Polycystic horseshoe kidney

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Introduction

Polycystic horseshoe kidney is thought to represent two separate renal diseases. Horseshoe kidney is a renal fusion anomaly during embryogenesis; autosomal dominant polycystic kidney disease (ADPKD) is a hereditary disorder due to mutations in the genes responsible for the expression of the proteins polycystin 1 (ADPKD1) and polycystin 2 (ADPKD2). Polycystic horseshoe kidney is a very rare occurrence with incidence ranges of 1 in 134,000 to 1 in 8,000,000 live births; about 20 cases have been reported in the literature.

Case

A 32-year-old woman was admitted to hospital complaining of increased belly volume. At physical examination, a large abdominal mass was found. An abdominal ultrasonography showed enlarged polycystic horseshoe kidney and liver cysts. Computerized tomography revealed enlarged kidneys with multiple cysts, fused by their lower poles (Figure 1) and multiple liver cysts (Figure 2). Her parents and four brothers were screened for ADPKD with renal ultrasound; no cases were found, although one brother was shown to have horseshoe kidney. She had been hypertensive since her first pregnancy at age 15 (she gave birth to three daughters, all with documented ADPKD) and at admission her renal function was noticed to be abnormal (serum creatinine of 2.1 mg/dl; creatinine clearance of 43 ml/min/1.73 m²).

Discussion

Adult polycystic kidney disease is a hereditary disorder characterized by accumulation of fluid-filled cysts in the kidney and other organs. It should be regarded as a systemic disease, as the genes responsible (PKD1 on chromosome 16 and PKD2 on chromosome 4) are widely expressed in extrarenal tissues and its encoded proteins, polycystin-1 and polycystin-2, respectively, are associated to cyst production, cerebral and aortic aneurysms, cardiac valvular abnormalities and colonic diverticuli, among other manifestations. It affects all ethnic groups worldwide with incidences ranging from 1 in 500 to 1 in 5000 births. About 5% of the reported cases of APKD occur without previous family history and probably represent incident mutation giving rise to a new cohort, as probably is the case of the patient reported here [1].

Horseshoe kidney is probably the most common of all renal fusion anomalies, occurring in about 0.25% of the population. It is usually asymptomatic, but can be associated with renal stones, ureteropelvic junction syndrome [2] and renovascular hypertension [3].

To date, no genetic association has been described between the PKD loci and horseshoe kidneys.

Conflict of interest statement. None declared.

References


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Fig. 1. Computerized abdominal tomography showing the patient’s enlarged, containing multiple cysts, horseshoe kidney.

Fig. 2. Computerized tomography of patient’s liver depicting inumerous cysts (polycystic liver).