

Unilateral Autosomal Dominant Polycystic Kidney Disease with Contralateral Renal Agenesis : A Case Report

Autosomal dominant polycystic kidney disease (ADPKD) is the most common hereditary renal disease. There are some reports in the literature concerning unilateral ADPKD. However, in adults, only a few cases of unilateral ADPKD with agenesis of contralateral kidney have been reported. We present a case of unilateral ADPKD with agenesis of contralateral kidney in a 66-yr-old man. Radiographic images showed the enlarged right kidney with multiple variable-sized cysts and the absence of the left kidney. The diagnosis of ADPKD was confirmed by the family screening. The patient received maintenance hemodialysis for end-stage renal disease. We report a case of unilateral ADPKD associated with contralateral renal agenesis in a 66-yr-old male patient with a literature review.

Key Words : Polycystic Kidney, Autosomal Dominant, Organogenesis, Renal Dialysis

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Received : 8 April 2002
Accepted : 10 June 2002

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INTRODUCTION

Autosomal dominant polycystic kidney disease (ADPKD) is characterized by progressive replacement of tissue by bilateral multiple renal cysts, often accompanied by cyst formation in the liver and pancreas. Although the disease is bilateral, renal involvement may be asynchronous and asymmetric. There are some reports on unilateral ADPKD in the literature (1-4). Only two cases of unilateral ADPKD with contralateral renal agenesis have been reported in the literatures (5, 6).

To our best knowledge, this is the first reported case of unilateral ADPKD with agenesis of contralateral kidney in Korea.

CASE REPORT

A 66-yr-old male patient was admitted to our hospital because of gross hematuria. One year prior to admission, the patient developed general weakness. He had been taking herb medicine intermittently. Ten years earlier, he was found to have hypertension. But he had not received any treatment. Other past medical history including diabetes mellitus, tuberculosis, or abdominal surgery had not been known. He smoked 20 cigarettes/day for 40 yr, and had moderate alcohol intake. His blood pressure was 190/100 mmHg. On physical exami-

nation, a large flank mass was palpable several centimeters below the right costal margin and his conjunctiva was pale. Auscultation of his chest was normal. Family screening was performed subsequently. His parent were dead earlier, and cause of death was not known. He had no sibling. Ultrasound studies were performed on his two sons and a daughter. One of the patient's two sons and a daughter had polycystic renal disease (Fig. 1). Family illness including renal failure or cerebrovascular accident were not observed.

Urinalysis showed hematuria (RBC 50-99/HPF) and proteinuria (protein 100 mg/dL). Biochemical analysis revealed hemoglobin level of 7.2 g/dL, white blood cell count 4,700/ μ L, platelet count 162,000/ μ L, blood urea nitrogen level of 69.6 mg/dL, and serum creatinine level of 7.0 mg/dL, sodium 133 mEq/L, potassium 5.2 mEq/L, total calcium 3.6 mEq/L, ionized calcium 2.0 mEq/L, aspartate aminotransferase 25 IU/L, alkaline phosphatase 59 IU/L, serum albumin 3.5 g/dL, and C-reactive protein (CRP) 0.46 mg/dL. Creatinine clearance rate was 5.71 mL/min/1.73 m², and normalized protein nitrogen appearance (nPNA) was 0.52 g/day. Chest radiography and electrocardiography (ECG) were normal findings.

Abdominal sonography revealed innumerable, variable-sized cysts in the right kidney and absence of the left kidney (Fig. 2). Abdominal CT revealed enlargement of the right kidney (16 × 14 × 10 cm) with multiple, variable-sized nonenhancing cystic lesions and the left kidney was not observed. Multi-

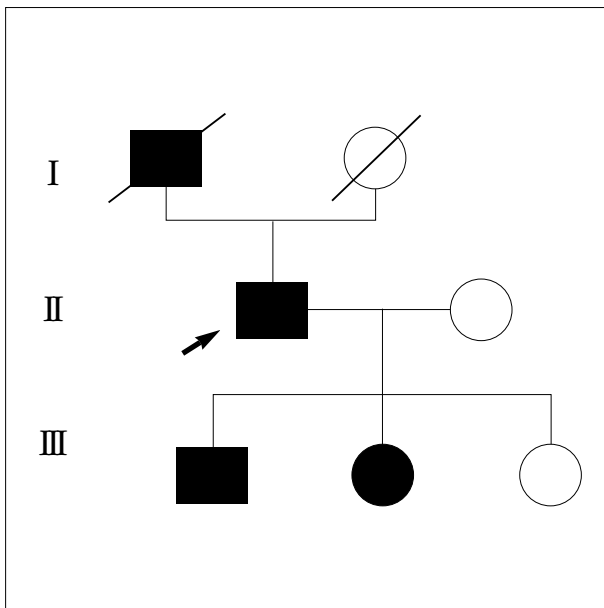


Fig. 1. Pedigree of family (square: male, circle: female, affected subject: filled symbol, affected proband: arrow).

