Recurrent Pneumothorax Associated with Polycystic Kidney Disease

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ABSTRACT

Autosomal dominant polycystic kidney disease (ADPKD) is usually revealed by renal or extra-renal manifestations or may be discovered by imaging techniques.¹ We report a case of a woman who presented with recurrent right-sided pneumothoraces associated with multiple lung cysts, hepatic cysts and bilateral polycystic kidneys favouring a diagnosis of ADPKD. This case is reported due to the rare external manifestation in ADPKD of lung cysts presenting clinically as recurrent pneumothorax. [Indian J Chest Dis Allied Sci 2008; 50: 233-235]

Key words: Recurrent pneumothorax, Lung cysts, ADPKD, Hypertension.

INTRODUCTION

Adult polycystic kidney disease is an autosomal dominant trait with an incidence of 1:500 to 1:1000.² Patients usually present with hypertension and progressive renal failure after the third decade of life. The disease is characterised by the presence of bilateral polycystic kidneys and variable extra-renal manifestations. Cysts in other organs, especially the liver occur in about 50% of patients with autosomal dominant polycystic kidney disease (ADPKD). However, cysts in other organs are less common with pulmonary cysts being rarely reported.³ Our patient presented with two episodes of right-sided pneumothoraces. Further investigations and work-up confirmed a diagnosis of ADPKD that was associated with the presence of multiple lung cysts. This cases, is thus, reported due to the rarity of ADPKD presenting clinically as recurrent pneumothoraces due to the rupture of pulmonary cysts.

CASE REPORT

A 46-year-old hypertensive, pre-menopausal lady presented during September 2004 with complaints of progressively worsening right-sided chest pain and breathlessness of 10 days duration. She denied any history of fever, cough, chest wall injury or relationship to her menstrual cycle. Clinical examination and investigations revealed a small right-sided pneumothorax. A simple aspiration was done following which she improved with complete radiological expansion of the lung. The underlying lung parenchyma appeared normal. The patient remained asymptomatic till March 2005, when she developed similar symptoms.

On clinical examination, she was afebrile and tachypnoeic at rest with a respiratory rate of 26 per minute and blood pressure of 190/110 mmHg. Examination of the respiratory system revealed a hyperresonant note and diminished breath sounds in the right hemithorax. Other systems were clinically normal. Past medical history was remarkable in childhood but she had hypertension for the past five years and was on regular medications. She was neither an asthmatic or diabetic nor did she have any history of anti-tuberculosis treatment. Family history revealed that her mother and brother were hypertensive.

Baseline blood investigations including blood sugar, erythrocyte sedimentation rate (ESR), hepatic and renal profiles were normal. The tuberculin skin test was negative. A plain chest radiograph revealed a small and a large right-sided pneumothorax (Figure 1) in September 2004 and March 2005, respectively. In an attempt to investigate the cause of her recurrent spontaneous pneumothoraces, a computerised tomographic (CT) scan of thorax was done that revealed multiple cysts of varying sizes in both the lungs from apex to base (Figure 2), and the presence of multiple hepatic cysts. This prompted us to evaluate the abdomen by sonography and CT that revealed similar cysts in the liver (Figure 3) and both kidneys as well as...
a solitary cyst in the left ovary (Figure 4). The above imaging features are a classical presentation of ADPKD.4-6 Computerised tomographic angiogram of the brain showed a basilar saccular aneurysm. The latter have been reported in patients with ADPKD.7-9

This time when she came back to us in March 2005 with a large pneumothorax on the right side, an intercostal chest drain was inserted. While breathlessness improved, the plain chest radiograph revealed incomplete lung expansion and a persistent air leak.

**DISCUSSION**

Polycystic kidney disease is of two types: (i) autosomal dominant/adult polycystic kidney disease (ADPKD); (ii) autosomal recessive/infantile type polycystic kidney disease (ARPKD). This is associated with pulmonary hypoplasia and pneumothorax.

The most common of the polycystic kidney disorders, ADPKD, with an incidence of 1:500 to 1:1000 is inherited as an autosomal dominant trait with complete penetrance.10 The disease may be revealed by a renal or extra-renal complication or discovered at clinical examination or by imaging techniques.2 Autosomal dominant polycystic kidney disease may not be clinically apparent until the third or fourth decade of life.3 Renal clinical manifestations include hypertension, abdominal pain, haematuria and renal failure. Approximately 50% of individuals with ADPKD have end-stage renal disease by the age of 60 years.16

Of the extra-renal manifestations, hepatic involvement is the most common with about 50% of patients with ADPKD having hepatic cysts.12 The prevalence of liver cysts in individuals with ADPKD increases from 20% in the third decade to approximately 75% after the sixth decade of life.13 The incidence of cysts reported in other organs are as follows: pancreas (9%), spleen (<5%), seminal vesicles (60%) and intracranial arachnoid cysts (8%).13 Cysts of thyroid, ovaries, endometrium and epididymis have been reported but are rare. Intracranial aneurysms occur in approximately 10% of individuals with ADPKD. The prevalence is higher in those with a positive family history of
aneurysms or subarachnoid haemorrhage (22%) than in those without (6%).

Cardiac valvular abnormalities have been found in conjunction with ADPKD. Mitral valve prolapse is the most common valvular abnormality and has been demonstrated in up to 25% of affected individuals. Other anomalies reported are dilation of aortic root, dissection of thoracic aorta, coronary artery and aortic aneurysms.

The diagnosis of ADPKD is established primarily by imaging studies of the kidney. Ultrasonography is the procedure of choice in the initial screening, work-up and diagnosis. The clinical diagnostic criteria relying on sonographic findings have a sensitivity of nearly 100 percent. Molecular genetic testing by sequence analysis can be helpful when the imaging results are equivocal and/or when a definite diagnosis is required in a younger adult. Gene linkage analysis can be used to determine obligate ADPKD gene carriers, but this method has not gained widespread clinical use and has many practical limitations.

In the present case, CT thorax done to investigate the cause of persistent air leak provided the clue to diagnosis by demonstration of lung and liver cysts. The patient also had multiple cysts in kidneys, and the left ovary with a berry aneurysm in the brain which fits into clinical criteria necessary for the diagnosis of ADPKD. The patient's hypertensive state was unlikely related to her polycystic kidney disease. In ADPKD, the incidence of pulmonary cysts is very rare. In published cases of ADPKD, pneumothorax as the clinical manifestation due to the rupture of underlying lung cysts has rarely been reported till date.

Due to the multiplicity of lung cysts, the possibility of recurrent episodes of pneumothoraces and role of pleurodesis was explained to the patient. The patient and her family members were counselled regarding the disease and its complications and were advised to undergo screening. She was subsequently discharged at request and advised to report if symptomatic.

REFERENCES

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