Autosomal Dominant Polycystic Kidney Disease

By Amar Udare, MBBS [2]

Case History: 45-year-old female with vague pain in the abdomen.

Case History: A 45-year-old female presented with vague pain in the abdomen. A USG was ordered, which revealed that both kidneys were enlarged and had innumerable cysts with minimal intervening parenchyma (Figure 1A and 1B). An ill-defined hyperechoic area with posterior acoustic shadowing was seen in the left kidney. The ureters and bladder were normal. A few similar cysts were also seen in the liver (Figure 2). The findings were confirmed on a plain CT scan which revealed bilateral nephromegaly and cysts in both kidneys and the liver. Multiple calculi were also seen in the left kidney (Figures 3-5). No cysts were identified in the pancreas and spleen. Features were suggestive of autosomal dominant polycystic kidney disease.

Figure 1 A and B. Ultrasound image shows that the right and left kidney are enlarged in size and have innumerable cysts. No definite renal parenchyma is identified. Ill-defined shadowing is seen in the left kidney.
Figure 2. Ultrasound of the liver shows cysts in the liver.
**Figure 3.** Non-contrast CT scan axial sections reveal multiple cysts in the liver and kidney. Multiple calculi are also noted at the lower pole of the left kidney.

**Figure 4.** Non-contrast CT scan axial sections reveal multiple cysts in the liver and kidney. Multiple calculi are also noted at the lower pole of the left kidney.

**Figure 5.** Non-contrast CT scan axial sections reveal multiple cysts in the liver and kidney. Multiple calculi are also noted at the lower pole of the left kidney.
Diagnosis: Autosomal dominant polycystic kidney disease

Incidence

- 1 in 400-1,000 persons in U.S.
- M:F- 1:1
- Presents late in life: 30-50 years (as compared to ARPKD which presents in early childhood)
- Presentation
  - Asymptomatic
  - Flank pain
  - Rarely hypertension and renal failure

Inheritance

- Autosomal dominant (90 percent); spontaneous mutations (10 percent)
- Chromosomes: 16 and 4

Associated abnormalities:

- Non cystic
  - Cerebral berry aneurysms
    - Intracranial dolichoectasia: hypertension: up to 80 percent adults
  - Colonic diverticulosis
  - Hernias
  - Bicuspid aortic valve
  - Mitral valve prolapse
  - Aortic dissection

- Cystic
  - Liver: most common, 75 percent by age 60
  - Ovaries
  - Spleen
  - Seminal vesicles
  - Prostate
  - Pancreas

Imaging

**IVP**

- Plain film
  - Increased renal size with curvilinear wall calcification
  - Renal calculi
  - "Swiss cheese" pattern: smoothly margined radiolucencies in cortex and medulla seen on nephrographic phase
  - Smooth, bosselated renal contour
  - Normal or effaced collecting system

**USG**

- Investigation of choice
  - Shows increased renal size, innumerable cysts in both kidneys
  - Simple cysts will have anechoic walls with posterior acoustic enhancement
  - Hemorrhagic cysts show echogenic material devoid of blood flow

**CT**
<table>
<thead>
<tr>
<th>Type of cyst</th>
<th>Non contrast CT</th>
<th>CECT</th>
</tr>
</thead>
<tbody>
<tr>
<td>Uncomplicated</td>
<td>Hypodense cysts (water attenuation) + thin wall</td>
<td>No enhancement</td>
</tr>
<tr>
<td>Hemorrhagic</td>
<td>Hyperdense; Perinephric hematomas; Mural calcification</td>
<td>Hypodense as compared to normal renal parenchyma</td>
</tr>
<tr>
<td>Infected</td>
<td>Hypodense; Thick irregular cyst wall</td>
<td>Wall enhancement</td>
</tr>
</tbody>
</table>

CT will also confirm the presence of cysts in other organs.

**MR Findings**

- T1: Hypointense
- T2: Hyperintense
- Post contrast: no contrast enhancement in simple cysts

**Differentials for Multiple Cysts**

- **Multiple simple cysts:**
  - Usually older presentation
  - No nephromegaly
  - History of renal cystic disease
  - Cysts are not found in other organs

- **Von Hippel-Lindau disease**
  - Multiple renal and pancreatic cysts, pheochromocytomas, and frequently multiple and bilateral RCCs
  - Associated lesions include retinal angiomas and cerebellar hemangioblastomas
  - Cysts are less numerous
  - Pancreatic cysts are more common

- **Tuberous sclerosis**
  - Multiple renal cysts and multiple fat containing angiomyolipoma
  - Cutaneous, retinal and cerebral hamartomas are associated

- **Acquired uremic cystic kidney disease**
  - Development of multiple cysts in the native kidneys of patients on long-term hemodialysis
  - Affected kidneys are usually small, reflecting the chronic renal disease
  - Cysts are predominantly cortical and rarely exceed 2cm

**References**


**Source URL:**
Links: