispadias, hypospadias, urethral discharge, superficial ulcers or vesicles, venereal warts, meatal stenosis
  - scrotum: testicular atrophy, testicular asymmetry, balanitis, dilated veins (varicocele) on standing, scrotal erythema/edema/cysts/hemangiomas
- ❑ palpation
  - examine all of the following: kidneys, bladder, penis, testes, vas deferens, epididymis, prostate
  - abdomen: masses, CVA tenderness, suprapubic distension/tenderness (examine for dullness on percussion), lymphadenopathy
  - penis: Peyronie's plaques, penile masses, penile tenderness
  - scrotum: scrotal tenderness/masses (size, consistency, location, mobility, shape), hernia, hydrocele, spermatocele, spermatic cord (varicocele, fusiform enlargement, thickening of the cord), absence of vas deferens, epididymal size/induration/tenderness
  - prostate (on DRE): size, consistency (rubbery, hard, boggy, indurated), nodularity (size, location), tenderness, warmth

# KIDNEY AND URETER

## RENAL STONE DISEASE

### Incidence

- ❑ 10% of population
- ❑ male:female ratio 3:1
- ❑ 50% chance of recurrence by 5 years

### Clinical Presentation

- ❑ urinary obstruction --> distension --> pain
  - flank pain from renal capsular distension (non-colicky)
  - severe waxing and waning pain radiating from flank to groin due to stretching of collecting system or ureter (ureteral colic)
  - never comfortable, always moving
- ❑ nausea, vomiting
- ❑ hematuria, usually microscopic, occasionally gross (90%)
- ❑ symptoms of trigonal irritation (frequency, urgency), diaphoresis, tachycardia, tachypnea
- ❑ +/- fever, chills, rigors secondary to pyelonephritis

### Differential Diagnosis of Renal Colic

- ❑ other causes of acute ureteral obstruction
  - UPJ obstruction
  - sloughed papillae
  - clot colic from gross hematuria

- ❑ radiculitis (L1 nerve root irritation)
  - herpes zoster
  - nerve root compression
- ❑ pyelonephritis (fever, chills, pyuria)
- ❑ acute abdominal crisis (biliary, bowel)
- ❑ leaking abdominal aortic aneurysm

#### Location of Stones

- ❑ calyx
  - may cause flank discomfort, recurrent infection or persistent hematuria
  - may remain asymptomatic for years and not require treatment
- ❑ pelvis
  - tend to cause UPJ obstruction
- ❑ renal pelvis and one or more calyces
  - staghorn calculi
    - often associated with infection
    - infection will not resolve until stone cleared
    - may obstruct renal drainage
- ❑ ureter
  - < 5 mm diameter will pass spontaneously in 75% of patients
- ❑ the three narrowest passage points for upper tract stones include: UPI, pelvic brim, UVJ

#### Investigations

- ❑ screening labs
  - CBC --> elevated WBC in presence of fever suggests infection
  - lytes, Cr, BUN --> to assess renal function
- ❑ urinalysis
  - routine and microscopic (WBCs, RBCs, crystals)
  - culture and sensitivity
- ❑ KUB x-ray
  - 90% of stones are radiopaque
- ❑ spiral CT
  - no contrast, good to distinguish radiolucent stone from soft tissue filling defect
- ❑ abdominal ultrasound
  - may demonstrate stone (difficult in ureter)
  - may demonstrate hydronephrosis
- ❑ IVP (see Colour Atlas M2)
  - establishes diagnosis
  - demonstrates
    - anatomy of urine collecting system
    - degree of obstruction
    - extravasation if present
    - renal tubular ectasia (medullary sponge kidney)
    - uric acid stones --> filling defect
- ❑ strain all urine --> stone analysis
- ❑ later (metabolic studies for recurrent stone formers)
  - serum lytes, calcium, phosphate and uric acid
  - PTH if hypercalcemic
  - creatinine and urea
    - 24 hour urine x 2 for creatinine,  $\text{Ca}^{2+}$ ,  $\text{PO}_4$ , uric acid, magnesium, oxalate and citrate

#### Acute Management

- ❑ medical
  - analgesic (Tylenol #3, demerol, morphine) +/- antiemetic
  - NSAIDs help lower intra-ureteral pressure
  - +/- antibiotics for UTI
  - IV fluids if vomiting
- ❑ indications for admission to hospital
  - severe persistent pain uncontrolled by oral analgesics
  - fever --> infection
  - high grade obstruction
  - single kidney with ureteral obstruction
  - bilateral ureteral stones
  - persistent vomiting
- ❑ surgical
  - ureteric stent
    - high grade obstruction
    - single kidney

- ❑ radiological
  - percutaneous nephrostomy (alternative to stent)

#### Elective Management

- ❑ medical
  - conservative if stone < 5mm and no complications
  - alkalinization of uric acid and cystine stones may be attempted (potassium citrate)
  - patient must receive one month of therapy before being considered to have failed
- ❑ surgical
  - kidney
    - extracorporeal shock wave lithotripsy (ESWL) if stone < 2.5 cm
    - + stent if 1.5-2.5 cm
    - percutaneous nephrolithotomy
      - stone > 2.5 cm
      - staghorn
      - UPJ obstruction
      - calyceal diverticulum
      - cystine stones (poorly fragmented with ESWL)
    - open nephrolithotomy
      - extensively branched staghorn
  - ureter
    - ESWL is primary modality of treatment
    - ureteroscopy
      - failed ESWL
      - ureteric stricture
      - reasonable alternative for distal 1/3 of ureter
    - open ureterolithotomy
      - rarely necessary (failed ESWL and ureteroscopy)

#### Stone Pathogenesis

- ❑ factors promoting stone formation
  - stasis (hydronephrosis, congenital abnormality)
  - medullary sponge kidney
  - infection (struvite stones)
  - hypercalciuria
  - increased oxalate
  - increased uric acid
- ❑ loss of inhibitory factors
  - Mg (forms soluble complex with oxalate)
  - citrate (forms soluble complex with calcium)
  - pyrophosphate
  - glycoprotein

### STONE TYPES

#### Calcium Stones

- ❑ account for 80% of all stones
- ❑ Ca<sup>2+</sup> oxalate most common, followed by Ca<sup>2+</sup> phosphate
- ❑ description
  - grey or brown due to hemosiderin from bleeding
  - radiopaque (see Colour Atlas M1)

#### Etiology

- ❑ hypercalciuria (60-70% of patients)
  - 95% of these patients have normal serum calcium levels
  - 5-10% of people without stones have hypercalciuria
  - absorptive causes (majority of patients)
    - increased vitamin D sensitivity --> idiopathic
- ❑ hypercalciuric Ca<sup>2+</sup> stones
  - sarcoidosis --> ↑ production of 1,25(OH)<sub>2</sub> vit D
  - abnormal vitamin D metabolism --> ↑ 1,25 (OH)<sub>2</sub> vit D
  - excess vitamin D intake
  - increased Ca<sup>2+</sup> intake (milk alkali syndrome)
  - renal phosphate leak --> ↓ PO<sub>4</sub> --> ↑ 1,25(OH)<sub>2</sub> vitamin D --> absorptive hypercalcemia
  - treatment
    - cellulose phosphate (decrease intestinal absorption of Ca<sup>2+</sup>) or orthophosphates (inhibit vitamin D synthesis)

- resorptive cause (i.e.  $\uparrow$   $\text{Ca}^{2+}$  from bones)
- hyperparathyroidism
- neoplasms (multiple myeloma, metastases)
- Cushing's disease
- hyperthyroidism
- immobilization
- steroids
- renal leak of calcium
- distal renal tubular acidosis (RTA I)  $\rightarrow$  6.0 pH
  - +  $\downarrow$  citrate  $\rightarrow$   $\uparrow$   $\text{CaPO}_4$  stones
    - treat with  $\text{HCO}_3^-$  to  $\uparrow$  citrate
- medullary sponge kidney (tubular ectasia)
  - anatomic defect in collecting ducts; 5-20% of  $\text{Ca}^{2+}$  stone formers
- idiopathic (25-40% of patients)
  - normocalcemic
  - normocalciuric
  - may have  $\downarrow$  citrate;  $\downarrow$  Mg;  $\uparrow$  oxalate;  $\uparrow$  urine acidity; dehydration
  - treatment
    - hydrochlorothiazide (HCTZ) 25 mg PO daily  $\rightarrow$   $\downarrow$   $\text{Ca}^{2+}$  in urine
    - increase water intake
- hyperuricosuria (25% of patients with  $\text{Ca}^{2+}$  stones)
  - uric acid acts as nidus for  $\text{Ca}^{2+}$  stone formation
  - treatment
    - add allopurinol if uric acid excretion  $>$  5 mmol/day
- hypocitraturia (12% of patients)
  - associated with type I RTA or chronic thiazide use
  - treatment
    - potassium citrate
- hypercalcemia (5% of patients)
  - primary hyperparathyroidism
  - malignancy
  - sarcoidosis
  - increased vitamin D
  - hyperthyroidism
  - milk-alkali syndrome
- hyperoxaluria ( $<$  5% of patients)
  - inflammatory bowel disease (IBD)
  - short bowel syndrome
  - dietary increase (caffeine, potatoes, rhubarb, chocolate, vitamin C)
  - primary increase in endogenous production
  - treatment
    - increase water intake, avoid oxalate-containing foods
    - oral calcium or cholestyramine

} 90% of cases

## Struvite Stones

- etiology and pathogenesis
  - account for 10% of all stones
  - contribute to formation of staghorn calculi
  - consist of triple phosphate (calcium, magnesium, ammonium)
  - due to infection with urea splitting organisms
    - $\text{NH}_2\text{CONH}_2 + \text{H}_2\text{O} \rightarrow 2\text{NH}_3 + \text{CO}_2$
    - $\text{NH}_4$  alkalinizes urine, thus decreasing solubility
- common organisms
  - *Proteus*
  - *Klebsiella*
  - *Pseudomonas*
  - *Providencia*
  - *S. aureus*
  - not *E. coli*
- treatment
  - complete stone clearance (ESWL/percutaneous nephrolithotomy)
  - acidify urine, dissolve microscopic fragments
  - antibiotics for 6 weeks
  - follow up urine cultures

## Uric Acid Stones

- account for 10% of all stones
- description and diagnosis
  - orange coloured gravel, needle shaped crystals
  - radiolucent on x-ray

- filling defect on IVP
- radiopaque on CT scan
- visualized with ultrasound
- etiology
  - hyperuricosuria (urine pH < 5.5)
    - secondary to increased uric acid production, or drugs (ASA and probenecid)
  - hyperuricemia
    - gout
    - myeloproliferative disease
    - cytotoxic drugs
    - defect in tubular NH<sub>3</sub> synthesis (ammonia trap for H<sup>+</sup>)
  - dehydration, IBD, colostomy and ileostomy
- treatment
  - increase fluid intake
  - NaHCO<sub>3</sub>
  - allopurinol
  - avoid high protein/purine diet

### Cystine Stones

- autosomal recessive defect in renal tubular transport
- seen in children
- aggressive stone disease
- description
  - yellow, hard
  - radiopaque (ground glass)
  - staghorn or multiple
  - decreased reabsorption of "COLA"
    - cystine
    - ornithine
    - lysine
    - arginine
- diagnosis
  - amino acid chromatography of urine --> see COLA in urine
- treatment
  - greatly increase water intake --> 3-4 L urine/day
  - HCO<sub>3</sub>
  - decrease dietary protein --> methionine
  - penicillamine chelators --> 2 g daily, soluble complex formed; use cautiously
  - a-mercaptopyronylglycine (MPG) --> similar action to penicillamine, less toxic
  - captopril (binds cystine)
  - irrigating solutions: N-acetylcystine (binds cystine), Tromethamine-E

