CYSTIC DISEASES OF THE KIDNEY
• Types

1-Simple Cysts

2-Autosomal Dominant (Adult) Polycystic Kidney Disease

3-Autosomal Recessive (Childhood) Polycystic Kidney Disease

4-Medullary Cystic Disease

1-Simple Cysts

• Multiple or single cystic spaces that vary widely in diameter (1-5 cm in diameter) filled with clear fluid.

• usually confined to the cortex.

• common post-mortem finding that has no clinical significance.

• The main importance of cysts lies in their differentiation from kidney tumors when they are discovered either incidentally or because of hemorrhage and pain
Simple renal Cysts
Dialysis-associated acquired cysts

- occur in the kidneys of patients with end-stage renal disease who have undergone prolonged dialysis.
- They are present in both cortex and medulla and may bleed causing hematuria.
- renal adenomas or even carcinomas may arise in the walls of these cysts.
Cystic change associated with chronic renal dialysis.
2-Autosomal Dominant (Adult) Polycystic Kidney Disease

- Characterized by multiple expanding cysts of both kidneys that ultimately destroy the intervening parenchyma.
- Incidence (1: 500-1000) persons
- Accounts for 10% of cases of chronic renal failure.
- It can be caused by inheritance of one of at least 2 autosomal dominant genes of very high penetrance.

1. **PKD1** on the short arm of chromosome 16

- In 85-90% of families
- This gene encodes a large and complex cell membrane-associated protein called polycystin-1
(2)- *PKD2* gene (10-15% of cases) on chromosome 4:

- encodes *polycystin 2*.
- Polycystin 2 is thought to function as a calcium-permeable membrane channel.
- polycystins 1 and 2 are believed to act together by forming heterodimers.
- mutation in either gene gives rise to essentially the same phenotype although patients with *PKD2* mutations have a slower rate of disease progression as compared with patients with *PKD1* mutations.
• **Clinical presentation**
  • asymptomatic until the 4th decade by which time the kidneys are quite large although small cysts start to develop in adolescence.
  • The most common presenting complaint is flank pain or a heavy dragging sensation.
  • Acute distention of a cyst either by intracystic hemorrhage or by obstruction may cause excruciating pain.
  • palpation of an abdominal mass.
  • Intermittent gross hematuria commonly occurs.
  • hemorrhage.

• **Complications**
  • 1-hypertension (75%).
  • 2-urinary infection.
  • 3-Saccular aneurysms of the circle of Willis are present in 10% to 30% of patients (subarachnoid hemorrhage).
  • 4-end-stage renal failure occurs at about age 50.
3-Autosomal Recessive (Childhood) Polycystic Kidney Disease

• autosomal recessive inheritance.
• 1:20,000 live births.
• Perinatal, neonatal, infantile, and juvenile subcategories have been defined, depending on time of presentation and the presence of associated hepatic lesions.
• Mutations in \textit{PKHD1} gene coding for a putative membrane receptor protein called \textit{fibrocytin}, localized to chromosome 6p.
• Fibrocytin may be involved in the function of cilia in tubular epithelial cells.
Normal term infant kidneys
Cysts are fairly small but uniformly distributed throughout the parenchyma so that the disease is usually symmetrical in appearance with both kidneys markedly enlarged.
4-Medullary Cystic Disease

- There are 2 major types of medullary cystic disease:
  - **1-medullary sponge kidney**
  - a relatively common and usually innocuous condition.
  - **2-nephronophthisis-medullary cystic disease complex**
  - is almost always associated with renal dysfunction.
  - usually begins in childhood.
  - 4 variants of this disease complex are recognized on the basis of the time of onset: infantile; juvenile (most common); adolescent; adult
• **Clinical features**

  • polyuria and polydipsia a consequence of diminished tubular function.
  
  • Progression to end-stage renal disease ensues over a 5-10-year period.
  
