

Urinary system 5 Congenital and cystic diseases of the kidney, urinary calculi and urinary obstruction

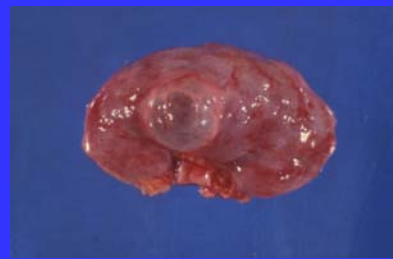
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Congenital renal disease

- ~ 10% born with potentially significant malformation of urinary tract, but congenital renal disease much less common
- renal agenesis and hypoplasia cause ~ 20 % renal failure in kids
- ectopic kidneys and horseshoe kidney also potentially important

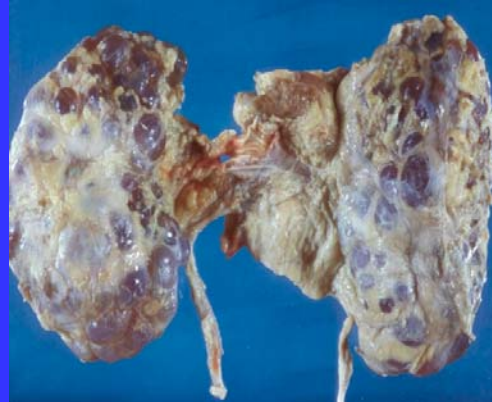
Cystic diseases of the kidney

- cysts may be
 - solitary or multiple
 - unilateral or bilateral
 - congenital (hereditary or not) or acquired
- many uncommon, but may be important if genetic screening & counselling available



Simple cysts

- common, usually noted on X ray
- increasing incidence with age
- single or multiple
- few mms to several cms
- smooth lining, clear fluid
- no effect on renal function
- occasionally haemorrhage, causing pain
- only real issue is distinction from tumour



AUTOSOMAL DOMINANT RENAL POLYCYSTIC DISEASE (ADPKD)

(there is also an ARPKD)

- relatively common in Europe (~ 1 in 500 births)
- multiple bilateral cysts - enlarge & compress renal parenchyma
- congenital, but presents any time from late childhood on (usually early/mid adulthood)
- autosomal dominant inheritance with high penetrance – so screening important if it's available
- genetic mutations for this condition now clear (one of 3 different genes - PKD 1-3)

ADPKD

- usually presents as one of -
 - chronic renal failure (up to 10% patients with CRF)
 - hypertension
 - or abdominal mass
- may also be
 - pain, haematuria, UTI, calculi
- occasionally cysts in other organs (clinically unimportant)
- and cerebral berry aneurysms cause death in up to 10%

ADPKD

- kidneys enlarged – usually at least 1000G each
- masses of cysts, up to 3 - 4 cm diameter - usually no obvious intervening parenchyma.
- cysts arise at all levels of the tubule
- filled with clear serous fluid +/- evidence of haemorrhage



Other cystic diseases

- dialysis-associated cystic disease
 - small cysts common in patients on long-term dialysis, prob due to tubular blockage in scarred kidneys: usually unimportant, but a few undergo malignant change - renal carcinoma
- uraemic medullary cystic disease (nephronophthisis) – cysts at CM junction
 - responsible for 20% CRF in children/adolescents
- medullary sponge kidney – cysts in papillae
 - no effect on renal function, but calculi can arise in cysts
- renal “dysplasia” – cysts all over
 - islands of undifferentiated tissue in kidney, usually cystic
 - only important if bilateral

Renal and urinary calculi – (nephrolithiasis & urolithiasis)

- maybe 1- 5% population at any one time in UK – much higher in Middle East (often bladder), less common in the tropics
 - ? protein (prothrombin fragment 1) in urine of black races inhibits crystal formation
- usually young/middle aged adults
- overall, twice as common in men than women
 - but calculi related to infection more common in women
- often recurrent - half will have another calculus within 10 yrs

Types of calculi

- calcium stones (Ca^{++} in complex with oxalate or phosphate or both) – most common stone
- triple ($\text{Mg NH}_4 \text{ PO}_4$) or struvite stones – quite common
- uric acid stones – 5%
- cystine or pure oxalate stones - inborn errors of metabolism -1%
- HIV – not really stones, but crystal precipitation due to antiviral agents

Pathogenesis of calculi

- most important factor is increased urinary concentration of stone's constituents
 - once solubility exceeded, precipitation occurs
- enhanced by any reduction in urinary solubility – especially rise in pH or lack of inhibitors of crystal formation (citrate or pyrophosphate)
- urinary protein may act as “nidus” for stone formation