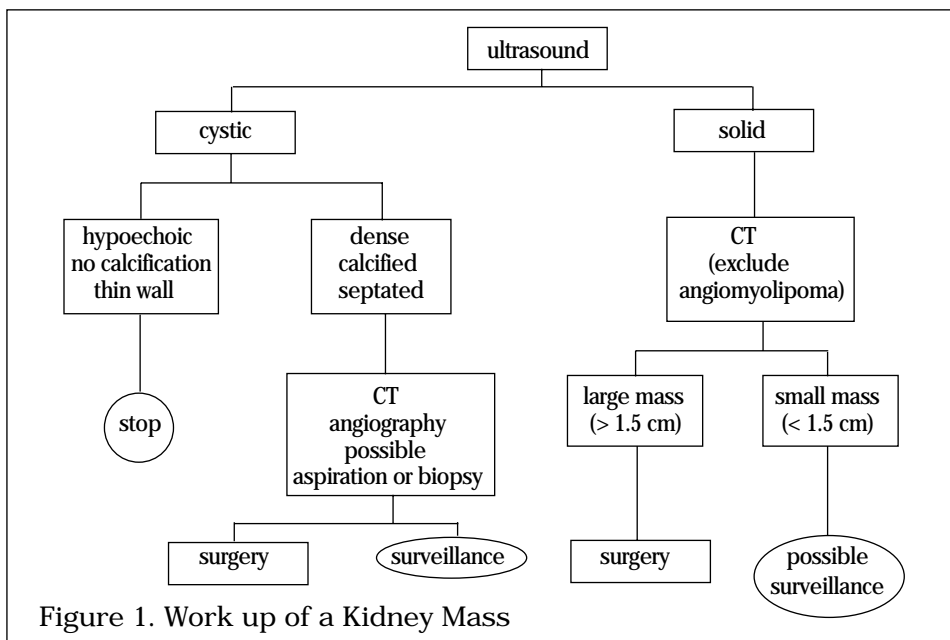


Figure 1. Work up of a Kidney Mass

## BENIGN RENAL TUMOURS

## Angiomyolipoma (Renal Hamartoma)

- rare benign tumour
- round, oval, expansible
- characterized by 3 major histologic components: blood vessels, smooth muscle and fat cells
- many asymptomatic, may spontaneously rupture, especially in pregnant females
- found in approximately 45-80% of patients with tuberous sclerosis which is characterized by
  - epilepsy
  - mental retardation
  - sebaceous adenoma
  - hamartomas of brain and kidney
- diagnose by CT --> fat (negative density on CT)  
observed in kidneys is pathognomonic for angiomyolipoma

## Renal Adenoma

- commonly found incidentally at autopsy or after nephrectomy for an unrelated disease
- 10-20% of the population
- asymptomatic
- need tissue diagnosis to definitively differentiate from renal cell carcinoma

## MALIGNANT RENAL TUMOURS

## Renal Adenocarcinoma (Renal Cell Carcinoma, RCC)

- also known as hypernephroma
- eighth most common malignancy (accounts for 3% of all newly diagnosed cancers)
- 85% of primary malignant tumours in kidney
- male:female = 3:1
- called the "internist's tumour" because of paraneoplastic symptomatology
- etiology
  - cause is unknown
  - originates from proximal convoluted tubule epithelial cell
  - risk factors: smoking (results in 2x increased relative risk), cadmium exposure, employment in leather industry
  - familial incidence seen with von Hippel Lindau syndrome which is characterized by
    - RCC (present in 2/3)
    - headache, ataxia, and blindness due to cystic lesions of cerebellum and retinal vessel aneurysms
- symptoms and signs
  - increasingly diagnosed incidentally with U/S and CT
  - poor prognostic indicators include
    - weight loss
    - weakness
    - anemia
    - bone pain
  - classic triad (too late triad!) found in 10-15%
    - gross hematuria 50%
    - flank pain < 50%
    - palpable mass < 30%
  - 30% have metastases when first seen
  - paraneoplastic syndromes
    - hematopoietic disturbances: anemia, polycythemia; raised ESR
    - endocrinopathies: hypercalcemia, production of other hormones including erythropoietin, renin, prolactin, gonadotropins, TSH, insulin, and cortisol
  - hemodynamic alterations: systolic hypertension (due to AV shunting), and peripheral edema (due to caval obstruction)
  - "Stauffer's syndrome": abnormal liver function tests, decreased WBC count, fever, areas of hepatic necrosis; reversible following removal of primary tumour

- ❑ diagnosis
  - routine labs for paraneoplastic syndromes (CBC, ESR)
  - urinalysis (60-75% have hematuria)
  - IVP
  - renal ultrasound
  - CT scan
  - angiography: no longer routinely done
- ❑ methods of spread
  - direct
  - venous
  - lymphatic
- ❑ staging
  - involves CT, chest x-ray, liver enzymes and functions, bone scan
  - T1: tumour less than 2.5 cm, confined to renal parenchyma
  - T2: tumour greater than 2.5 cm, confined to renal parenchyma
  - T3: tumour extends into major veins or adrenal, but not beyond Gerota's fascia
  - T4: tumour extends beyond Gerota's fascia
  - N0: no regional nodes
  - N1: metastasis to a single node, less than 2 cm
  - N2: metastasis to a single node between 2 and 5 cm or multiple nodes, less than 2 cm
  - N3: node greater than 5 cm
  - M0: no evidence of metastasis
  - M1: presence of distant metastasis
- ❑ treatment
  - surgical (mainstay)
    - radical nephrectomy
    - en bloc removal of kidney, tumour, ipsilateral adrenal gland and intact Gerota's capsule and periaortic lymphadenectomy
    - surgical removal of solitary metastasis may be considered
  - radiation for palliation
    - for painful bony lesions
  - chemotherapy: NOT effective
  - immunotherapy: experimental
- ❑ prognosis
  - stage at diagnosis is the single most important predictor of survival
  - 5 year survival of T1 is 90-100%
  - 5 year survival of T2-T3 is approximately 60%
  - 5 year survival of patients presenting with metastasis is 0-20%

## CARCINOMA OF THE RENAL PELVIS AND URETER

- ❑ incidence
  - rare, accounts for 4% of all urothelial cancers
  - frequently multifocal
  - papillary transitional cell cancer 85%
  - male:female = 3:1
- ❑ relative incidence
  - bladder:pelvis:ureter = 100:10:1
- ❑ predisposing factors
  - chemical exposure (industrial dyes and solvents)
  - smoking
  - analgesic abuse (acetaminophen, aspirin, and phenacetin)
  - Balkan nephropathy
- ❑ symptoms and signs
  - gross painless hematuria (70-90% of patients)
  - microscopic hematuria found incidentally
  - flank pain
  - tenderness over kidney
  - flank mass caused by either tumour or associated hydronephrosis (10-20% of patients)
- ❑ diagnosis
  - made by noting a radiolucent filling defect on IVP
  - differential diagnosis of filling defect
    - transitional cell carcinoma (differentiate via cytology and CT scan)
    - uric acid stone (differentiate via cytology and CT scan)
    - blood clot
    - pyelitis cystica
    - papillary necrosis
    - fungus ball
    - gas bubble from gas producing organisms



- ❑ treatment
  - radical ureteronephrectomy with cuff of bladder

### RENAL TRAUMA

- ❑ etiology: blunt (80%, MVA, assaults, falls) vs. penetrating (20%, stab and gunshots)
- ❑ history: mechanism of injury
- ❑ P/E: ABCs, renal vascular injury --> shock
  - flank contusions, lower rib/vertebral #, upper abdominal/flank tenderness suggest blunt trauma
- ❑ U/A: hematuria, (> 5 RBC/HPF), degree of hematuria does not correlate with the degree of injury
- ❑ imaging: IVP, CT if patient stable --> look for renal laceration, urinary extravasation, retroperitoneal hematoma, and associated intra-abdominal organ injury
- ❑ classification according to severity
  - minor: contusions and superficial lacerations, 90% of all blunt traumas, surgical exploration seldom necessary
  - major: laceration that extends into deep medulla and collecting system, injuries to renal artery/vein and segmental branches
- ❑ management
  - microscopic hematuria + isolated well-staged minor injuries do not need hospitalization
  - gross hematuria + contusion/minor lacerations: hospitalize, bedrest, repeat CT if bleeding persists
- ❑ surgical management
  - absolute indications: hemorrhage and hemodynamic instability
  - relative indications
    - nonviable tissue and major laceration
    - urinary extravasation
    - vascular injury
    - incomplete staging
    - laparotomy for associated injury
- ❑ outcome
  - F/U with IVP or CT before discharge, and at 6 weeks
  - hypertension in 5% of renal trauma

## BLADDER

### BLADDER CARCINOMA

- ❑ incidence
  - male:female = 3:1
  - usually > 55 years
  - may be characterized by frequent recurrences
- ❑ classification
  - transitional cell carcinoma (TCC) 92%
  - squamous cell carcinoma (SCC) 7%
  - adenocarcinoma 1%
  - others < 1%
- ❑ stages of transitional cell carcinoma at diagnosis
  - superficial papillary (75%)
    - 15% of these will progress to invasive TCC
    - the majority of these patients will have recurrence
  - invasive (25%)
    - 85% have no prior history of superficial TCC
    - 50% have occult metastases at diagnosis
  - carcinoma in situ
    - may progress to invasive TCC
- ❑ etiology
  - smoking (main factor)
  - chemicals
    - naphthylamines, benzidine, tryptophan metabolites
  - cyclophosphamide
  - phenacetin metabolites
  - *Schistosoma hematobium* (associated with SCC)
  - chronic irritation (cystitis)

- ❑ symptoms
  - hematuria 85-90%
  - pain 50%
  - clot retention 17%
  - no symptoms 20%
  - occult hematuria
  - irritative urinary symptoms - consider carcinoma in situ
- ❑ investigation
  - history and physical
  - urinalysis
  - urine cytology
  - ultrasound
  - cystoscopy with bladder washings (gold standard)
  - new advances with specific bladder tumour markers (NMP-22, BTA, Immunocyt, FDP)
  - intravenous pyelogram (IVP)
  - for invasive disease
  - CT, chest x-ray, liver tests
- ❑ TNM classification (see Figure 2)
  - Ta noninvasive papillary carcinoma
  - Tis carcinoma in situ; flat tumour
  - T1 tumour invades subepithelial connective tissue
  - T2a tumour invades superficial muscle
  - T2b tumour invades deep muscle
  - T3 tumour invades perivesical fat
  - T4a adjacent organ involvement; prostate, uterus or vagina
  - T4b adjacent organ involvement; pelvic wall or abdominal wall
  - N, M status: as for renal cell carcinoma
- ❑ treatment
  - carcinoma in situ (CIS)
    - TURBT
    - intravesical immunotherapy with BCG, thiotepa, or mitomycin C
  - superficial disease (Tis, Ta, T1)
    - TURBT + intravesical chemotherapy or intravesical BCG prophylaxis to decrease recurrence rate
  - invasive disease (T2a, T2b, T3)
  - radical cystectomy with urinary diversion and/or irradiation
  - metastatic disease (T4a, T4b, N+, M+)
    - irradiation +/- systemic chemotherapy

