Autosomal recessive polycystic kidney disease: a case description and early urologic management

García-de León Gómez José Manuel,1 Navarro-González Alfonso,2 Hernández-Valdés María Guadalupe,3 Aguirre-Ramírez Pedro.4

ABSTRACT

Polycystic kidney disease is a pathology that affects both kidneys symmetrically. It is usually diagnosed by obstetric ultrasound or at birth and is the result of a specific mutation of a gene on chromosome 16. This disease has a low incidence of 1 in 20 000 births and represents 4.24% of neonatal abdominal masses and 6.4% of all renal masses at this stage of life. It usually manifests as hard echogenic giant kidneys.

The case is presented of a neonate with antenatal detection of autosomal recessive polycystic disease and renal failure, treated in the first days of life with left nephrectomy and then, twelve months later, with right nephrectomy and kidney transplantation.

The neonatal patient was attended to at our hospital and had a past medical history of prenatal ultrasound diagnosis of large, echogenic kidneys and oligohydramnios, with no evidence of hydronephrosis. After multidisciplinary

RESUMEN

La enfermedad renal poliquística (ERPI) es un padecimiento que afecta ambos riñones, de manera simétrica. Usualmente es diagnosticada mediante ultrasonido obstétrico o al nacimiento, como resultado de una mutación específica de un gen del cromosoma 16. Esta enfermedad tiene una baja incidencia 1 en 20 000 nacimientos, representa el 4.24% de las masas abdominales del recién nacido, y 6.4% de todas las masas de origen renal en esta etapa. Frecuentemente, se manifiesta como riñones gigantes duros ecogénicos.

Se presenta el caso de un recién nacido, con detección antenatal de enfermedad poliquística infantil e insuficiencia renal, tratado en los primeros días de vida con nefrectomía izquierda y 12 meses después, trasplante renal y nefrectomía del riñón derecho.

Recién nacido atendido en nuestro Hospital, con antecedente de diagnóstico antenatal mediante ultrasonido de
INTRODUCTION

Autosomal recessive polycystic kidney disease (ARPKD) involves both kidneys, symmetrically, and is transmitted by autosomal recessive inheritance. It is generally diagnosed in utero or at birth and occurs as the result of a genetic mutation of the PKD1 genes that are located on chromosome 16.

It is a rare disease and its incidence is estimated to be 1 in every 20,000 births. It represents 4.24% of abdominal masses in neonates and 6.4% of neonatal masses of renal origin.

It is characterized by a fusiform dilation of renal collecting tubes and is frequently associated with hepatic fibrosis.

The disease has a wide presentation spectrum, and studies indicate that the abnormality occurs in just one locule, which is responsible for all the phenotypes.

Prenatal ultrasound usually shows very large, echogenic kidneys, along with oligohydramnios that is the result of a reduction of fetal diuresis.

Symptoms at birth can include abdominal tumors on both sides, renal failure, or both.

Death occurs in 25-30% of affected neonates due to respiratory failure. Close to 50% of individuals born with this disease survive, but progress to kidney failure within the first decade of life. The patients that survive may have concomitant diseases such as high blood pressure, portal hypertension, and hepatic fibrosis.

CASE PRESENTATION

The patient is a male neonate with antenatal diagnosis of oligohydramnios and enlarged kidneys with no hydrenephrosis, attended to in the intensive care unit for respiratory failure and for presenting with solid, giant, palpable abdominal tumors. The infant was unable to eat due to the significant respiratory and digestive restrictions secondary to abdominal mass compression. The clinical case was jointly presented to the departments of nephrology, neonatology, transplants, and urology and the decision was made to remove the left kidney in order to improve ventilation and to decompress the

Keywords: Renal transplantation, autosomal recessive, polycystic kidney, Mexico.

Palabras clave: Trasplante renal, autosómica recesiva, riñón poliquístico, México.
gastric chamber and facilitate oral feeding, despite the fact that the patient already presented with moderate renal failure.

Surgery revealed a solid, giant kidney with a diameter larger than 13 cm and histopathologic report confirmed autosomal recessive polycystic kidney disease (ARPKD) (Figure 1).

Medical management was carried out by the department of nephrology and renal failure continued to progress. The patient also presented with high blood pressure that was managed with medications. At the age of one year, the patient, then weighing 8 kilograms, required substitutive treatment and so the decision was made to perform a living related kidney transplantation. At the moment of transplantation, right kidney nephrectomy was carried out to place the graft on the right side, performing anastomosis to the aorta and to the inferior vena cava (Figures 2 to 5).

Patient progression is satisfactory at one year from the transplantation, with good function of the kidney graft. High blood pressure disappeared at the moment the right native kidney was removed in the transplantation.

**DISCUSSION**

Autosomal recessive polycystic kidney disease (ARPKD) is hereditary and its outstanding characteristic is numerous fluid-filled microcysts that arise from the kidney tubules. It is the most frequent genetic cause of terminal kidney failure in fetuses and neonates. It is a recessive autosomal disorder located on chromosome 16 that is the result of mutations of the PDK1 (found in locus 16p13.3-p13.12) and PKD2 (4q21-q23) genes that encode the fibrocystin and polycystin proteins. Parents that do not present with ARPKD can have a child with this disease if each one carries an abnormal gene and both genes are transmitted to the child. If only one of the parents has the abnormal gene, the child will not inherit the disease.

There are 4 types of ARPKD: In type I, or perinatal disease, there is serious kidney lesion, symmetric nephromegaly, early death due to acute respiratory failure, secondary pulmonary hypoplasia, and Potter’s syndrome. Type II, or neonatal disease, is the same as type I except that death is caused by renal failure. Type III, or infantile disease, corresponds to the case presented herein. And type IV, or juvenile disease, is associated with congenital hepatic fibrosis and portal hypertension.
In ARPKD, the two kidneys are enlarged due to microcysts in the cortex and medulla that represent dilations of the collecting tubules, interstitial fibrosis, and tubular atrophy and are often associated with hepatic fibrosis.

Unilateral or bilateral nephrectomy in the neonate with giant polycystic kidneys that restrict respiratory and gastrointestinal function, is indicated as palliative treatment in selected cases. 1-3-5

In the present case, left nephrectomy improved respiratory function and allowed food intake, and therefore the survival of the neonate. At the age of one year, the patient had 15% renal function and so it was decided to perform a living relative transplant, before dialysis was required. Transplantation was carried out when the patient weighed 8 kilograms and was one year old. One year after the transplant, the kidney graft is functioning well.

We consider aggressive neonatal management of unilateral or bilateral nephrectomy in patients with solid, giant, polycystic kidneys to be recommendable in order to facilitate respiratory and digestive function and to allow early renal transplantation. In addition nephrectomy usually reverts the high blood pressure that is always present in these patients. In the present case we could observe the comparative size of the adult living donor kidney graft and the right native kidney removed from the patient at the moment of transplantation (Figure 6).

REFERENCES