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Unilateral autosomal dominant polycystic kidney disease with co-existent renal cell carcinoma: A rare entity

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ABSTRACT

Bilateral ADPKD is a well-known entity, but there are only a few reports on unilateral ADPKD in adults, most of which had associated contralateral agenesis. Further rare is the development of RCC in unilateral ADPKD. We present an exceedingly rare case of true unilateral ADPKD with normal contralateral kidney and an associated renal cell carcinoma in the same kidney.

1. Introduction

ADPKD is the most common cystic disease of kidney [8]. Bilateral multiple renal cysts of variable sizes, often accompanied by cyst formation in liver, spleen, and pancreas are its characteristic features. Although the disease is usually bilateral, renal involvement may occasionally be asynchronous and asymmetrical [1]. The risk of renal cell carcinoma in patient with ADPKD is reported to be low at < 1% [2].

Unilateral cystic kidney disease often poses diagnostic confusion as more sinister cystic neoplasm needs to be excluded. ADPKD presenting with unilateral cysts has been seen in children [3–5] but in adults, unilateral presentation is quite rare. Most of the reported adult ADPKD cases either had contralateral renal agenesis [1,6] or nephrectomy [7], and to the best of our knowledge no case of unilateral ADPKD with normal contralateral kidney and positive family history has been reported.

We report one interesting case, unilateral ADPKD with associated renal cell carcinoma, a unique presentation not reported so far. The clinical, radiological, and pathological appearances of the case are described; also the differential diagnoses and the possible pathogenic mechanisms are discussed.

2. Case report

A 45-year-old female patient presented to the outpatient department of our hospital with the right hypochondriac pain of 4 months duration with occasional episodes of painless hematuria. There was a history of low grade fever for past 2 months with some weight loss. Patient was known to be asthmatic and chronic smoker.

Her general physical examination was unremarkable except mild pallor. Respiratory examination revealed mild generalized wheeze on auscultation; cardiovascular and neurological systems being normal.

On per abdominal examination, fullness was noted in the right hypochondrium and the renal angle was dull on percussion. A firm non-tender mass, extending several centimeters below right costal margin was palpable in the right renal area.

Routine laboratory investigations were normal except a hemoglobin level of 7 gm%; there was microcytic hypochromic pattern of peripheral smear examination.

Abdominal sonography revealed that the right kidney was enlarged (18.5 × 12.1 cm) and was replaced by innumerable, thin walled anechoic cysts (Fig. 1A). These cysts were of varying sizes and were non-communicating. The kidney was maintaining its reniform shape and was seen crossing the midline. In addition, there was a well-defined hypoechoic solid mass measuring 6.0 × 5.2 cm in mid-lower pole region of this kidney. The mass showed internal vascularity on Doppler imaging (Fig. 2).

Left kidney was normal in size, shape and echo texture (Fig. 1B). Rest of abdominal solid organs, urinary bladder and bowel loops were unremarkable.

Contrast enhanced computed tomography confirmed the ultrasound findings of multiple renal cysts (Fig. 3). No intra-cystic calcification, hemorrhage or wall enhancement was seen. There was stretched out normal functional renal parenchyma tissue in between the cysts. There was excretion of contrast into the pelvi-calyceal system, which appeared distorted by the cysts. The renal mass (6.3 × 5.7 × 6.1 cm) at the mid pole of right kidney was exophytic from renal outline and was seen to focally infiltrate into the anterior abdominal wall. It was

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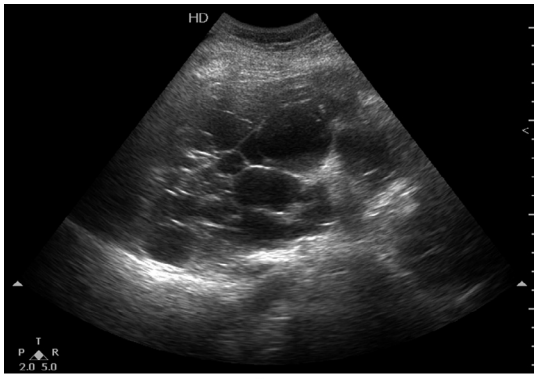

A

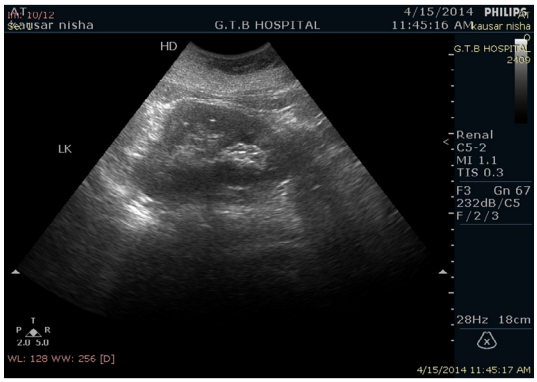

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Fig. 1. Ultrasound image showing: (A) An enlarged right kidney replaced with variably sized cysts. (B) Normal contralateral left kidney.