Calcium stones

- most patients have hypercalciuria
 - but only 10% have hypercalcaemia
 - e.g. due hyperparathyroidism, sarcoid, vit D intoxication, prolonged immobilisation etc
- so ? defect in tubular calcium reabsorption
- (excretion of uric acid in urine also favours calcium stone formation)
- (*“nephrocalcinosis” is different pathology – refers not to stones/calculi but flecks of calcification in kidney - often due to hypercalcaemia and usually clinically unimportant*)

Triple (struvite) stones

- almost always females with persistently alkaline urine due to UTI
- certain bacteria hydrolyse urea to form NH_4
 - especially *P vulgaris/mirabilis*
- bacteria also serve as nidi for stone formation
- (calculi often “staghorn” shaped – cast of part/all of pelvi-calyceal system)

Uric acid stones

- associated with gout (25% of patients with gout) or leukaemias (high cell turnover)
- hyperuricaemia causes hyperuricosuria
- but 50% of patients have neither
- (hyperuricosuria also favours calcium stone formation)

Renal and urinary calculi

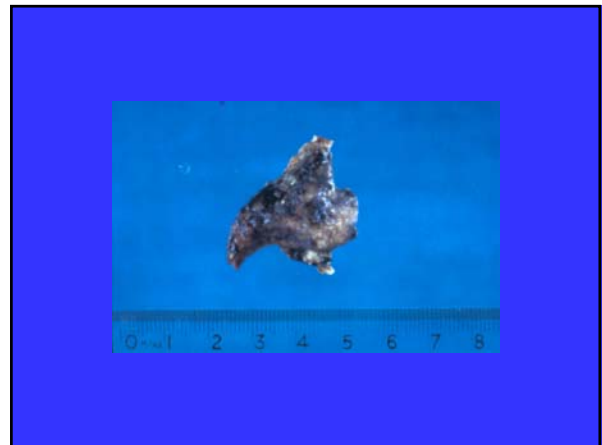
- usually unilateral (80%)
- may be multiple
- common sites for formation are pelvi-calyceal system or bladder
- mostly small (2-3 mm)
- but “staghorn” calculi are very large

Clinical effects

- depends on type, size and site of origin and/or arrest
- can be asymptomatic
- migration into ureter producing “renal” colic
- urinary obstruction
- erosion of mucosa - haematuria
- recurrent, intractable ascending urinary infection
- renal damage (hydronephrosis and pyelonephritis)
- squamous metaplasia of urothelium, so slight risk of squamous carcinoma

Where do stones stick in urinary tract?

- staghorn – usually in pelvi-calyceal system
- the other types –
 - pelvi-ureteral junction
 - where ureter crosses pelvic brim/iliac artery
 - lower end of ureter
- stones formed in bladder usually stay there



Urinary obstruction (obstructive uropathy)

- can occur at any level of urinary tract from renal pelvis to external meatus
- like obstruction of any hollow organ may be
 - partial or total
 - and acute or chronic
- variety of causes – e.g. lumen, wall and external
- increases susceptibility to urinary infection and stone formation
- if unrelieved usually leads to dilatation of pelvi-calyceal system and then renal atrophy

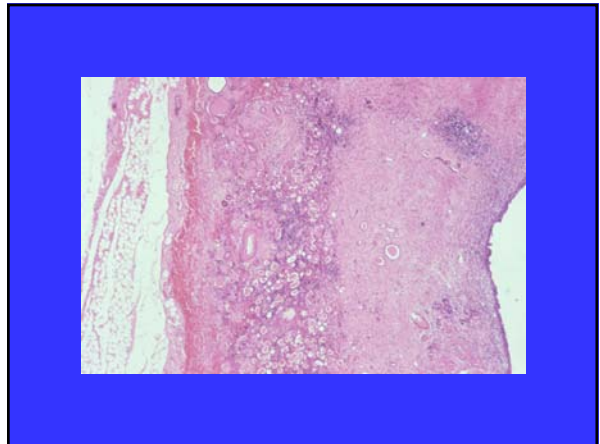
Hydronephrosis

- dilation of pelvi-calyceal system
- progressive pressure atrophy of the kidney
- obstruction also triggers interstitial nephritis, causing interstitial fibrosis
- any associated infection (pyelonephritis) will add to pressure effect and interstitial nephritis and so magnify renal damage
- (acute obstruction will cause acute renal “failure”)