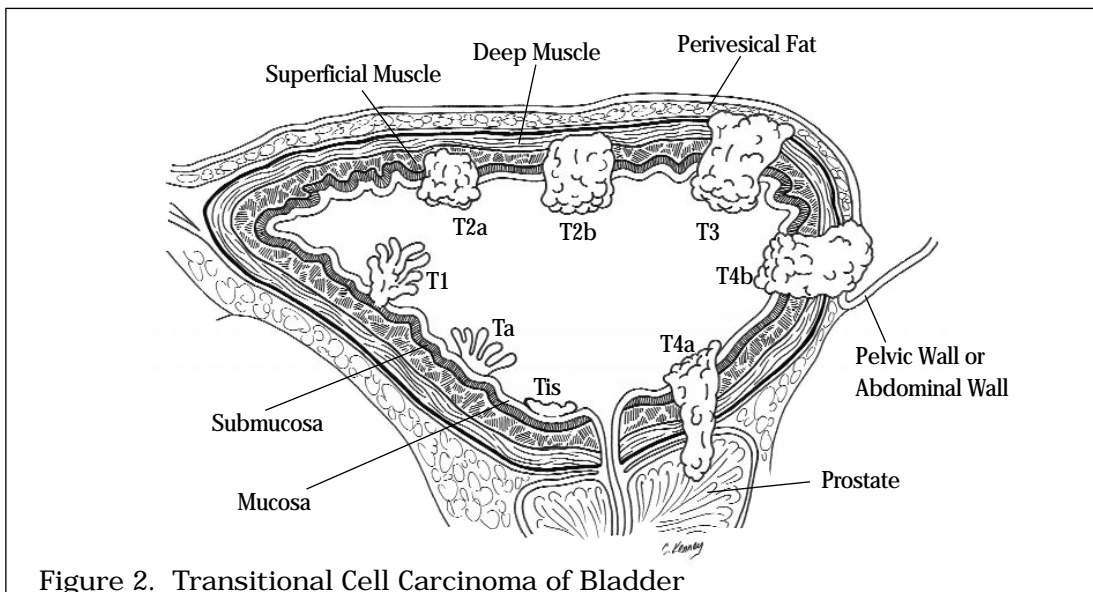


Figure 2. Transitional Cell Carcinoma of Bladder

*Drawing by Christine Kenney*

## NEUROGENIC BLADDER

## Definition

- ❑ a bladder deficient in some aspect of its innervation
- ❑ lesion above brainstem (pontine micturition centre)
  - > hyperactive bladder
- ❑ lesion at brainstem or below but above S2-S4
  - > detrusor sphincter dyssynergia
- ❑ lesion at or below sacral cord S2-S4 --> bladder areflexia

## Higher Micturition Control

- ❑ cerebellum, basal ganglia, thalamus, hypothalamus, and cerebral cortex --> all have input at pontine micturition centre
- ❑ cerebral cortex
  - superomedial portion of the frontal lobes and the genu of the corpus callosum
  - net effect is inhibitory
  - damage results in hyperactivity of detrusor
- ❑ basal ganglia
  - inhibits bladder overactivity
  - correlates with detrusor hyperactivity in Parkinson's
- ❑ cerebellum
  - receives sensory input from bladder and pelvic floor
  - maintenance of tone in the pelvic floor striated muscle
  - coordinates emptying
  - damage results in hyperactivity of detrusor
- ❑ brain stem (pontine micturition centre)
  - coordinates micturition reflex: detrusor contraction with simultaneous sphincter relaxation
  - destruction of it or below i.e. spinal cord injury --> hyperactive bladder and sphincter (external)
  - injury above it --> hyperactive bladder only

## Switch Theory of Micturition

- ❑ switch from storage to micturition increases with higher intensity of afferent input (stretching of bladder wall, increased intravesical pressure) (see Figure 3)
- ❑ urine storage
  - low level of afferent activity causes
    - efferent input via somatic pudendal nerve (S2-S4) to urethral sphincter --> contraction
    - efferent input via hypogastric nerve (sympathetics T11-L1) to bladder neck --> contraction
    - inhibition of detrusor muscle via hypogastric nerve --> relaxation
- ❑ micturition
  - high level of afferent activity causes
    - signal via pelvic nerve up to brain stem and cortex, initiating the voiding mechanism
  - pontine micturition centre causes:
    - inhibition of pudendal nerve --> relaxation of urethral sphincter
    - inhibition of hypogastric nerve --> relaxation of bladder neck
    - efferent input to pelvic nerve (parasympathetics) --> detrusor contraction

## Classification of Neurogenic Bladder

- ❑ failure to store
  - bladder problem - hyperreflexia
  - urethral problem - flaccid urethra
- ❑ failure to empty
  - bladder problem - areflexic bladder
  - urethral problem - hyperactive sphincter

## Investigations

- ❑ urodynamic studies
  - measure pressures during filling and emptying of bladder

- also measure flow rates
- EMG is incorporated
- ❑ imaging
  - IVP, ultrasound --> rule out hydronephrosis and stones
- ❑ urine C&S and microscopy
- ❑ cystoscopy --> to assess lower urinary tract

### Treatment

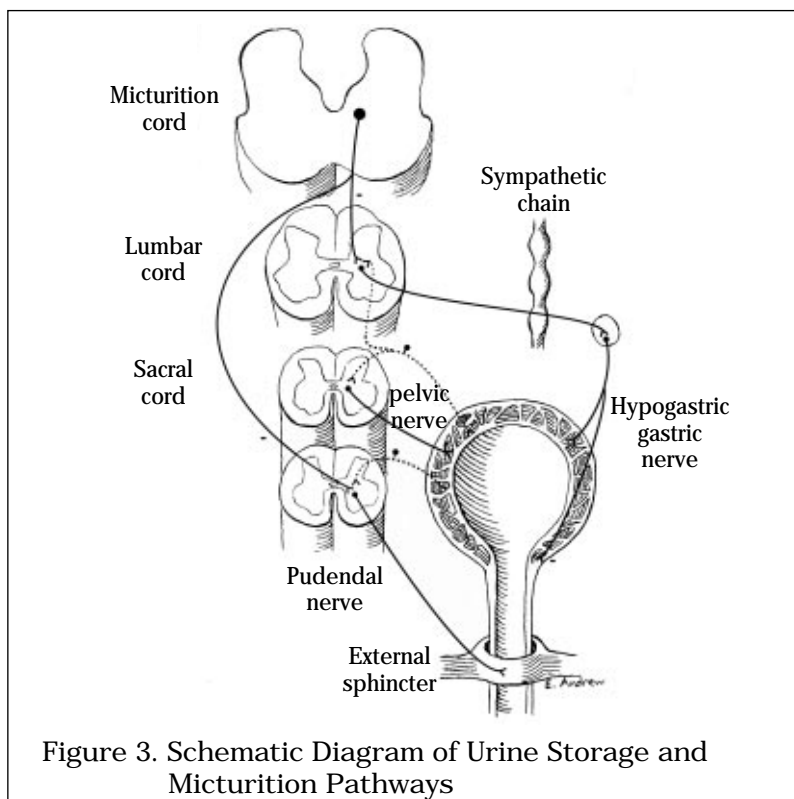
- ❑ goals of treatment (in order of importance)
  - prevent renal failure
  - prevent infections
  - prevent incontinence
- ❑ treatment options: depends on status of bladder and urethra
  - bladder hyperactivity --> medications to relax bladder
  - flaccid bladder --> intermittent catheterization

### Detrusor External Sphincter Dyssynergia

- ❑ contraction of bladder and external sphincter at the same time
- ❑ caused by injury between brainstem and sacral cord

### Autonomic Dysreflexia

- ❑ syndrome of exaggerated sympathetic activity in response to a noxious stimuli (distended bladder in a para or quadriplegic) below the lesion which is usually above T6, 7
- ❑ hypertension, reflex bradycardia, sweating and vasoconstriction below lesion
- ❑ red and vasodilated above level of lesion
- ❑ treatment: remove noxious stimulus (insert a catheter), parenteral ganglionic or  $\alpha$ -blockers or chlorpromazine



*Drawing by Eleanor Andrew*

## INCONTINENCE

### Definition

- ❑ the involuntary passage of urine sufficiently severe to cause social or hygiene problems

## Epidemiology

- ❑ most frequent in the elderly, affecting over 15% of those living in the community and 50% of nursing home residents
- ❑ F:M = 2:1

## Classification

- ❑ total: constant or periodic loss of urine without warning
  - exstrophy of bladder
  - epispadias
  - vesico-vaginal fistulas
  - ectopic ureteral orifices
- ❑ stress: urine loss with sudden increase in intra-abdominal pressure (e.g. coughing or sneezing)
  - weakness of pelvic floor musculature (child bearing, previous abdominal/pelvic surgery)
  - damage/weakness of urethra or sphincter
- ❑ urge: urine loss due to uninhibited bladder contractions
  - local bladder irritation (e.g. cystitis, stone, tumour)
  - CNS disorder
- ❑ overflow: urine loss when intravesical pressure exceeds urethral pressure
  - obstructive (e.g. BPH)
  - hypotonic bladder
  - detrusor-sphincter dyssynergia
- ❑ functional: urine loss caused by inability to reach toilet in time
  - physical immobility

## Assessment

- ❑ clinical
- ❑ urine C&S
- ❑ ultrasound
- ❑ cystoscopy
- ❑ voiding cystourethrogram (VCUG)
- ❑ cystometrogram (CMG)
- ❑ uroflowmetry

## Management

- ❑ goals
  - preserve renal function
  - maintain infection free
  - low pressure system with minimal tubes and devices
- ❑ non-medical
  - pads
  - bladder training
  - self-stimulation voiding
  - intermittent catheterization
  - indwelling catheterization
  - condom drainage
  - penile clamp
- ❑ medical
  - drugs that promote urine retention
    - smooth muscle depressant (flavoxate)
    - anticholinergics (oxybutinin, propantheline)
    - sympathomimetics (ephedrine, phenylephrine)
    - tricyclic antidepressants (imipramine)
  - drugs that promote micturition
    - cholinergic agonists (bethanechol, carbachol)
    - adrenergic antagonists (phenoxybenzamine, propranolol)
    - sphincteric relaxants (diazepam, baclofen, terazosin)
- ❑ surgical
  - TURP
  - TURBT
  - bladder or sphincter denervation
  - bladder augmentation (ileocystoplasty)
  - bladder neck reconstruction
  - artificial sphincter
  - neurostimulation
  - periurethral collagen injection
  - urinary diversion

## URINARY TRACT INFECTIONS

## Definition

- greater than 100 000 bacteria/mL - midstream urine
- if symptomatic, 100 bacteria/mL may be significant

## Classification

- first infection: first documented UTI
- unresolved bacteriuria: urinary tract is not sterilized during therapy (most commonly due to resistant organisms or noncompliance)
- bacterial persistence: urine cultures become sterile during therapy but resultant reinfection of the urine by the same organisms occur
- reinfection: new infections with new pathogens, 80% of recurrent UTIs

## Source

- ascending (commonest)
- hematogenous (TB, perinephric abscess)
- lymphatic
- direct (IBD fistulas)

## Predisposing Factors

- stasis and obstruction
  - posterior urethral valves
  - reflux
  - residual urine
  - drugs (anticholinergics)
- foreign body
  - catheter
  - stone
  - instrumentation
- decreased resistance
  - diabetes mellitus
  - malignancy
  - immunosuppression
- other factors
  - trauma
  - anatomic variance (congenital)

## History

- irritative symptoms
- obstructive symptoms
- previous UTIs
- renal calculi
- sexual activity
- personal hygiene
- hematuria
- pain
- tenderness
  - costovertebral
  - abdominal
  - rectal
- pyuria
- +/- fever, chills, nausea, vomiting
- sepsis/shock

## Organisms

- routine cultures, mnemonic "KEEPS"
  - *Klebsiella*
  - *E. coli* (90%)
  - *Enterococci*, other Gram negatives
  - *Proteus mirabilis*, *Pseudomonas*
  - *S. saprophyticus*, *S. fecalis*
- non-routine cultures
  - TB
  - *Chlamydia*
  - *Mycoplasma (Ureaplasma urealyticum)*
  - fungi (*Candida*)

## Indications for Investigations

- persistence of pyuria/symptoms after adequate therapy
- severe infection with an increase in creatinine
- hematuria
- recurrent/persistent infections
- any male
- infection in children (see Pediatric Urology Section)

## Investigations

- midstream urine R&M, C&S (routine)
- urine cytology (if indicated)
- IVP/ultrasound (if indicated)
- cystoscopy (if indicated)
- spiral CT (if indicated)
- voiding cystourethrogram (VCUG) if recurrent and/or hydronephrosis