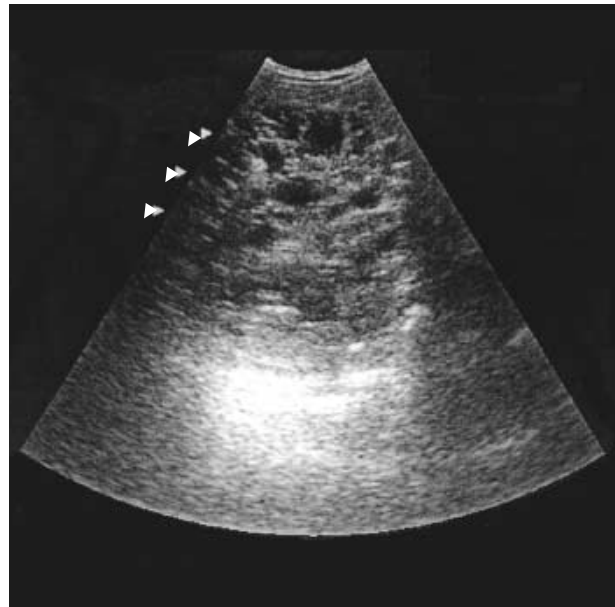


Fig. 2. Abdominal US shows the enlarged right kidney filled with innumerable, variable-sized cysts and the absence of the left kidney.



Fig. 3. Abdominal CT scan reveals the enlarged right kidney with multiple, variable-sized nonenhancing cystic lesions and the absence of renal tissue in the left side on all levels of scanning (A: L1 level, B: L3 level). Also note multiple cysts in the liver and pancreas. Left adrenal gland is indicated by arrow.

ple nonenhancing cysts and a single cyst were also noted in the liver and pancreas body, respectively (Fig. 3). ^{99m}Tc -DMSA scan demonstrated no activity of the left kidney (Fig. 4). He has received chronic maintenance hemodialysis for the end-stage renal disease (ESRD).

DISCUSSION

We described an extremely rare case of unilateral ADPKD with contralateral renal agenesis. In 1974, Bear (5) reported 2 cases of ADPKD with solitary kidney. First case was 48-yr-old man of unilateral ADPKD with agenesis of the con-

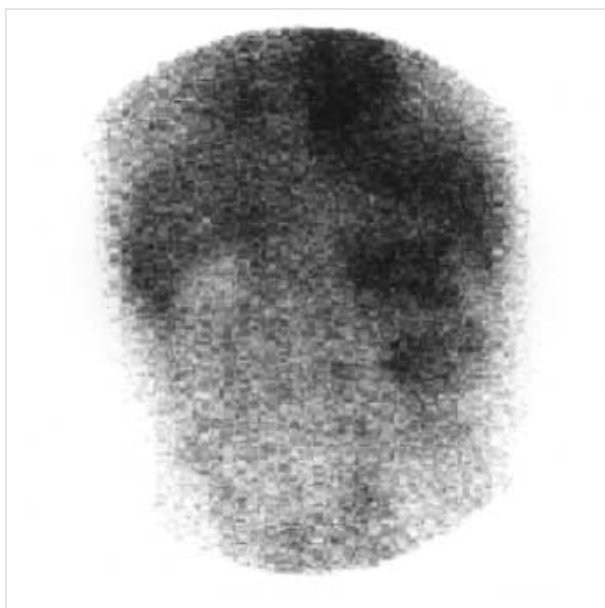


Fig. 4. 99mTc-DMSA demonstrates no radioactivity of the left kidney.

tralateral kidney. His father died of hypertension and renal failure, and autopsy revealed bilateral polycystic kidney disease. An excretory urogram (IVP) revealed absence of the one kidney and early changes of polycystic disease in the contralateral kidney. He was planned renal transplantation for the ESRD. Second case was ADPKD with contralateral nephrectomized kidney. Therefore, pure case of ADPKD with renal agenesis was only one. In 1999, Todorov (6) reported another case of unilateral ADPKD with agenesis of the contralateral kidney. The patient was a 45-yr-old woman, and her mother suffered from polycystic kidney disease and also found in one sibling by ultrasonography. She was confirmed the absence of one kidney and typical picture of ADPKD by using CT.

ADPKD is morphologically indistinguishable from unilateral renal cystic disease (URCD). Levine et al. (7), stated that URCD had at least three different aspects from ADPKD, that is unilateral localization, negative family history, and no progression to chronic renal failure. URCD patients usually have no cysts in other intra-abdominal organs (8, 9). ADPKD is an inherited disorder, transmitted in an autosomal dominant pattern, with 100% penetrance but variable expression. Overall, 50% of ADPKD patients have reached an ESRD by the ages of 57 to 73 yr (1-4). The mechanism by which ADPKD causes renal failure is not completely

understood.

The diagnosis of unilateral ADPKD is made by family history and radiological studies. If there is no family history of ADPKD, the differential diagnosis includes hydronephrosis, multicystic dysplastic kidney, cystic nephroma, and cystic Wilm's tumor. In the past, most of the nephrectomies in children with unilateral polycystic kidney disease were performed because of concern of a Wilm's tumor. However ultrasonography and CT can usually differentiate polycystic kidney disease from Wilm's tumor, avoiding unnecessary surgery (10, 11).

Management may vary depending on the renal function and presence of other complications. Our patient has received maintenance hemodialysis for ESRD and gross hematuria was resolved spontaneously by resting.

In summary, to our best knowledge, the present case is the first report of a unilateral ADPKD with contralateral renal agenesis in Korean.

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