Hydronephrosis

- progressive damage
- gross thinning of cortex due to parenchymal atrophy
- eventually, kidney becomes thin-walled "cyst"
- if blockage is in ureter or lower, ureter(s) may also dilate (hydroureter)

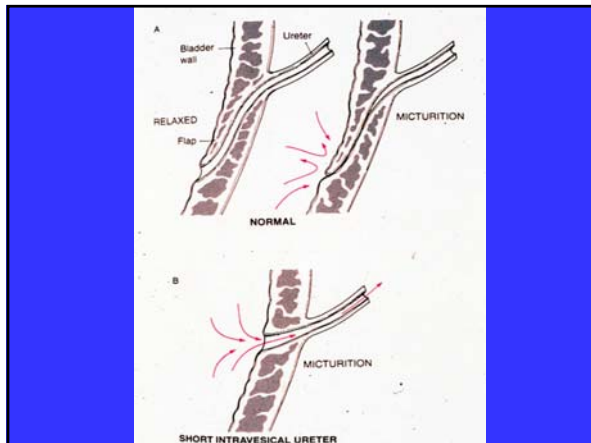


Unilateral disease

- block must be at level of vesico-ureteral (VU) valve or above
- may be silent for long time
- unilateral renal damage can cause secondary hypertension
 - and so possible effects on other kidney

Bilateral disease

- blockage must be at level of internal bladder sphincter or beyond
- if obstruction incomplete
 - bladder muscle hypertrophies, causing trabeculation
 - diverticula may form
 - bladder dilatation may make VU junctions incompetent
- will always cause renal failure if unrelieved

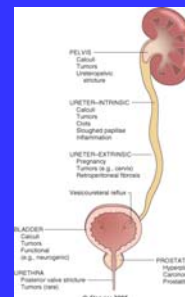


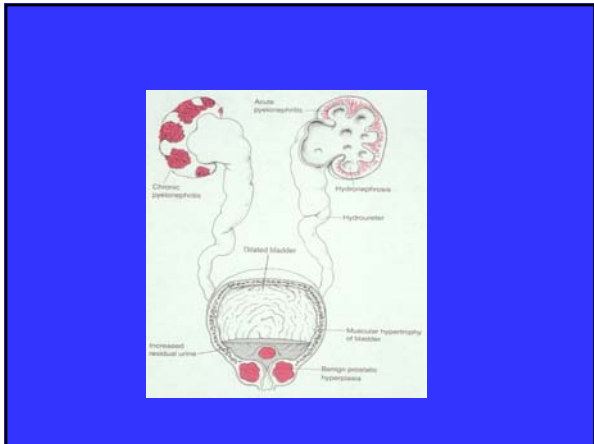
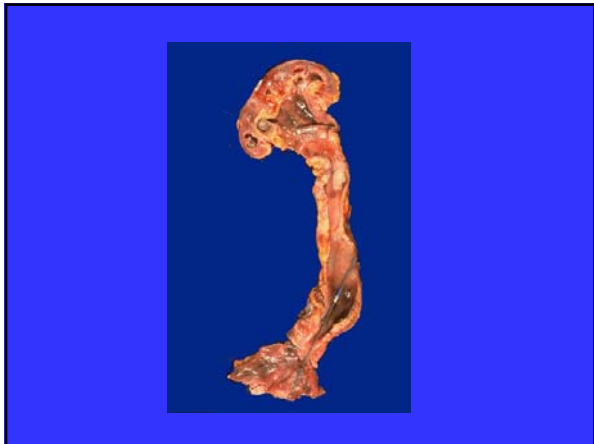
Causes of obstruction

- in the lumen
 - stones, blood clots, necrotic renal papillae
 - in the wall
 - intrinsic tumours, strictures (post-inflammatory*, congenital), neuropathic bladder, "pelviureteric dysfunction"
- *schistosomiasis important cause

Causes of obstruction

- outside the wall
 - inflammation
 - diverticulitis, salpingitis, prostatitis, retroperitoneal fibrosis
 - tumours (prostate, cervix, uterus, colon, enlarged retroperitoneal nodes)
 - others
 - benign prostatic hypertrophy, endometriosis, aortic aneurysm, prolapsed uterus etc – even (temporarily) pregnancy





and finally -

Diseases of the urethra

Often present with urinary obstruction

- congenital valves
- traumatic rupture/stricture
- urethritis – e.g. due to gonococcus
- tumours – viral condyloma, transitional tumours