AN INTERESTING CASE OF ABDOMINAL MASS

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PRESENTATION OF CASE

A 52-year-old lady, a housewife, presented to the medicine OPD with the complaints of swelling in the right flank which she had initially noticed 2 years back, but had neglected. But since last 1 month, she noticed an increase in size of this swelling as well as another similar swelling in her left flank as well. At the same time, she developed bilateral pedal oedema. There was no h/o dysuria, haematuria, lithuria or decreased urine output, neither was there any h/o vomiting, recent loss of weight or loss of appetite.

She was detected to be hypertensive at the age of 30 years and since then she is on 2 anti-hypertensive drugs. There was a family history of kidney disease in her elder sister, who is now dialysis dependent and her mother who expired few years back from a complication of some kidney disease the details of which is not known to her.

On examination, she was conscious and oriented, moderately built and nourished. She was pale and has bilateral pitting pedal oedema. Her pulse rate was 82 per minute, regular and all peripheral pulses were palpable. Her blood pressure was 130/90 mmHg on 2 antihypertensive drugs. Abdominal examination showed a distended abdomen and flanks appeared full. There was epigastric hernia (Fig. 1). Liver was palpable 10 cm below the right costal margin and the surface was smooth. The liver span was 20 cm. The spleen was not palpable though. The mass in the lumbar regions were ballotable (Fig. 2). Examination of other systems were essentially normal, except for a grade 2 hypertensive retinopathy in the fundus.

DIFFERENTIAL DIAGNOSIS

After History and Examination, the Differential Diagnosis were

1. Polycystic kidney disease.
2. Renal cell carcinoma.

INVESTIGATIONS

Routine blood investigation revealed anaemia with a haemoglobin of 9.2. Her renal function tests and liver function tests were essentially normal. Her urine routine examination did not show any RBCs. ECG was also normal. So proceeded to imaging methods.

The ultrasonography of abdomen showed liver size of 24 cm with large complex cystic lesion of size 18 x 11.8 cm and multiple cystic lesions in both lobes. The spleen size was 12 cm and it also had multiple cystic lesions. Both the kidneys were grossly enlarged and had multiple complex cystic lesions. CECT abdomen confirmed the ultrasonographic findings (Fig. 3, 4). Another interesting finding in the CECT was a mild pericardial effusion (Fig. 5), which was further confirmed by ECHO.
DISCUSSION OF MANAGEMENT
Autosomal dominant polycystic kidney disease (ADPKD) is the most common life-threatening monogenic disease. The mutation is in PKD1 or PKD2 gene (85%, 15%) respectively. The prevalence of the disease is 1:1000 and only 50% is clinically diagnosed. It is usually asymptomatic till 4th-5th decade. The differential diagnosis of renal cysts with autosomal dominant inheritance pattern include ADPKD, tuberous sclerosis, medullary cystic kidney disease and Von Hippel-Lindau disease.

The renal complications of ADPKD include haemorrhage into cysts, which is seen in 30%-35% and it presents as flank pain and haematuria. The other complications include renal cyst infection, nephrolithiasis with predominantly uric acid stones, urine concentrating defects and rarely renal cell carcinoma. The extra renal complications are hepatic cysts which is seen in 50%-70% cases, especially in women with multiple pregnancies, cerebral aneurysms in 20% cases, pancreatic cysts and various cardiac manifestations like valvular diseases, pericardial effusion and coronary artery aneurysms. Abdominal wall hernia is seen in 45% cases.

Our patient was also evaluated for other extra renal complications like cerebral aneurysms, which is seen in 20% cases. So a Magnetic Resonance Angiography of brain was done, which did not show any evidence of aneurysm. Her two children were also evaluated for ADPKD, and they both did not have any evidence of disease at present, and hence they are kept under followup.

Her blood pressure was well controlled with antihypertensive drugs and she is kept under followup to look for development of further complications.

FINAL DIAGNOSIS
Autosomal Dominant Polycystic Kidney Disease with multiple extra renal complications including polycystic liver disease, epigastric hernia and pericardial effusion.

REFERENCES