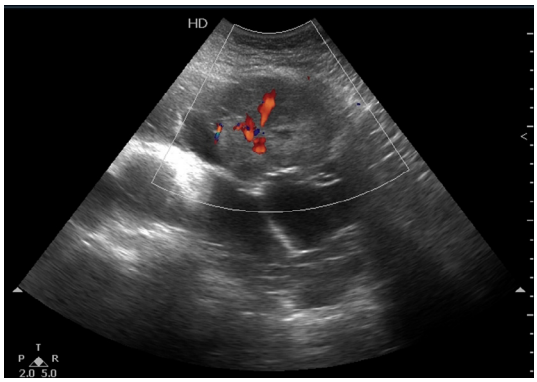

Fig. 2. Ultrasound showing a well defined mass lesion in right kidney with internal vascularity.

heterogeneously hypodense on plain scans and on contrast administration, showed moderate enhancement with central non enhancing areas, suggestive of necrosis (Fig. 4). No vascular invasion or lymphadenopathy was seen. Left kidney, liver, spleen and pancreas were normal. No cysts were seen within them.

The imaging differentials in a case of unilateral cystic kidney with solid mass, as seen in the present case, are unilateral renal cystic disease with malignancy, cystic nephroma, multicystic dysplastic kidney, cystic renal cell carcinoma.

The presence of functional parenchyma and opacified distorted calyces in between the cysts, which indicated that the cysts were separate and not part of a cystic mass, helped us in excluding the possibilities of cystic nephroma, multicystic dysplastic kidney and cystic renal cell carcinoma.

Thus, a working diagnosis of unilateral cystic renal disease with

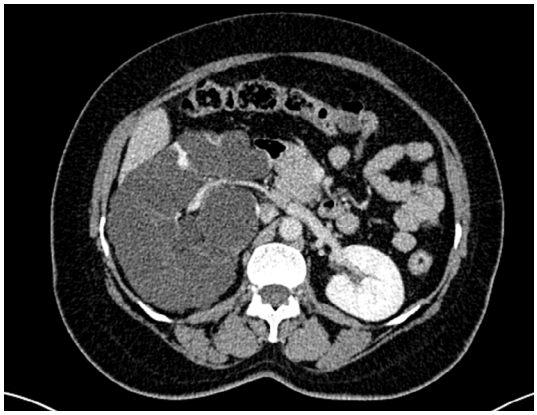

Fig. 3. Axial CECT image showing an enlarged right kidney with multiple cysts and a normal left kidney.

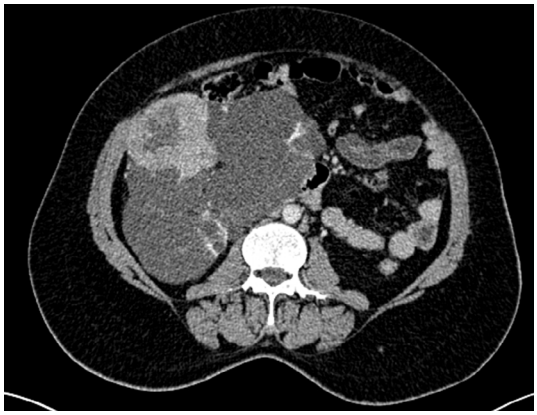

Fig. 4. Axial CECT image showing a well-defined, heterogeneously enhancing mass in the polycystic right kidney.

malignancy was considered. The possibility of unilateral polycystic kidney disease was also considered and patient's family was called for screening. There was frank bilateral ADPKD in patient's daughter. On questioning, it was also disclosed by the patient that her mother had died of some kidney disease, though no records were available with them.

Based on imaging appearances and patient's family history, a confident diagnosis of unilateral ADPKD with secondary malignancy was kept. Diagnostic ultrasound guided aspiration cytology was positive for malignant cells.

The patient underwent unilateral radical nephrectomy. She had an uneventful post-operative period and was normal at one year follow up. No evidence of recurrence was seen.

The nephrectomy specimen was sent for histopathology. The cut surface of the same revealed multiple cysts of varying sizes with translucent walls along with a 7 × 5 cm solitary gray white area.

On microscopy, the cysts were found to be lined with a single layer of cuboidal epithelium with hyperplastic epithelial cells focally (Fig. 5). The section from solid tumor tissue revealed cells with moderate anisonucleosis, hyperchromatism and inconspicuous nucleoli, arranged in a papillary-trabecular, papillary solid pattern with fibro vascular core (Fig. 6). There were few areas of hemorrhage and necrosis. A histopathological diagnosis of papillary renal cell carcinoma (type 2) in the background of polycystic kidney was made.

3. Discussion

ADPKD is a common hereditary disorder, with autosomal dominant inheritance, seen in 1 in 500 to 1 in 1000 live births and in 10 – 20%