## Treatment

- confirm diagnosis
- establish predisposing cause (if any) and correct
- identify organism and treat (TMP/SMX, norfloxacin, nitrofurantoin)
- for mild infections 3 day course is sufficient
- consider self administered antibiotics
- consider long term, low dose prophylaxis
- if febrile, consider admission with IV therapy and rule out obstruction

## RECURRENT/CHRONIC CYSTITIS

- incidence of bacteriuria in females
  - pre-teens: 1%
  - late teens: 4%
  - 30-50 years: 6%
- can be caused by perineal colonization in females
- investigations include IVP, cystoscopy, ultrasound
- relation to intercourse (postcoital antibiotics?)
- prophylaxis if greater than three or four attacks per year (long term low dose antibiotics)
- self-administered antibiotics

## INTERSTITIAL CYSTITIS

- commonly in middle-aged women
- irritative symptoms
  - frequency, nocturia, urgency, hematuria, dysuria and pain due to bladder distention
- usually no specific physical or lab findings
- small bladder capacity on cystometric studies
- rule out cancer
- diagnosis via cystoscopy and biopsy
- treatment
  - intravesical dimethylsulfoxide (DMSO)
  - intravesical steroids
  - bladder distension
  - intravesical pentosan polysulfate
  - amitriptyline
- controversial topic

## BLADDER STONES

- etiology
  - stasis (bladder outflow obstruction)
  - foreign body
- description
  - large
  - often multiple
- signs and symptoms
  - frequency and urgency
  - pain at end of urination
  - pyuria
  - hematuria
  - obstructive symptoms
- stone types
  - often Ca<sup>2+</sup> oxalate/phosphate
  - uric acid

- treatment
  - transurethral litholapaxy
  - remove outflow obstruction (TURP or dilatation of stricture)

## URINARY RETENTION

### Etiology

- outflow obstruction
  - BPH
  - prostate cancer
  - prostatitis
  - meatal or urethral stricture
  - calculus/clot at bladder neck
  - urethral disruption due to trauma
  - bladder or urethral foreign body
- loss of bladder innervation
  - disk herniation
  - spinal cord injury
  - stroke
  - DM
  - post-pelvic surgery
- pharmacological
  - major tranquilizers
  - anticholinergics
  - narcotics
  - antihypertensives (ganglionic blockers, methyldopa)

### History and Physical

- vitals
- palpable and/or percussable bladder in lower abdomen
- possible purulent/bloody meatal discharge
- DRE (size of prostate + anal tone)
- neurological: deep tendon reflexes, "anal wink", normal sensation

### Investigations

- CBC, lytes, Cr, BUN, urine R&M, C&S, ultrasound, and possibly cystoscopy

### Management

- catheterization
  - urethral anesthetic lubricant, 16-18 Fr Foley catheter (if this fails, try a coude tip catheter if patient has BPH)
  - rarely filiform and followers or percutaneous suprapubic cystostomy
  - catheterization is contraindicated in trauma patient unless urethral disruption has been ruled out
  - watch for post-obstructive diuresis after catheterization
- in post-operative patients
  - encourage ambulation
  - cholinergics to cause bladder contraction
  - alpha-blockers to relax bladder neck
- definitive treatment will depend on etiology

## BLADDER TRAUMA

- blunt (MVA, falls, and crush injury) vs. penetrating trauma to lower abdomen, pelvis, or perineum
- blunt is associated with pelvic # in 97% of cases

### History and Physical

- abdominal tenderness and distension, and unable to void
- may be few peritoneal signs or symptoms
- associated injuries such as pelvic and long bone # are common
- hemodynamic instability also common due to extensive blood loss in the pelvis

### Investigations

- U/A: gross hematuria in 95% of bladder ruptures

### Imaging

- cystogram (extravasation)



## Classification

- contusions: no urinary extravasation, damage to mucosa or muscularis
- intraperitoneal ruptures: often involve the dome
- extraperitoneal ruptures: involve anterior or lateral bladder wall

## Management

- depends on the type of bladder injury and the extent of associated injuries
- contusion: urethral catheter until hematuria completely resolves
- extraperitoneal bladder perforations can be managed non-operatively if associated injuries do not require a laparotomy and the urine is sterile at time of the injury
  - others will need surgical management
- intraperitoneal injuries require drainage and a suprapubic catheter

## Complications

- mortality is around 20%, and is usually due to associated injuries due to trauma rather than bladder rupture
- complications of bladder injury itself are rare

**PROSTATE****BENIGN PROSTATIC HYPERPLASIA (BPH)**

## Features

- extremely common (80% of 80 year olds)
- 25% of men will require treatment
- etiology unknown (testes required)
- hyperplasia in periurethral area of prostate
- symptoms not proportional to size

## Signs and Symptoms

- obstructive
- irritative
- due to residual urine
  - infection
  - bladder instability
  - acute retention
- silent prostatism
  - incontinence
  - bladder instability
- decompensated
  - secondary renal insufficiency

## Workup

- history
  - WHO Symptom Score Assessment out of 30 (can be used to follow progression of disease and response to therapy)
  - AUA (American Urology Association) Symptom Score with Quality of Life Score
  - IPSS (International Prostate Symptom Score with Quality of Life Score)
- DRE
- urinalysis
- creatinine to assess renal function
- PSA to rule out malignancy (if life expectancy > 10 years)
- cystoscopy (optional)
- uroflowmetry (optional)
- ultrasound / bladder scan to determine residual urine (optional)

## Treatment

- conservative
  - watchful waiting
  - most common
  - may include lifestyle changes e.g. evening fluid restriction, planned activities

- ❑ medical treatment
  - $\alpha$ -adrenergic antagonists to reduce prostatic smooth muscle tone (e.g. terazosin)
  - 5- $\alpha$  reductase inhibitors to reduce prostatic size if > 40 cc (e.g. finasteride)
- ❑ absolute indications for surgery
  - refractory urinary retention
  - recurrent UTIs
  - recurrent gross hematuria
  - bladder stones
  - large bladder diverticula
  - renal insufficiency
- ❑ transurethral resection of prostate (TURP) --> 95% of surgery
  - complications
    - incontinence (1%)
    - erectile dysfunction (3-35%)
    - retrograde ejaculation (25-99%)
  - in 20%, it does not relieve irritative symptoms
  - 80-90% have improvement in symptoms with increase in flow by 100%
  - approximately 5% will require reTURP within 5-10 years
- ❑ open prostatectomy --> 5% of surgery
  - for large prostates or associated problems (e.g. bladder stones)
  - suprapubic (through the bladder)
  - retropubic (through the prostatic capsule)
- ❑ surgical alternatives
  - stents, microwave therapy, laser ablation, cryotherapy
  - although available, no RCT to prove effectiveness of high intensity focused ultrasound (HIFU) and transurethral needle ablation (TUNA)

### PROSTATE SPECIFIC ANTIGEN (PSA)

- ❑ enzyme produced by epithelial cells of prostate gland to liquefy the ejaculate
- ❑ leaks into circulation but is present at < 4 ng/mL
- ❑ measured total serum PSA is a combination of free (unbound) PSA (15%) and complexed PSA (85%)
- ❑ used for
  - detection and screening
  - monitoring of cancer and relapse after treatment
  - PSA > 0.4 ng/mL after radical prostatectomy is associated with 100% chance of relapse
  - prognosis of tumour extent in general
- ❑ may also be increased by
  - prostate cancer
  - BPH
  - prostatitis
  - prostatic ischemia/infarction
  - acute urinary retention
  - prostate biopsy/surgery
  - vigorous prostatic massage (not normal DRE)
- ❑  $t_{1/2}$  = 2-3 days, must wait 2-3 weeks after transient increase to test

### PSA Screening

- ❑ recommended cutoff for positive screen: > 4.1 ng/mL
  - prevalence of cancer if PSA < 4.0 ng/mL: 1.4 %
  - prevalence of cancer if PSA > 10.0 ng/mL: 54 %
- ❑ sensitivity: 68-80%
- ❑ specificity: 49-90%
- ❑ positive predictive value: 30%
- ❑ advantages of screening
  - high sensitivity resulting in increased cancer detection rate
  - specificity exceeding that of mammography
  - detection of tumours early, when they can be treated
- ❑ disadvantages of screening
  - no demonstrated decrease in mortality rate in screened patient populations
  - unnecessary detection of slow growing tumours that would not cause problems if untreated
  - unnecessary investigations because of high false positive rate

- ❑ agencies recommending annual DRE and PSA in men over 50 with no risk factors and starting at age 40 for those with risk factors if life expectancy is > 10 years
  - American Urology Association
  - American College of Surgeons
  - American Cancer Society
- ❑ agencies recommending against screening (as of 1997)
  - US Preventive Services Task Force
  - Canadian Task Force on the Periodic Health Examination
  - Canadian Urology Association
- ❑ strategies to increase specificity of PSA test
  - age adjusted PSA reference ranges
    - different positive PSA test cutoff for different age groups
    - increase specificity by 11% but decrease sensitivity by 9%
  - PSA density
    - PSA divided by prostate volume as found on TRUS
    - > 0.15 ng/mL/g associated with increased risk of cancer
  - PSA velocity
    - PSA change divided by time - measured over a 2 year period
    - > 0.75 ng/mL/year associated with increased risk of cancer
  - percent free PSA
    - free PSA divided by total PSA in serum
    - < 20% associated with increased risk of cancer
    - because complexed PSA increases in prostate cancer decreasing the percentage of the free fraction

## PROSTATIC CARCINOMA

### Incidence

- ❑ most prevalent cancer in males
- ❑ second leading cause of male cancer deaths
- ❑ 10-30% males > 50 years
- ❑ 50% of males > 80 years
- ❑ median age at diagnosis = 70 years
- ❑ urban blacks have increased incidence

### Etiology

- ❑ not known (but requires testes as disease is not present in eunuchs)
- ❑ role of diet still being assessed
- ❑ family history
  - 1st degree relative = 2x risk
  - 1st and 2nd degree relatives = 9x risk
- ❑ vasectomy (controversial)

### Pathology

- ❑ adenocarcinoma
  - > 95%
  - often multifocal
- ❑ transitional cell carcinoma of prostate
  - associated with TCC of bladder
  - not hormone-responsive
- ❑ endometrial (rare)
  - carcinoma of the utricule

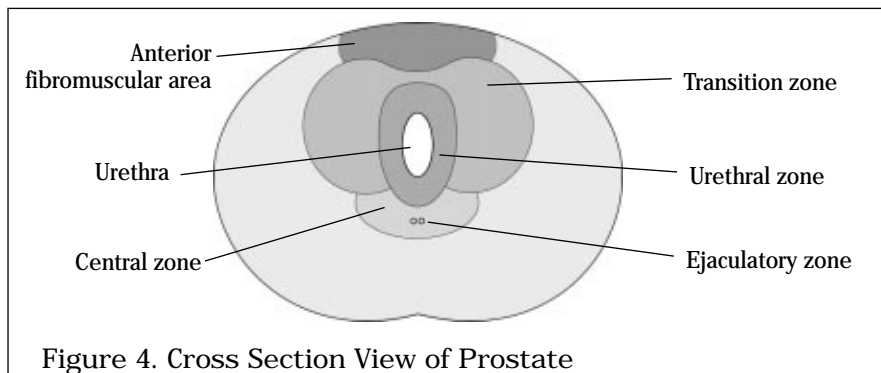


Figure 4. Cross Section View of Prostate

*Drawing by Meaghan Brierley*

## Morphology (see Figure 4)

- 75% of nodules arise in the peripheral zone
- 15-20% arise in the central zone
- 10-15% arise in the transition zone

## Methods of Spread

- local invasion
- lymphatic spread to regional nodes
  - obturator > iliac > presacral/para-aortic
- hematogenous dissemination occurs early
- bony metastasis to axial skeleton is very common (osteoblastic)
- soft tissue metastasis less common with liver and lung metastases occurring most frequently

