Unilateral and segmental localised polycystic kidney disease

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Abstract

Unilateral and segmental localised polycystic disease is a rare type of cystic disease of the kidney. It takes the form of a segmental cystic abnormality in one kidney morphologically identical to the autosomal dominant adult form of polycystic kidney disease. The clinical, radiological, and pathological appearances of a case are described. The differential diagnosis and a possible pathogenic mechanism are discussed.

Keywords: kidney; polycystic disease; localised polycystic disease

Case report

A 45 year old male presented with left abdominal and loin pain associated with sterile pyuria. Tenderness in the left upper abdominal quadrant was elicited by clinical examination. An abdominal ultrasound showed a multicystic mass at the lower pole of the right kidney, the

Figure 1 (A) Computed tomography through the level of the lower poles of the kidneys. Note the multicystic abnormality on the right (arrowed). (B) A scan at the level of the upper poles shows normal renal parenchyma on both sides.
left kidney being normal. Abdominal computed tomography confirmed the ultrasound findings (fig 1). The radiological appearances were interpreted as a renal cell carcinoma with cystic degeneration, with a differential diagnosis of a multilocular cystic nephroma being thought less likely. The disparity between the side of symptoms and the lesion in the right kidney was never explained. It is possible the cystic abnormality was asymptomatic.

Past medical history was of ischaemic heart disease including a myocardial infarct at the age of 33 years, type IV hyperlipidaemia, and a hiatus hernia.

There was no family history of renal disease.

A right nephrectomy was carried out and postoperative recovery was uneventful.

**Pathological examination**
The kidney measured 13.5 × 8.5 × 5 cm and weighed 147 g. At the lower pole there was a multicycstic abnormality measuring 8.5 × 5.2 × 5 cm. The cysts ranged from 0.3 to 2.0 cm in maximum dimension and many contained altered blood. No solid or papillary areas were seen and the cysts abutted on the underlying collecting system.

A single isolated cyst, 0.7 cm across, was present at the equator of the kidney but the remainder of the kidney was macroscopically normal.

Microscopically the cysts were lined largely by a simple low cuboidal epithelium (fig 2A) with occasional foci of oncocytic change. There were no papillary areas. Haemorrhage into the cysts was confirmed, with an inflammatory reaction in which macrophages were prominent. The interstitial tissues between the cysts contained compressed glomeruli and tubules (fig 2B). The tubules were mature and there was no loose mesenchymatous tissue or evidence of cartilaginous metaplasia. No blastema or immature fetal elements were identified.

The cystic area was not encapsulated.

The separate simple cyst was lined by low cuboidal epithelium.

The renal parenchyma elsewhere was normal apart from mild intimal fibrosis within interlobular arteries.

**Discussion**
Many different forms of cystic disease of the kidney exist ranging from simple cysts of no clinical significance to congenital abnormalities incompatible with life.

Excluding multiple simple cysts localised segmental forms of cystic disease are unusual. The differential diagnosis of such abnormalities includes multilocular cystic nephroma, cystic partially differentiated Wilm’s tumour,
segmental cystic dysplasia, and unilateral and segmental so-called localised polycystic kidney disease.

Multilocular cystic nephroma shows many similarities to this case but the presence of mature nephron elements in the septa between cysts precludes this diagnosis.\(^1\) The cysts of partially differentiated Wilm's tumour again are similar but the absence of blastema and immature fetal elements is incompatible with this entity.\(^2\) In segmental cystic dysplasia the nephrons in the septa between cysts are rarely fully formed and are usually associated with metaplastic elements such as cartilage. Despite extensive sampling no such elements were found in this case. Although excluding multiple simple cysts is difficult, the extreme clustering of cysts in this case, taken together with their microscopic appearances, indicates a diagnosis of unilateral segmental localised polycystic kidney disease.

Unilateral segmental localised polycystic kidney disease is a rare condition, with only six cases reported.\(^3\)\(^-\)\(^6\) The age range of these cases was 3 to 59 years, and there were four males and two females. They presented with one or more of haematuria, proteinuria, flank/back pain, palpable renal mass, or renal abnormality on x-ray. The collection of cysts involved up to 75% of the kidneys, could be found anywhere within the kidney, and could extend to the collecting system. The cysts varied in size and were lined by flattened/low cuboidal epithelium. Foci of epithelial cell multilayering were identified up to 10 cells thick. Mature nephron elements were seen in the septa between cysts but no immature or metaplastic changes were present. Haemorrhage into cysts was a common feature accompanied by haemosiderin deposition. No epithelial atypia was seen. One case had an accompanying solitary simple cyst.

The case we describe shares many of these features. There was no family history of renal disease. This is an important lesion to diagnose as there is no evidence for progressive renal disease, and simple surgical resection is curative.

The pathogenesis of this form of cystic renal disease is unknown. In view of the morphological similarity of the cystic change to the autosomal dominant form of polycystic kidney disease it is tempting to speculate that the pathogenesis of the cysts is the same. This would require a somatic mutation within the autosomal dominant polycystic kidney gene in the affected segment of the kidney. The germ cells would not be affected and thus there would be no hereditary element. Such a mechanism would also be consistent with the absence of any of the other manifestations of autosomal dominant polycystic kidney disease such as hepatic cysts. As one of the previously reported cases occurred in a three year old, the potential rate of cyst formation is faster than that usually observed in the autosomal dominant form where cysts are not detected until approximately 20 years of age.

We describe a case of unilateral and segmental polycystic kidney disease. This condition has to be distinguished from other localised forms of renal cystic disease and from cystic change in renal tumours. While this condition is morphologically identical to the autosomal dominant form of polycystic kidney disease it is not inherited and has no significance for further renal function.

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