



“Why does my stomach hurt?”

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Ken, a 39-year-old single accountant, presents with a history of :

- general tiredness,
- recurrent headaches and
- insomnia.

For the previous three months, he has had upper abdominal discomfort on both sides. His appetite has also deteriorated and he has lost about 3 kg in weight.

Medical history

Ken's medical history reveals the following:

- his last medical exam was done about 10 years ago,
- he denies excessive drinking, although he agrees that he does drink sociably,
- he smokes 30 cigarettes per day and
- his family history is unknown as he was adopted.

Physical examination

Upon examination, the following is noted:

- Pulse: 88 bpm
- BP: 195/108 mmHg
- A soft systolic murmur is heard over the precordium
- General tenderness is found in both the upper sides of the abdomen
- Pitting oedema of both ankles
- Fundi shows a narrowing and tortuosity of the retinal arteries with arterio-venous nipping

Clinical investigations

Clinical investigations show:

- Enlarged cardiac silhouette on chest x-ray
- Left ventricular hypertrophy on ECG
- Hemoglobin: 119 g/L
- Creatinine (serum): 139 $\mu\text{mol/L}$
- Creatinine clearance: 45 ml/minute
- Urine analysis: proteinuria and hematuria

CT scans of the abdomen are taken (Figures 1 and 2)

What's your diagnosis?

- a) Simple renal cysts
- b) Polycystic kidney disease
- c) Tuberosus sclerosis
- d) Bilateral kidney tumours

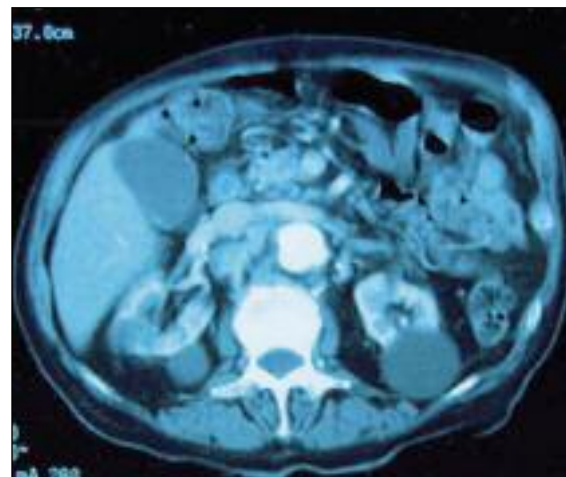


Figure 1. CT scan of abdomen.



Figure 2. CT scan of chest.

Answer: B
Polycystic kidney disease

What is polycystic kidney disease?

Polycystic kidney disease (PKD) is one of the most common inherited disorders in humans. It is the most frequent genetic cause of renal failure in adults, accounting for 10% of patients on dialysis in the US.

Characteristics

PKD is a multisystemic and progressive disorder characterized by the formation and enlargement of renal cysts in the kidney and other organs, such as the:

- liver,
- pancreas and
- spleen.

Clinical features usually begin in the third to fourth decade of life, but cysts may be detectable in childhood and in utero.

PKD is associated with:

- brain aneurysms,
- diverticula of the colon and
- cysts in the:
 - liver,
 - pancreas and
 - testes.

As many as half of those with PKD also have cysts on their livers.

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Pathophysiology

In the early stages of the disease, the cysts on the kidneys enlarge and interfere with kidney function, resulting in chronic high BP and kidney infections. The cysts may cause the kidneys to increase their production of erythropoietin (the hormone that stimulates production of red blood cells) resulting in too many red blood cells, rather than the expected anemia of chronic kidney disease. Bleeding in a cyst can cause flank pain. Kidney stones are more common in people with PKD. Hypertension (high BP) caused by polycystic kidneys may be difficult to control.

Slowly, the disease progresses, eventually resulting in end-stage kidney failure.

PKD is also associated with liver disease, including infection of liver cysts.

Classic clinical findings in PKD are a positive family history for renal disease in 75% of reported cases, bilateral flank masses, elevated BP and uremia.

Varying forms

An autosomal recessive form of PKD also exists and appears in infancy or childhood; it tends to be very serious and progresses rapidly, resulting in end-stage kidney failure and generally causing death in infancy or childhood.

Clinical findings

Classic clinical findings in PKD are:

- a positive family history for renal disease in 75% of reported cases,
- bilateral flank masses,
- elevated BP and
- uremia.

Flank pain is the earliest symptom. It begins months-to-years before detectable renal enlargement. Renal failure follows nephromegaly. Patients come to the attention of physicians because of:

- pain,
- bleeding,

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- urinary tract infections,
- nephrolithiasis, or
- obstructive uropathy.

Treatment

Treatment is nonspecific and symptomatic. Renal transplantation and dialysis will prolong life once end-stage renal failure occurs. Genetic counselling is important. 