## Signs and Symptoms

- symptoms of outflow obstruction uncommon
- suspect with prostatism and incontinence +/- back pain
- carcinoma is suspected when a hard irregular nodule or diffuse dense induration involving one or both lobes is noted
- prostate nodule has 30% chance of being malignant

## Diagnosis

- digital rectal exam (DRE)
  - differential diagnosis of a prostatic nodule
  - prostate cancer
  - benign prostatic hyperplasia
  - prostatic infarct
  - prostatic calculus
  - chronic prostatitis
  - tuberculous prostatitis
- PSA (prostate specific antigen) elevated in the majority of patients with prostate cancer (see PSA section)
- transrectal ultrasound (TRUS)
- TRUS guided needle biopsy
- incidental finding on TURP
- bone scan
- lymphangiogram and CT scanning to assess local invasion or metastases
- prostatic acid phosphatase rarely used

## Staging (TNM 1997)

- T1: clinically undetectable tumour, not palpable nor visible by imaging
- T2: confined within prostate
- T3: tumour extends through prostate capsule
- T4: tumour fixed or invades adjacent structures (besides seminal vesicles)
- N: spread to regional lymph nodes
- M: metastatic spread
- tumour grade (Gleason score out of 10) is also important
  - 2-4 represent well differentiated
  - 5-7 represent moderately differentiated
  - 8-10 represent poorly differentiated

## Treatment

- T1
  - if young consider radical prostatectomy
  - follow in older population
- T2
  - radical prostatectomy or radiation
- T3, T4
  - staging lymphadenectomy and radiation or hormonal treatment
- N > 0 or M > 0
  - requires hormonal therapy/palliative radiotherapy to metastasis
  - bilateral orchiectomy - removes 95% of testosterone
  - estrogens (e.g. DES)
    - inhibits luteinizing hormone (LH), and cytotoxic effect on tumour cells
    - increased risk of cardiovascular side effects

- LHRH agonists e.g. leuprolide (Lupron), goserelin (Zoladex)
  - initially stimulates LH, increasing testosterone and causing “flare”
  - later causing low testosterone
  - side effects include “hot flashes”
- steroidal antiandrogens e.g. cyproterone acetate
  - central effect: lowers LH
  - peripheral effect: competes with dihydrotestosterone (DHT) for cytosolic receptors
  - blocks both adrenal and testicular androgens
- non-steroidal antiandrogens e.g. flutamide
  - competes with DHT for cytosolic receptors
  - blocks both adrenal and testicular androgens
  - testosterone levels do not decrease (and may increase) so that potency may be preserved
- inhibitors of steroidogenesis e.g. ketoconazole, spironolactone
  - block multiple enzymes in the steroid pathway
- local irradiation of painful secondaries or half body irradiation

Prognosis

- Stage T1-T2: excellent, compatible with normal life expectancy
- Stage T3-T4: 40% survival at 10 years
- Stage N+ and/or M+: 40% survival at 5 years
- prognostic factors
  - tumour stage
  - tumour grade
  - PSA value

PROSTATITIS/PROSTATODYNIA

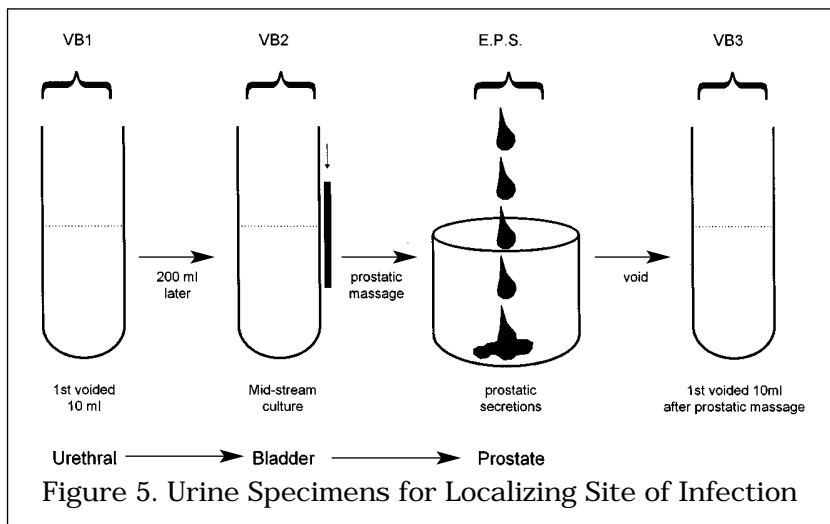
Acute Bacterial Prostatitis

- etiology
  - organisms: *E. coli*, *Pseudomonas*, *S. fecalis*
  - ascending infection in urethra (young)
  - obstructive uropathy (older age)
  - bladder calculi (older)
  - urinary catheter/instrumentation
- features
  - systemic symptoms
    - myalgia
    - fevers, chills
    - arthralgia
  - irritative symptoms
  - hematuria
  - perineal pain, worsened by defecation
- diagnosis
  - rectal exam
    - enlarged, tender, boggy prostate
    - multiple rectal exams are painful and contraindicated due to risk of secondary sepsis, abscess, or epididymo-orchitis
  - urine R&M, C&S
  - blood culture
  - rule out retention, obstruction
- treatment
  - rest/admission
  - hydration
  - PO antibiotics (TMP/SMX)
  - IV antibiotics (ampicillin and gentamycin)
  - stool softener

Chronic Bacterial Prostatitis

- the most common cause of relapsing urinary infection in males
- features
  - often asymptomatic bactiuria
  - irritative symptoms of urgency
  - low back and perineal pain
  - erectile dysfunction and incontinence

- rectal exam usually normal
- discharge uncommon
- ❑ diagnosis
  - split urines for C&S to determine site of infection; collect 4 specimens
    - initial stream (voided bladder 1 or VB1), midstream specimen VB2, then expressed prostatic secretions (EPS), and post-massage urine VB3 (see Figure 5)
  - small number of bacteria in the prostatic fluid can be evidence of disease (expressed prostatic secretions)
  - common organisms
    - *E. coli*, *Proteus*, *Klebsiella*
- ❑ treatment
  - prolonged course of antimicrobials (up to 3 months)
    - either TMP/SMX, erythromycin, or fluoroquinolone



Chronic Non-Bacterial Prostatitis

- ❑ most common of prostatic syndromes
- ❑ similar symptoms, WBC in urine but absence of bacteria in cultures
- ❑ *Chlamydia* and *Ureaplasma* may be culprits
- ❑ psychological, autoimmune
- ❑ treatment
  - tetracycline or erythromycin
  - treat both partners
  - $\alpha$ -adrenergic blocker (e.g. prazosin) to relieve sphincter spasms and symptoms
  - NSAIDs (experimental)
- ❑ other causes
  - mycotic
  - *Trichomonas*

Prostatodynia

- ❑ similar symptoms to chronic non-bacterial prostatitis
- ❑ typically young men
- ❑ possible psychosomatic component
- ❑ EPS has no WBC and no bacteria
- ❑ frequently difficult to treat

EPS Contents	Acute Bacterial	Non Bacterial	Prostatodynia
WBC	+++	++	-
bacteria	+++	-	-

Table 2. Classification of Painful vs. Painless Scrotal Swelling

Painful	Painless
1. Epididymitis	1. Hydrocele
2. Orchitis	2. Spermatocele
3. Torsion	3. Varicocele
4. Tumour (hemorrhagic)	4. Tumour (non hemorrhagic)
5. Hematocele	5. Hernia
6. Strangulated Indirect Hernia	

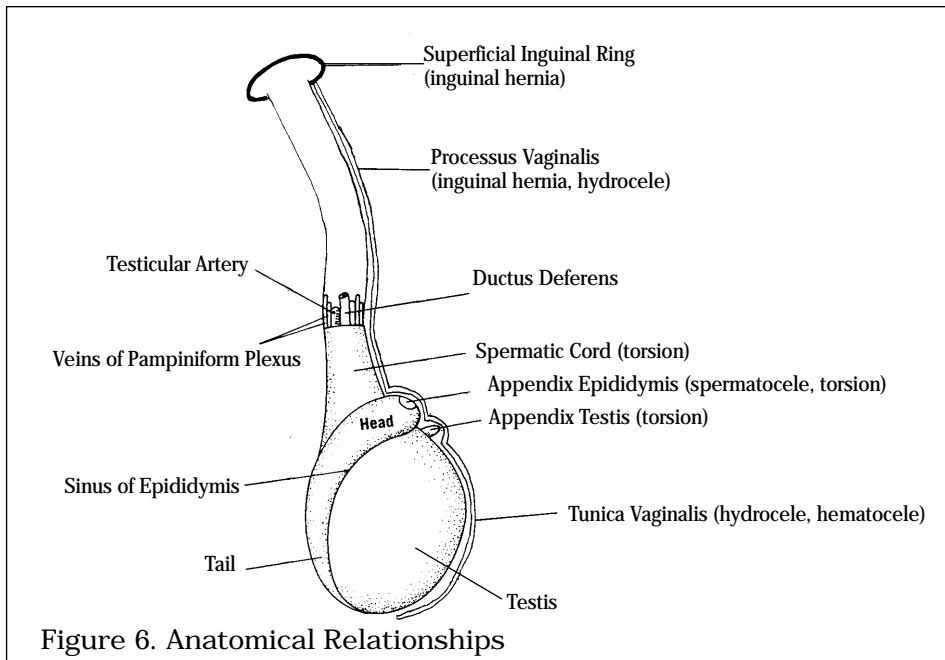


Figure 6. Anatomical Relationships

Reprinted with permission from O'Rahilly, R. *Basic Human Anatomy*. c 1983.

## EPIDIDYMITIS

### □ etiology

- infection
  - < 35 years - GC or *Chlamydia* (STD's)
  - > 35 years - coliforms (from GI tract)
- prior instrumentation
- reflux
  - physical exertion (heavy lifting) causes reflux of urine along vas deferens --> sterile epididymitis

### □ signs and symptoms

- sudden onset scrotal pain +/- radiation along cord to flank
- swelling and tenderness
- fever
- reactive hydrocele

### □ diagnosis

- urinalysis (pyuria), urine C&S
- +/- urethral discharge: Gram stain for GC
- pain may be relieved with elevation of testicle (Prehn's sign), absent in testicular torsion
- if diagnosis clinically uncertain, must do
  - colour-flow Doppler ultrasound
  - nuclear medicine scan
  - examination under anesthesia (EUA)

- ❑ treatment
  - antibiotics
    - GC or *Chlamydia* - ceftriaxone 250 mg IM once followed by doxycycline 100 mg BID x 21 days
    - coliforms - ampicillin
  - scrotal support
  - ice
  - analgesia

## ORCHITIS

- ❑ etiology
  - 25% with mumps get orchitis
  - mumps orchitis usually follows parotitis by 3-4 days
  - post-pubertal males
  - 10% bilateral
  - other rare causes
    - tuberculosis (TB)
    - syphilis
    - granulomatous (autoimmune) in elderly men
- ❑ signs and symptoms
  - fever and prostration
  - +/- hydrocele
- ❑ diagnosis
  - red, swollen scrotum
  - blue testis
  - no urinary symptoms
- ❑ treatment
  - mumps hyperimmune globulin
  - analgesics, antipyretics
  - ice
  - steroids
  - bedrest
  - scrotal elevation
- ❑ complications
  - if severe, testicular atrophy
  - 30% have persistent infertility problems

## TORSION

- ❑ two types: torsion of testicular appendage or testicular torsion

### I. Torsion of Testicular Appendages

- ❑ twisting of testicular/epididymal appendix
- ❑ often < 16 year of age
- ❑ signs and symptoms
  - clinically similar to testicular torsion
  - "blue dot sign" - blue infarcted appendage seen through scrotal skin
  - point tenderness over the superior-posterior portion of testicle
- ❑ treatment
  - analgesia - most will subside over 5-7 days
  - surgical exploration and excision if diagnosis uncertain or refractory pain