  • The disease is difficult to diagnose, since there are no serologic markers and the cysts may be too small to be seen with radiologic imaging.
  
  • cysts may not be apparent on renal biopsy if the cortico-medullary junction is not well sampled.
  
  • A positive family history and unexplained chronic renal failure in young patients should lead to suspicion of medullary cystic disease.
URINARY OUTFLOW OBSTRUCTION

- **Renal Stones** *Urolithiasis*
  - Calculus formation at any level in the urinary collecting system.
  - Most common arise in the kidney.
  - (1%) of all autopsies.
  - Symptomatic urolithiasis is more common in men than in women.
  - Familial tendency toward stone formation
  - *Pathogenesis*
  - Renal stones are composed of:
    - 1- calcium oxalate or calcium oxalate mixed with calcium phosphate (80%) .
    - 2- 10% are composed of magnesium ammonium phosphate.
    - 3- 6%-9% are either uric acid or cystine stones
    - In all cases there is an organic matrix of mucoprotein that makes up about 2.5% of the stone by weight.
• **Causes**

• *1-increased urine concentration of the stone's constituents so that it exceeds their solubility in urine (supersaturation).*

• 50% of patients who develop *calcium stones* have hypercalciuria that is not associated with hypercalcemia.

• **Hypercalciuria:**
  
  A. absorptive hypercalciuria.
  
  B. renal hypercalciuria due to primary renal defect of calcium reabsorption.

• In 5% to 10% of persons there is hypercalcemia and consequent hypercalciuria.
- The presence of a nidus
  - Urates provide a nidus for calcium deposition.
  - Desquamated epithelial cells

- urine pH
  - High urine pH favors crystallization of calcium phosphate and stone formation.
  - *Magnesium ammonium phosphate (struvite) stones* almost always occur with a persistently alkaline urine due to UTIs.
  - Uric acid stones formed in acidic urine (under pH 5.5).
  - *Cystine stones* are more likely to form when the urine is relatively acidic.
• **4-infections**
  • The urea-splitting bacteria such as *Proteus vulgaris* and the staphylococci predispose the person to urolithiasis.

• **5-lack of substances that normally inhibit mineral precipitation.**
  • Inhibitors of crystal formation in urine include Tamm-Horsfall protein, osteopontin, pyrophosphate, mucopolysaccharides, diphosphonates, and a glycoprotein called nephrocalcin.
  • No deficiency of any of these substances has been consistently demonstrated in individuals with urolithiasis.
• Stones are unilateral in about 80% of patients.
• Common sites of formation are renal pelvis and calyces and the bladder.
• They tend to be small (average diameter 2-3 mm) and may be smooth or jagged.
• Progressive precipitation of salts leads to the development of branching structures known as staghorn calculi.
• These massive stones are usually composed of magnesium ammonium phosphate.
Hydronephrosis

• Refers to dilation of the renal pelvis and calyces, with accompanying atrophy of the parenchyma.

• The obstruction may be sudden or insidious and it may occur at any level of the urinary tract from the urethra to the renal pelvis.

• The most common causes are as follows:
  
  • **1-Congenital:**
    • Atresia of the urethra
    • Valve formations in either ureter or urethra
    • Aberrant renal artery compressing the ureter
    • Renal ptosis with torsion or kinking of the ureter
• **2-Acquired:**
  - Foreign bodies: Calculi, necrotic apillae
  - Tumors: Benign prostatic hyperplasia, carcinoma of the prostate, bladder tumors (papilloma and carcinoma), contiguous malignant disease (retroperitoneal lymphoma, carcinoma of the cervix or uterus)
  - Inflammation: Prostatitis, ureteritis, urethritis, retroperitoneal fibrosis
  - Neurogenic: Spinal cord damage with paralysis of the bladder
  - Normal pregnancy: Mild and reversible
Hydronephrosis of the kidney, with marked dilation of the pelvis and calyces and thinning of renal parenchyma.