Renal Cystic Diseases

Classification

Non-Genetic

- **Developmental:**
  - Medullary sponge kidney
  - Renal cystic dysplasia:
    - Multicystic dysplasia
    - Cystic dysplasia with lower urinary tract obstruction
  - Diffuse cystic dysplasia (syndromal / nonsyndromal)
- **Acquired:**
  - Simple cysts
  - Solitary multilocular cysts
  - Hypokalemic cystic disease
  - Acquired cystic disease (advanced renal failure)

Genetic

- ☑ **Autosomal Dominant**
  - Autosomal dominant polycystic kidney disease (ADPKD)
  - Adult-onset medullary cystic disease
  - Tuberous sclerosis
  - Von Hippel–Lindau disease
- ☑ **Autosomal Recessive**
  - Autosomal recessive polycystic kidney disease (ARPKD)
  - Juvenile nephronophthisis
  - Meckel–Gruber syndrome
- ☑ **X-Linked**
  - Orofaciodigital syndrome type I

Autosomal Dominant Polycystic Kidney Disease (ADPKD)

Definition & Epidemiology

- **Inheritance:** Autosomal dominant
- **Incidence: 1:400–1:1000** live births
- **ESRD in USA: 6.9–8.7 / million / year**

✧ Characteristics

- Multiple bilateral renal cysts
- Extra-renal cysts: liver, pancreas, arachnoid membranes

✧ Diagnosis

☑ **Ultrasound (Revised Unified Criteria)**

- **Age 15–29: >3 cysts** unilateral or bilateral
- **Age 30–39: ≥3 cysts** unilateral or bilateral
- **Age 40–59: ≥2 cysts** in each kidney
- **Age ≥60: ≥4 cysts** in each kidney

☑ **Genetic Testing**

- Indications:
  - Indeterminate imaging
  - Presymptomatic screening after counseling
- Outcomes:
  - If present: genetic counseling + early BP control
  - If absent: reassurance

✧ Renal Manifestations

- **Functional:**
  - Concentrating defect
  - Reduced renal blood flow
- **Hypertension:**
  - Cardiac
  - Cerebrovascular
  - Arteriolosclerosis
  - Peripheral vascular disease
- **Pain:**
  - Cyst hemorrhage
  - Gross hematuria
  - Nephrolithiasis
  - Infection
  - Renal enlargement
- **Renal failure:**
  - Interstitial inflammation
  - Tubular apoptosis
  - Hypertensive glomerulosclerosis
  - Compression atrophy

✧ Extra-Renal Manifestations

☑ **Polycystic Liver Disease**

- Usually asymptomatic
- Conservative:
  - Avoid ethanol, hepatotoxins, caffeine
- Invasive:
  - Cyst aspiration & sclerosis
  - Fenestration
  - Partial hepatectomy
  - Liver transplantation

☑ **Intracranial Aneurysms**

- **Screening NOT for all**
- Indications:
  - Family history
  - Previous rupture
  - Pre-op high-risk surgery
  - High-risk occupations
  - Severe patient anxiety
- **Modality:** MR angiography
- Management:
  - **<5 mm:** follow-up imaging
  - **6–9 mm:** controversial
  - **>10 mm:** surgery or endovascular coils

☑ **Other Vascular Abnormalities**

- Thoracic aortic dissection
- Cervicocephalic dissection
- Coronary aneurysms
- Retinal artery/vein occlusion

☑ **Cardiac**

- Mitral valve prolapse (**25%**)
- Mitral regurgitation
- Tricuspid regurgitation/prolapse

☑ **Other**

- Pancreatic cysts (**5%**)
- Seminal vesicle cysts (**40%**)
- Arachnoid membrane cysts (**8%**)
- Colonic & duodenal diverticulae

✧ Treatment of ADPKD Complications

☑ **Flank Pain**

- Exclude stones, infection, tumors
- Avoid NSAIDs
- Opioids (acute)
- SSRIs + lifestyle (chronic)
- Cyst decompression

☑ **Hematuria & Cyst Hemorrhage**

- Conservative:
  - Bed rest
  - Analgesia
  - Fluids
  - Tranexamic acid
- Severe:
  - Hospitalization
  - Blood transfusion
- Persistent:
  - Arterial embolization
  - Surgery

☑ **Infection**

- Prompt treatment of ASB & cystitis

☑ **Nephrolithiasis**

- Potassium citrate
- ESWL
- Percutaneous nephrolithotomy

☑ **Progressive Renal Failure**

- BP target **125/75 mmHg**
- ACEIs or ARBs
- Increased water intake

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

✧ Definition & Genetics

- Severe early-onset cystic disease
- Gene: **PKHD1**
- Protein: fibrocystin/polyductin
- Tubular abnormality: fusiform/saccular dilation of collecting ducts

✧ Epidemiology

- Incidence: **1:20,000**
- More common in Caucasians

✧ Clinical Manifestations

☑ **In Utero / At Birth (Potter Phenotype)**

- ◈ Enlarged echogenic kidneys
- ◈ Oligohydramnios
- ◈ Pulmonary hypoplasia
- ◈ Characteristic facies
- ◈ Limb & spine deformities
- ◈ Perinatal mortality **~30%**

☑ **Survivors (>1 month)**

- ◈ **5-year survival: 85–90%**
- ◈ Hypertension (**70–80%**)
- ◈ Diluting & concentrating defects
- ◈ Recurrent UTIs
- ◈ Initial GFR improvement → progressive decline
- ◈ Portal hypertension

☑ **Childhood / Adolescence**

- ◈ Portal hypertension:
  - ↳ Hepatosplenomegaly
  - ↳ Variceal bleeding
  - ↳ Hypersplenism
- ◈ Preserved hepatocellular function
- ◈ Ascending suppurative cholangitis

✧ Diagnosis

- ◈ Imaging:
  - ↳ Kidneys enlarged **10×**
  - ↳ Hyperechogenic medulla
- ◈ Genetic testing:
  - ↳ Detection rate **80–87%**
  - ↳ Prenatal & preimplantation diagnosis

✧ Treatment

- ◈ Ventilatory & neonatal support
- ◈ BP control
- ◈ Renal supportive care
- ◈ Hydration monitoring
- ◈ Portal hypertension management
- ◈ Aggressive antibiotics for cholangitis

Medullary Sponge Kidney

Epidemiology

- Incidence: **1:5000**
- Onset: **4th–5th decade**
- Sporadic

Characteristics

- Dilated medullary & papillary collecting ducts
- Spongy medulla

Clinical Features

- Nephrocalcinosis
- Recurrent calcium nephrolithiasis
- Hematuria
- UTIs
- Reduced concentrating & acidifying ability

Treatment

- Potassium citrate
- High fluid intake
- Thiazides for hypercalciuria
- Antibiotics for UTI
- Renal impairment uncommon

Simple Renal Cysts

Features

- Most common acquired cystic lesion
- Usually unilateral
- Single or multiple
- Usually asymptomatic

Investigation (Ultrasound Criteria)

- Smooth walls
- No septae
- No intracystic debris

Treatment

- None usually required
- Symptomatic or renin-dependent HTN:
  - US-guided aspiration
  - Sclerotherapy