### II. Testicular Torsion

- ❑ twisting of testis on cord
- ❑ often < 30 years of age
- ❑ commonly after physical activity
- ❑ etiology
  - bell clapper congenital deformity (see Figure 7)
  - anomalous development of tunica vaginalis and spermatic cord
- ❑ signs and symptoms
  - acute onset of severe scrotal pain, swelling
  - raised and transverse testicle (horizontal lie)
  - no pain relief with testicle elevation (negative Prehn's sign)
- ❑ diagnosis
  - ultrasound with colour-flow Doppler probe over testicular artery (if torsion, no blood flow)
  - decrease uptake on 99M Tc Perchnetate Scintillation Scan
  - examination under anesthesia and surgical exploration



- treatment
  - emergency detorsion (rotate "inward"), failure of manual detorsion requires surgical detorsion, and bilateral orchiopexy (fixation)
  - < 12 hours - good prognosis
  - 12-24 hours - uncertain prognosis
  - > 24 hours - poor prognosis

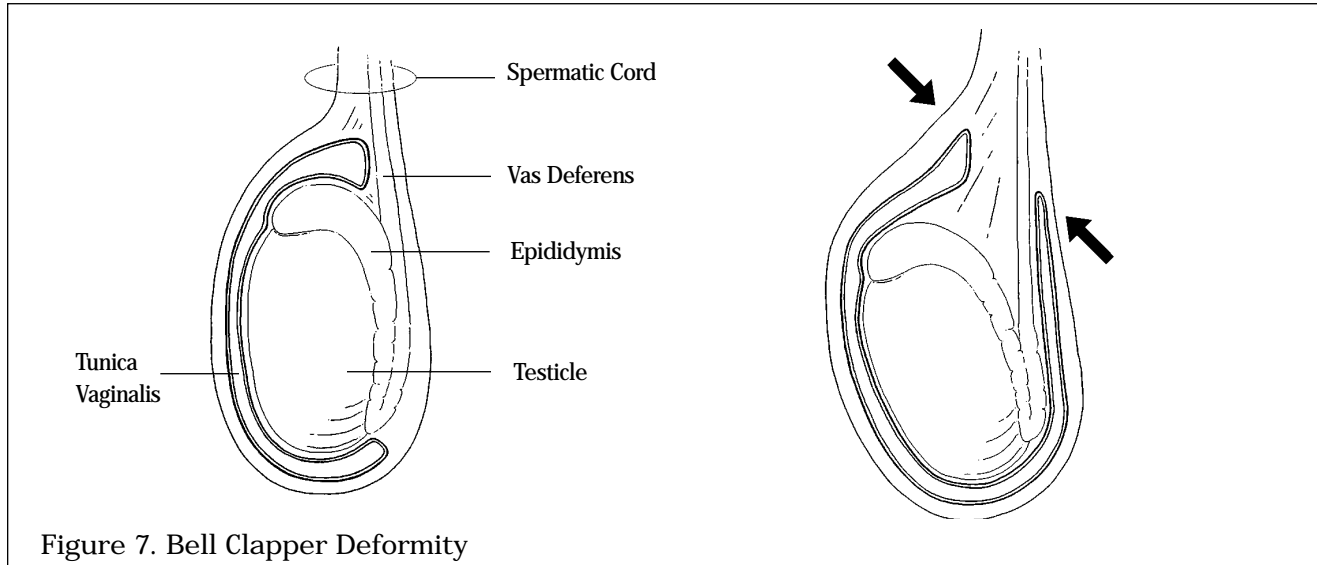


Figure 7. Bell Clapper Deformity

Drawings by Brett Clayton

Table 3. Differential Diagnosis of Torsion vs. Epididymo-orchitis

	Torsion	Epididymo-orchitis
age	most common 12-18 years	any
onset	acute	may be gradual
nausea	common	none
fever	25%	30%
pyuria	20%	50% +
scrotal elevation	no effect	decreases pain
testicular position	elevated / transverse	normal
opposite testes	bell clapper	normal
colour doppler ultrasound	absent flow to the epididymis and testes	increased flow to the epididymis and testis
nuclear imaging	central photon-deficient areas	increased perfusion of the affected testes and hemiscrotum
management	surgical	antibiotics, surgery if uncertain

## TESTICULAR TUMOURS

- 95% are malignant
- any solid testicular mass in young patient must rule out malignancy
- primary
  - 1% of all malignancies in males
  - most common solid malignancy in males aged 15-34 years
  - undescended testicle has increased risk (40 x) of malignancy
  - germinal cell 95% (all are malignant)
    - seminoma (30-50%)
    - non-seminoma (50-70%)
      - embryonal
      - choriocarcinoma
      - teratoma
      - yolk sac
      - combinations

- non-germinal cell 5% (usually benign)
  - Leydig --> testosterone, precocious puberty
  - Sertoli --> gynecomastia, decreased libido
- ❑ secondary
  - male > 50 years of age
  - usually a lymphoma
  - others include metastases from
    - lung
    - prostate
    - gastrointestinal
- ❑ signs and symptoms
  - painless swelling in the testicle
  - painful if hemorrhage into tumour, otherwise painless
  - slight heaviness
  - rubbery, hard mass
  - associated hydrocele in 10%
  - coincidental trauma in 10%
  - gynecomastia due to secretory tumour effects
  - back pain
  - supraclavicular nodes
  - abdominal mass (retroperitoneal lymph node metastases)
- ❑ investigations
  - testicular ultrasound to confirm diagnosis
  - chest x-ray
  - markers for staging (βhCG, AFP)
  - CT abdomen (after diagnosis; retroperitoneal nodes enlarged)
  - needle aspiration contraindicated
- ❑ staging
  - Stage I: disease limited to testis, epididymis, or spermatic cord
  - Stage II: disease limited to the retroperitoneal nodes
  - Stage III: disease metastatic to supradiaphragmatic nodal or visceral sites
  - methods of spread include direct extension, blood, and lymphatics (retroperitoneal)
  - “cross-over” metastases from right to left are fairly common, but they have not been reported from left to right
- ❑ tumour markers
  - βhCG and AFP are positive in 85% of non-seminomatous tumours
  - pre-orchietomy elevated marker levels return to normal post-operatively if no secondaries
  - βhCG positive in 8% of seminomas, AFP never elevated with seminoma
- ❑ treatment
  - never use a transscrotal approach for biopsy or orchietomy, due to chance of metastases via lymph drainage
  - seminoma
    - radical inguinal orchietomy and radiation (90% survival)
    - adjuvant chemotherapy for metastatic disease
  - non-seminoma
    - radical inguinal orchietomy and staging
    - retroperitoneal lymphadenectomy or surveillance
    - surveillance includes monitoring CXR, (hCG, and AFP levels
    - chemotherapy if evidence of secondary disease
- ❑ spread
  - direct
  - lymphatic
  - right --> medial, paracaval anterior and lateral nodes
  - left --> left lateral and anterior paraaortic nodes
- ❑ prognosis
  - 99% cured with Stage I, Stage II
  - 70-80% complete remission with advanced disease

### Clinical Pearl

- ❑ Surgical descent of undescended testis does not reduce the risk of malignancy (40 x)

## HEMATOCELE

- ❑ trauma with bleed into tunica vaginalis
- ❑ ultrasound helpful to exclude fracture of testis which requires surgical repair
- ❑ treatment
  - ice packs, analgesics, surgical repair

## HYDROCELE

- ❑ definition
  - collection of fluid within the processus vaginalis
  - may occur within the spermatic cord, most often seen surrounding the testis
- ❑ etiology
  - usually idiopathic, found in 5-10% of testicular tumours
  - associated with tumour, orchitis, epididymitis
- ❑ types
  - communicating hydrocele: patent processus vaginalis
  - non-communicating hydrocele: processus vaginalis is not patent
- ❑ diagnosis
  - ultrasound (definitive), especially if < 40 years of age (rule out tumour)
- ❑ treatment
  - nothing if tolerated and no complications
  - surgical
  - sclerotherapy
- ❑ complications
  - hemorrhage into hydrocele sac following trauma
  - compression of testicular blood supply

## SPERMATOCELE/EPIDIDYMAL CYST

- ❑ definition
  - collection of sperm in the appendix epididymis
  - located at superior pole of testicle
- ❑ diagnosis
  - aspirate for diagnosis, not as treatment
  - transilluminates
- ❑ treatment
  - usually no treatment
  - remove only if symptomatic

Table 4. Differentiation between Hydrocele, Spermatocele, Hernia

	Palpation	Transillumination
hydrocele	testis is not separable from hydrocele spermatic cord is palpable above the swelling	+ve
spermatocele	testis is separable from spermatocele spermatic cord is palpable above the swelling	+ve
hernia	testis is separable from hernia spermatic cord is not palpable above the swelling cough impulse may be transmitted may be reducible	-ve

## VARICOCELE

- ❑ etiology
  - dilated veins in the pampiniform plexus (97% on left side)
  - if on the right side --> suspect a retro-peritoneal tumour (renal cell carcinoma)
  - 15% incidence in young men (left sided)
  - associated with infertility
  - usually asymptomatic
- ❑ diagnosis
  - upright - mass of dilated, tortuous veins - "bag of worms"
  - heavy sensation after walking or standing
  - supine - venous distention abates
  - pulsates with Valsalva or cough
- ❑ treatment
  - surgical ligation of testicular vein above inguinal ligament
  - percutaneous vein occlusion (balloon catheter, sclerosing agents)

- in the presence of oligospermia, surgically correcting the varicocele may improve sperm count and motility in 50-75% of patients

## HERNIA

- bowel vs. omentum
- if bowel
  - direct vs. indirect inguinal hernia
  - bowel sounds often audible
  - does not transilluminate
  - may have bowel obstruction
- painful if strangulated indirect hernia
- twists and becomes ischemic
- surgical emergency
  - treatment: surgical correction (see General Surgery Notes)

## PENIS AND URETHRA

### PEYRONIE'S DISEASE

#### Etiology

- inflammatory process involving tunica albuginea of corpora cavernosa resulting in fibrotic plaque
- commonly on dorsal surface resulting in upward curvature of erect penis due to scar tissue --> pain on erection or sexual intercourse
- chordee = ventral bend
- in children, chordee associated with hypospadias (1/300)

#### Treatment

- watchful waiting
- topical vitamin E cream + aminobenzoic acid
- surgical excision of plaque, prosthesis for erectile dysfunction

### PRIAPISM

#### Definition

- painful tumescence (swelling) of corpora cavernosa with flaccid glans penis (no corpora spongiosum involvement)

#### Etiology

- 60% idiopathic
- intracorporeal drug injection (papaverine, phentolamine, PGE<sub>1</sub>)
- increased incidence with
  - sickle cell disease
  - leukemia
  - pelvic tumours
  - pelvic infections
  - penile trauma
  - spinal cord trauma

#### Treatment

- needle aspiration and drainage
- intracorporeal injection with phenylephrine (Neo-Synephrine) (a vasoconstrictor)
- cavernosal-spongiosal shunt if necessary (drain through spongiosum)
- STAT leukopheresis if leukemia

#### Complication

- erectile dysfunction due to corporal fibrosis if treatment delayed (50%)

## PHIMOSIS

### Definition

- inability to retract foreskin over glans penis
- may occur due to or be caused by balanitis (infection of glans)

### Treatment

- circumcision

## PARAPHIMOSIS

### Definition

- foreskin caught behind glans leading to edema; unable to reduce foreskin

### Treatment

- squeeze edema out of the glans with manual pressure / elastic wrap
- manual reduction with analgesia/benzodiazepine and dorsal slit
- pull on foreskin with fingers while pushing on glans with thumbs (must push very hard)
- elective circumcision for definitive treatment, as paraphimosis tends to recur

## PENILE TUMOURS

- rare

### Benign

- cyst
- hemangioma
- nevus
- papilloma

### Pre-malignant

- balanitis xerotica obliterans
- leukoplakia
- Buschke-Lowenstein tumour (large condyloma)
- Bowen's disease/CIS
- erythroplasia of Queyrat (velvet red plaques, indurated)

### Malignant

- squamous cell (most common)
- basal cell
- Paget's disease
- melanoma

## ERECTILE DYSFUNCTION

### Definition

- persistent inability to obtain or sustain an adequate erection for intercourse
- physiology
  - erection
    - veno-occlusive mechanism: relaxation and engorgement of arterioles of corpora (through release of NO by parasympathetics) and occlusion of penile venous drainage
  - emission
    - sensory from glans
    - secretions from prostate, seminal vesicles and ejaculatory ducts enter prostatic urethra (sympathetics)
  - ejaculation
    - spasmodic contraction of peri-urethral and pelvic floor musculature (sympathetics and somatics --> urethral bulb)

### Classification

- psychogenic (50%)
  - ethanol, tension, and/or premature ejaculation often involved

- patient usually characterized by
  - younger age
  - intermittent difficulty
  - no risk factors for organic disease
  - nocturnal penile tumescence present
  - often able to achieve erection using self-stimulation
- ❑ organic (50%)
  - endocrine: diabetes (20%), gonadal dysfunction, pituitary
  - vasculogenic (12%)
  - neurogenic: multiple sclerosis
  - iatrogenic: drugs (antihypertensives, sedatives), radiation, surgery (radical prostatectomy)
  - penile: post-priapism, Peyronie's
  - patient usually characterized by
    - older age (> 50 years old)
    - constant difficulty
    - risk factors present (atherosclerosis, HTN, DM)
    - nocturnal penile tumescence absent (Rigiscan)

#### Investigations

- ❑ history, physical
- ❑ testosterone (free and total), prolactin, LH, FSH
- ❑ glucose, cholesterol
- ❑ non-invasive
  - nocturnal penile tumescence monitor
  - doppler studies, penile-brachial index < 0.6 suggestive of vascular cause
- ❑ invasive
  - penile injections
  - angiography (pudendal artery)
  - cavernography

#### Treatment

- ❑ psychological (sex counselling)
- ❑ oral medication
  - sildenafil (Viagra): cyclic guanosine monophosphate inhibitor
  - yohimbine: alpha-blocker
  - trazodone: serotonin antagonist and reuptake inhibitor
- ❑ intracorporal vasodilator injection/self-injection
  - triple therapy (papaverine, phentolamine, PGE<sub>1</sub>) or PGE<sub>1</sub> alone
  - complications
    - priapism (overdose)
    - thickening of tunica albuginea at site of repeated injections (Peyronie's plaque)
    - hematoma
- ❑ vacuum devices: draw blood into penis, then put ring on base of penis
- ❑ implants: malleable, inflatable - last resort
- ❑ arterial revascularization of penis - limited success

#### URETHRAL SYNDROME

- ❑ dysuria in females with consistently sterile urine cultures or low bacterial counts
- ❑ some have bacterial urethrocystitis (*C. trachomatis* or other organisms) and require antimicrobial treatment
- ❑ treatment
  - tetracycline or erythromycin
- ❑ rule out psychological, vaginitis, cancer

#### URETHRAL STRICTURE

- ❑ refers only to anterior urethral scarring (posterior strictures not included)
- ❑ involves scar in corpus spongiosum
- ❑ contraction of this scar will decrease size of urethral lumen
- ❑ more common in males

#### Etiology

- ❑ congenital
  - may cause hydronephrosis
  - treat at time of endoscopy with dilatation, internal urethrotomy

- ❑ trauma
  - instrumentation (most common, at fossa navicularis)
  - external trauma
    - urethral trauma with stricture formation
- ❑ infection
  - common with gonorrhea in the past (not common now)
  - long-term indwelling catheter
  - balanitis xerotica obliterans
    - causes meatal stenosis

#### Diagnosis and Evaluation

- ❑ signs and symptoms
  - decreased force/amount of urinary stream
  - spraying
  - double stream
  - post-void dribbling
  - other UTIs (prostatitis, epididymitis)
- ❑ laboratory findings
  - flow rates < 10 mL/s (normal = 20 mL/s)
  - urine culture usually negative, but may show pyuria
- ❑ radiologic findings
  - urethrogram, VCUG, or ultrasound will demonstrate location
- ❑ urethroscopy

#### Treatment

- ❑ dilatation
  - temporarily increases lumen size by breaking up scar tissue
  - healing will reform scar tissue and recreate stricture
  - not usually curative
- ❑ internal urethrotomy
  - endoscopically incise stricture without skin incision
  - cure rate 70-80% with single treatment, 90% with repeated courses
- ❑ open surgical reconstruction
  - completely excise strictures < 2 cm, extending 1 cm beyond each end
  - patch graft urethroplasty if > 2 cm
  - full-thickness skin graft obtained from penis to replace urethra

## URETHRAL TRAUMA

#### Etiology

- ❑ most common site is membranous urethra due to blunt trauma, MVAs
  - associated with pelvic fractures (10% of such fractures)
- ❑ other causes: iatrogenic instrumentation, prosthesis insertion, penile fracture, masturbation with urethral manipulation

#### Diagnosis

- ❑ do not perform cystoscopy or catheterization before retrograde urethrography if urethral trauma suspected
- ❑ signs and symptoms
  - high riding prostate
  - blood at urethral meatus
  - sensation of voiding without urine output
  - swelling and hematoma
- ❑ retrograde urethrography
  - demonstrates extravasation and location of injury

#### Treatment

- ❑ simple contusions - no treatment
- ❑ partial urethral disruption
  - with no resistance to catheterization - Foley x 2-3 weeks
  - with resistance to catheterization
    - suprapubic cystostomy
    - periodic flow rates/urethrograms to evaluate for stricture formation
- ❑ complete disruption
  - immediate repair if patient stable, delayed repair if unstable

## Classification

- pseudo vs. true
- by age (see Table 5)
- by location: pre-renal, renal, ureter, bladder, urethra
- gross vs. microscopic (see Nephrology Notes)
  - up to 2-3 RBC/HPF is normal
  - microscopic hematuria normal in 10% of population

Table 5. Etiology of Hematuria by Age Group

Age (years)	Etiology (in order of decreasing frequency)
0-20	glomerulonephritis, UTI, congenital anomalies
20-40	UTI, stones, bladder tumour
40-60	male: bladder tumour, stones, UTI female: UTI, stones, bladder tumour
>60	male: BPH, bladder tumour, UTI female: bladder tumour, UTI

## Etiology

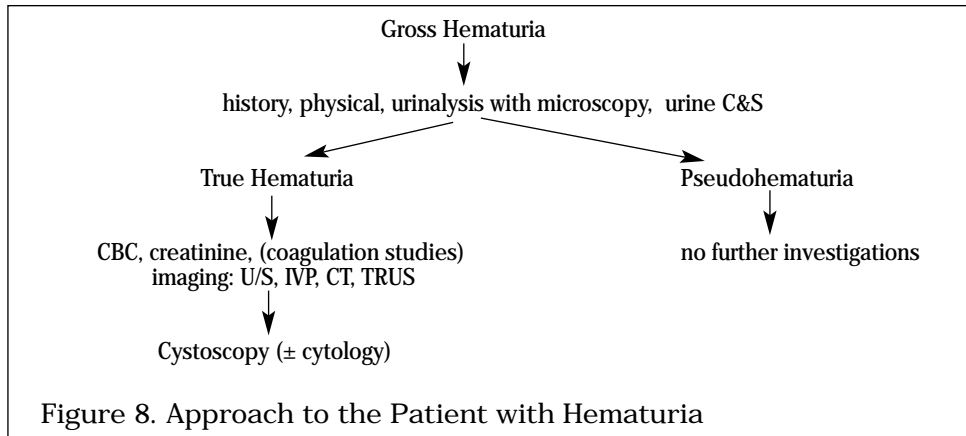
- pseudohematuria
  - menses
  - dyes
    - beets
    - rhodamine B in drinks, candy and juices
  - hemoglobin
    - hemolytic anemia
  - myoglobin
    - rhabdomyolysis
  - porphyria
  - laxatives
    - phenolphthalein
- based on age (see Table 5)
- based on source of bleeding
  - pre-renal
    - throughout urinary stream
    - anticoagulants
    - coagulation defects
    - sickle cell disease
    - leukemia
  - renal
    - throughout urinary stream
    - renal cell carcinoma, transitional cell carcinoma, Wilm's tumour, pyelonephritis, tuberculosis, glomerulonephritis, trauma, stone, infarct, polycystic kidneys, arteriovenous malformation
  - ureter
    - stone, tumour
  - bladder
    - cystitis, tumour, stone, polyps
  - urethra
    - urethritis, stone, tumour, urethral stricture
- timing to urinary stream
  - initial
    - anterior urethral lesions
  - terminal
    - bladder neck/trigone
  - total
    - bladder and/or above bladder

## Diagnosis

- approach (see Figure 8)
- history
  - timing to urinary stream
  - flank pain
  - provoking factors (e.g. exercise, trauma)



- irritative or obstructive symptoms
- previous kidney or urologic disease
- history of recent UTI, STDs, TB exposure, pelvic irradiation, bleeding diathesis, smoking
- drugs (NSAIDs, anticoagulants)
- physical exam
  - fluid status
  - blood pressure
  - abdominal masses or pain
  - DRE for prostate
  - external genitalia in males



### Investigations

- urinalysis: (the 4 C's)
  - casts vs. RBCs
  - crystals
  - culture and sensitivity
  - cytology
- ultrasound
- cystoscopy
- intravenous pyelogram (IVP)

### Clinical Pearl

- In patients with hematuria, particularly if over age 40, malignancy must be ruled out, especially bladder tumours

## INFERTILITY

- failure to conceive after one year of unprotected, properly timed intercourse
- primary vs. secondary (see Gynecology Notes)
- incidence
  - 10% of all couples - investigate both partners
  - 1/3 female, 1/3 male, 1/3 combined problem

### Male Reproduction

- hypothalamus-pituitary axis
- LH --> Leydig cells --> testosterone --> sperm support
- FSH --> Sertoli cells --> sperm support
- sperm --> epididymis --> vas deferens --> ejaculatory ducts
- emission
  - semen 2/3 from seminal vesicle
- ejaculation
  - bladder neck closure (sympathetic control)
  - rhythmic contraction of pelvic floor and bulbo-cavernosus muscle

Etiology

- ❑ hormonal (see Endocrinology Notes)
  - hypothalamic-pituitary-testicular axis (2-3%)
- ❑ scrotal
  - varicocele
  - common (40% of men with infertility have it)
  - rule out with scrotal ultrasound
  - 50-75% of men have improved semen quality after varicocelectomy
  - tubal obstruction (congenital, chronic infection)
- ❑ testicular
  - tumour
  - congenital (Klinefelter's - XXY, undescended testes)
  - post infectious (epididymo-orchitis, STDs)
- ❑ iatrogenic
  - radiation, antineoplastic and antiandrogen drugs can interfere with sperm transport and production
- ❑ surgical complications
  - testes (vasectomy, hydrocelectomy)
  - inguinal (inadvertant ligation of vas deferens)
  - bladder/prostate (damage to bladder neck causing retrograde ejaculation, damage to ejaculatory ducts)
  - abdomen (damage to sympathetic nerves causing retrograde ejaculation)
- ❑ transport
  - cystic fibrosis, Kartagener's syndrome
  - absence of vas deferens, obstruction of vas deferens
- ❑ investigations
  - normal semen analysis (at least 2 specimens)
    - volume: 2-5 mL
    - count: > 20 million sperm/mL
    - morphology: > 60% normal forms
    - motility: > 50%
    - liquefaction: complete in 20 minutes
    - pH: 7.2-7.8
    - WBC: < 10 per high power field
  - hormonal evaluation
  - chromosomal studies
  - immunologic studies (sperm antibodies in ejaculate and blood)
  - testicular biopsy
  - scrotal U/S (varicocele, testicular size)
  - vasography (assess patency of vas deferens)
- ❑ treatment
  - medical
    - endocrine therapy (see Endocrinology Notes)
    - therapy for retrograde ejaculation: discontinue anti-sympathomimetic agents, may start alpha-adrenergic stimulation (phenylpropanolamine, pseudoephedrine, or ephedrine)
    - treat underlying infections
  - surgical
    - varicocelectomy
    - vasovasostomy, epididymovasostomy, transurethral resection of ejaculatory duct
    - microsurgical epididymal sperm aspiration (MESA) (for vas deferens compromise by creating an artificial sperm reservoir over epididymis)

**CONGENITAL ABNORMALITIES**

- not uncommon; 1/200 have congenital abnormalities of the GU tract
- UTI is the most common presentation postnatally
- hydronephrosis is the most common finding antenatally

**HYPOSPADIAS**

- very common; 1/300
- a condition in which the urethral meatus opens on the ventral side of the penis, proximal to the glans penis
- classified as
  - glandular
  - coronal
  - penile
  - penoscrotal
  - perineal
- may be associated with chordee, intersex states, or undescended testicles
- depending on the severity, there may be difficulty directing the urinary stream or even infertility
- treatment is surgical correction
- circumcision should be deferred because the foreskin may be utilized in the correction

**EPISPADIAS-EXSTROPHY COMPLEX**

- incidence 1/30 000
- represents failure of closure of the cloacal membrane, resulting in the bladder and urethra opening directly through the abdominal wall
- high morbidity --> incontinence and infertility
- treatment: surgical correction

**ANTENATAL HYDRONEPHROSIS**

- 1% of fetuses
- unilateral or bilateral
- differential diagnosis:
  - UPJ obstruction
  - multi-cystic kidney
  - reflux
  - PUV
  - duplex anomalies
- antenatal in utero intervention rarely indicated

**POSTERIOR URETHRAL VALVES**

- the most common obstructive urethral lesion in infants
- abnormal mucosal folds at the distal prostatic urethra
- presents with obstructive symptoms, UTI, or complications of obstruction (depending on the severity)
- now detected antenatally --> hydronephrosis
- diagnosis: VCUG
- treatment: cystoscopic destruction of the valves

**UPJ OBSTRUCTION**

- the most common congenital defect of the ureter
- unclear etiology: ? adynamic segment of ureter
- symptoms depend on severity and age of diagnosis
  - infants: abdominal mass
  - children: pain, vomiting
- diagnosis: U/S, renal scan +/- furosemide
- treatment: surgical correction
- prognosis: good, usually unilateral disease

**VESICoureTERAL REFLUX (VUR)**

- condition wherein urine passes retrograde from the bladder through the UVJ into the ureter
- common causes: trigonal weakness, lateral insertion of the ureters, short submucosal segment (all part of "primary reflux")
- many other causes including secondary reflux, subvesical obstruction, iatrogenic, secondary to ureteric abnormalities (e.g. ureterocele, ectopic ureter, or duplication), and secondary to cystitis

- ❑ symptoms
  - UTI, urosepsis
  - pyelonephritis
  - pain on voiding
  - symptoms of renal failure (uremia, hypertension)
- ❑ diagnosis and staging is done using VCUG, +/- U/S
- ❑ grading based on cystogram
  - grade I: ureters only fill
  - grade II: ureters and pelvis fill
  - grade III: ureters and pelvis fill with some dilatation
  - grade IV: ureters pelvis and calyces fill with significant dilatation
  - grade V: ureters, pelvis and calyces fill with major dilatation and tortuosity
- ❑ complications
  - pyelonephritis
  - hydrouréteronephrosis
- ❑ management
  - many children “outgrow” reflux (60% of primary reflux)
  - treatment is dependent on the grade
  - minimal reflux: keep urine free of infection,
  - observe with repeat VCUG, U/A, and monitor renal function
  - moderate reflux: trial of conservative therapy
  - medical
    - antibiotics
  - surgical (ureteroneocystostomy +/- ureteroplasty)
    - indications
      - failure of observation
      - new renal scars
      - breakthrough infections
      - severe reflux
- ❑ prognosis depends on degree of damage done at the time of diagnosis

### URINARY TRACT INFECTION

- ❑ 50% of children with UTI will have a GU abnormality
- ❑ indications for investigation of UTI
  - < 1 year old with symptomatic UTI
  - all males, regardless of age
  - all febrile UTIs with significant systemic symptoms
- ❑ investigations
  - clean catch urine for urine R&M, C&S
  - assessment of renal function (creatinine)
  - U/S abdomen
  - VCUG
- ❑ treatment is of the underlying cause if any, then treat the UTI as per adults

#### Clinical Pearl

- ❑ Diagnosis of UTI in children requires both:
  - (a) positive culture and
  - (b) pyuria

### DAYTIME INCONTINENCE

- ❑ age and culture related
- ❑ commonest cause: maturational lag
- ❑ treatment
  - rule out organic causes, explore psychological/psychiatric causes
  - positive reinforcement for dry days
  - bladder training
  - pharmacotherapy: imipramine, DDAVP (anti-diuretic hormone), oxybutynin (Ditropan)

### NEPHROBLASTOMA (WILM'S TUMOUR)

- ❑ arises from metanephric blastema
- ❑ 5% of all childhood cancers, 5% bilateral
- ❑ average age of incidence is 3 years
- ❑ 1/3 hereditary and 2/3 sporadic
  - familial form associated with other congenital abnormalities and gene defects

- ❑ presentation
  - abdominal mass (most common presentation)
  - hypertension
  - flank tenderness
  - hematuria
- ❑ treatment
  - treatment of choice is surgical resection +/- radiation +/- chemotherapy
- ❑ prognosis
  - generally good; overall 5-year survival about 80%
  - metastatic disease may respond well

## ECTOPIC TESTES

- ❑ cryptorchidism refers to testes located somewhere along the normal path of descent (prepubic > external inguinal ring > inguinal canal > abdominal)
- ❑ incidence
  - 2.7% of full term newborns
  - 0.7%-0.8% at 1 year old
- ❑ consider
  - retractile testes
  - atrophic testes
  - intersex states
- ❑ treatment
  - undescended testes must be brought down before age 1-2 years as irreversible changes occur; after age 2 they should be brought down to monitor for malignancy
  - hormonal therapy (hCG or LH may facilitate their descent)
  - surgical descent
- ❑ prognosis
  - untreated bilateral cryptorchidism ~ 100% infertility
  - treated bilateral: 60-70% fertility rate
  - treated/untreated unilateral: fertility is still less than the general population
  - risk of malignancy is 40 x increased in undescended testes; this risk does not decrease with surgical descent, but monitoring is made easier

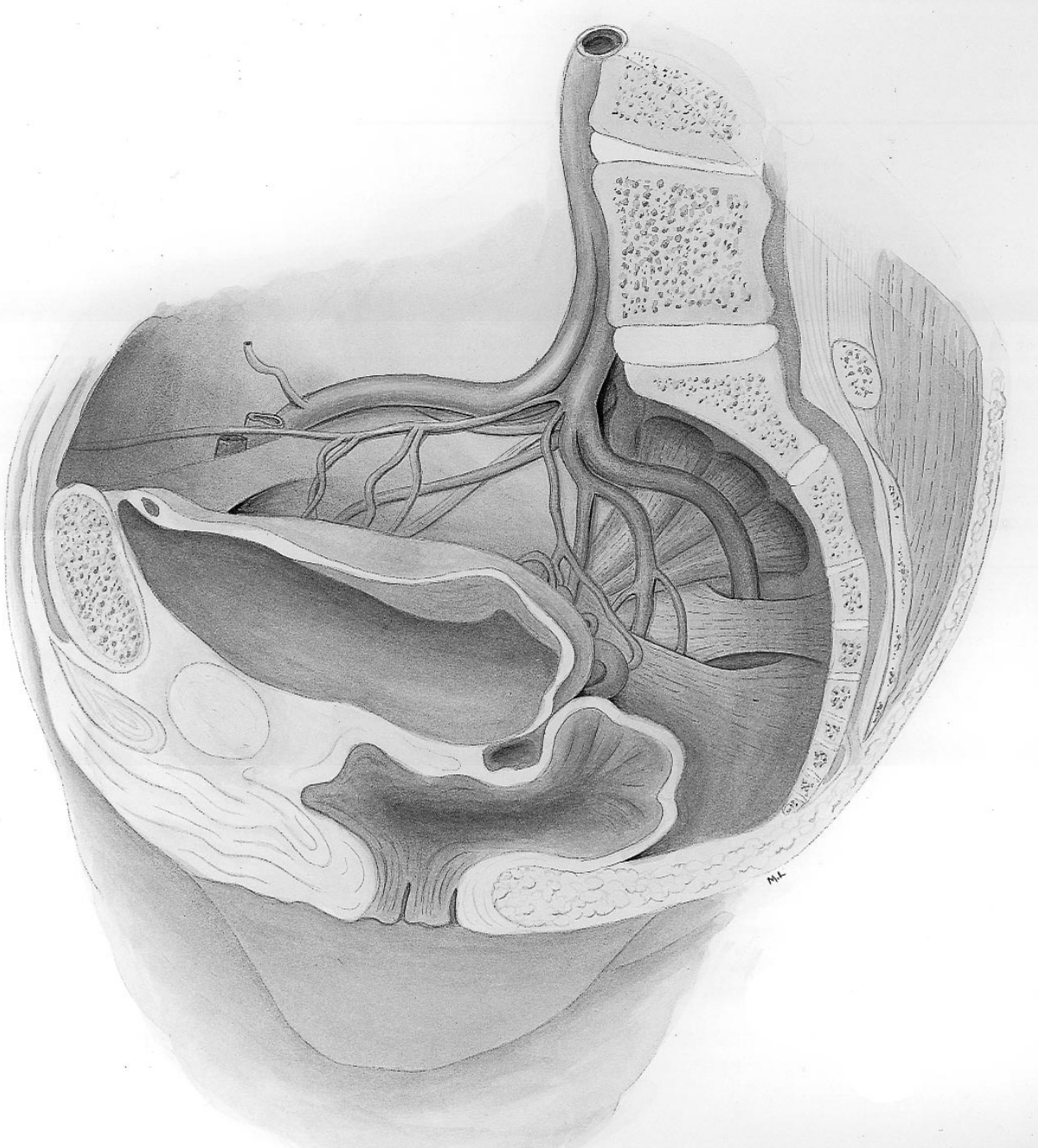
## AMBIGUOUS GENITALIA

### Definition and Classification

- ❑ genitalia that do not have a normal appearance based on the chromosomal sex of the child
- ❑ five major categories (proposed by Allen, 1976) based on histology and etiology
  - female pseudohermaphroditism (ovary only)
    - all are 46XX
    - most due to congenital adrenal hyperplasia
  - male pseudohermaphroditism (testis only)
    - all are 46XY
    - androgen insensitivity syndromes
  - true hermaphrodite (ovary plus testis)
  - mixed gonadal dysgenesis (testis plus streak gonad)
  - pure gonadal dysgenesis (two streak gonads)

### Diagnosis and Treatment

- ❑ thorough maternal and family history needed
  - other forms of abnormal sexual development
  - parent consanguinity
- ❑ physical exam very important in determining appropriate gender to assign
- ❑ laboratory tests
  - plasma 17-hydroxyprogesterone levels - elevated in 21-hydroxylase deficiency
  - DHEA and pregnenolone - elevated in 3-beta-hydroxysteroid dehydrogenase deficiency
  - corticosterone and deoxycorticosterone - elevated in 17-hydroxylase deficiency
  - testosterone levels alone not helpful
- ❑ ultrasound of adrenals, gonads, uterus
- ❑ endoscopy of urogenital sinus
- ❑ chromosomal evaluation - karyotype
- ❑ repair of external genitalia before 1 year of age
  - clitoral recession or clitoroplasty
- ❑ long-term psychological guidance and support for both patient and family



*Drawing by Monique LeBlanc*