Autosomal recessive polycystic kidney disease (ARPKD) affects the liver and the kidney. Renal involvement presents early in life, whereas hepatic involvement manifests slightly later with portal hypertension. A male toddler came with chronic abdominal distension, prominent abdominal wall vessels, and umbilical protuberance. Ultrasonography findings of hepatic fibrosis with portal hypertension, mildly prominent biliary radicals, bilateral cystic renal enlargement, and a striated nephrogram on contrast-enhanced computed tomography suggested the diagnosis. Congenital hepatic fibrosis is invariably associated with ARPKD leading to portal hypertension and the development of portosystemic collaterals; those located in the umbilical region appear as caput medusae.

**Keywords:** Autosomal recessive polycystic kidney disease, caput medusae, computed tomography, portal hypertension, ultrasonography

**INTRODUCTION**

Autosomal recessive polycystic kidney disease (ARPKD) is the most common heritable cystic renal disease. The gene attributed to this condition is PKHD1. It presents during perinatal period and childhood. There is no gender predilection. The reported incidence is in between 1 in 6000 and 1 in 55,000 births.

**CASE REPORT**

An 1-year and 9-month-old male child presented to the pediatrician with symptoms of progressive abdominal swelling over 6 months associated with swelling in the umbilical region. On examination, the prominent superficial veins were seen over the abdomen wall with a tuft of vessels in the umbilicus giving the appearance of caput medusae [Figure 1]. There was no history of hematemesis or melena. Blood pressure was normal. The parents gave the history of consanguineous marriage. His first sibling had died at 4 years of age due to a similar illness. He has one more normal living sibling. Both the parents and the living sibling were screened by ultrasonography (USG), which was normal.

The biochemical investigations showed, normal liver function tests: aspartate transaminase – 52 U/L, alanine transaminase – 26 U/L, alkaline phosphatase – 162 U/L, albumin – 3.2 g/dl, globulin – 4.1 mg/dl, total bilirubin – 0.4 mg/dL, and direct bilirubin – 0.1 mg/dL. The renal function tests are within normal limits: urea – 28 mg/dl and creatinine – 0.6 mg/dl. The hemoglobin level was reduced (6.7 g/dl). The patient was given blood transfusion.

The patient was referred to the Department of Radiology for imaging. USG showed mild hepatomegaly with regular hepatic outlines. There were prominent portal triads with increased periportal echogenicity [Figure 2a] and mild prominent biliary radicals [Figure 2a and b]. Color Doppler USG showed dilated recanalized paraumbilical vein [Figure 3a] in the falciform ligament connecting the left portal vein with a cluster of dilated venous channels in the umbilicus [Figure 3b] suggestive of portosystemic shunting. There was mild splenomegaly. Both the kidneys were enlarged and normal in shape. Parenchyma was hypeerechoic with numerous small anechoic cysts in the cortex as well as medulla [Figure 4a].

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Contrast-enhanced computed tomography of the abdomen was performed, which showed bilateral nephromegaly with maintained reniform shape, multiple small renal cysts, and classical striated nephrogram [Figure 4b]. Multiplanar reformatted image demonstrated the entire course of recanalized left paraumbilical vein [Figure 3c]. Three-dimensional volume-rendered image showed prominent abdominal vessels with umbilical caput medusa [Figure 3d]. A molecular genetic testing was advised for the patient, and the parents were referred for genetic counseling. An upper GI endoscopy was advised to look for gastroesophageal varices. However, the patient was lost to follow-up.

**Discussion**

ARPKD affects the kidneys and the liver to a variable degree. The severity of hepatic and renal involvement varies inversely. Renal involvement tends to occur in younger age, and hepatic involvement manifests in older children. The underlying pathology in the kidney is renal tubular ectasia and interstitial fibrosis. ARPKD inevitably presents with congenital hepatic fibrosis that gradually leads to the development of portal hypertension. Hepatic involvement comprises of ductal plate malformation leading to abnormal proliferation and dilatation of bile ducts and fibrosis of portal tracts. Historically, childhood PKD was classified into four types, namely perinatal, neonatal, infantile, and juvenile forms using clinical and histologic findings. The most severe form of renal disease presents in the perinatal period. It can be diagnosed in utero by sonography. Grossly impaired renal function causes severe oligohydramnios.

*Figure 1:* Distended abdomen with prominent superficial veins and tuft of vessels in the umbilical region suggestive of caput medusae

*Figure 2:* (a) High-resolution ultrasound of liver showing echogenic portal triads (short arrow) and prominent tubular anechoic biliary radicals (long arrow). (b) Ultrasonography of liver showing prominent tubular anechoic biliary radicals (arrow)

*Figure 3:* (a) Color Doppler ultrasonography of liver showing dilated recanalized left paraumbilical vein in the falciform ligament. (b) Color Doppler ultrasonography of the umbilical swelling shows a cluster of dilated veins suggestive of caput medusae. (c) Contrast-enhanced computed tomography of abdomen with multiplanar reformatted image showing the entire course of the paraumbilical vein from liver (short arrows) up to the umbilicus (long arrow). (d) Three-dimensional volume-rendered image showing prominent abdominal wall vessels and caput medusae

*Figure 4:* (a) Ultrasonography of kidney shows anechoic cystic tubular structures suggestive of renal tubular ectasia. (b) Coronal contrast-enhanced computed tomography of the abdomen shows symmetrically enlarged bilateral kidneys with hypodense cysts and striated nephrogram
Enlarged kidneys compress the thoracic cavities with pulmonary hypoplasia, which is the cause of perinatal mortality. Patients with milder renal involvement survive the perinatal period and present with renal impairment during the later part of infancy. Renal impairment presents with oliguria, azotemia, and hypertension. Gradually, the patients land up with end-stage renal disease requiring frequent dialysis and renal transplantation.[1]

On USG, both kidneys appear enlarged with maintained reniform shape. The renal parenchyma appears hypechoic due to many interfaces between the radially arranged dilated anechoic ducts and the ultrasound beam. Corticomедullary differentiation is lost in later stages, but a thin rim of the preserved cortex is seen in the milder form. Multiple small cysts are seen in the renal parenchyma. The maximum diameter of cysts does not exceed 1.5 cm.[1,9]

CT scan shows enlarged kidneys with multiple cysts and a striated nephrogram due to the accumulation of contrast in dilated tubules.[1,10]

Patients with very mild renal involvement show normal renal function. Patient survival is prolonged due to a milder form of renal disease providing time for the progression of congenital hepatic fibrosis. USG shows increased periportal echogenicity and various degrees of intrahepatic biliary radical dilatation (IHBRD). IHBRD is nonobstructive and due to duct ectasia. The cystic dilated intrahepatic biliary radicals amount to Caroli’s disease. These patients present with features of portal hypertension during first few years of life. Portal hypertension manifests as splenomegaly, ascites, and portosystemic collateral formation in the form of esophageal varices, caput medusae around the umbilicus which is also demonstrated on radiological imaging. The child may present with variceal bleeding needing sclerotherapy or banding.[11]

There is no impairment of hepatocellular function even in advanced hepatic fibrosis due to intact hepatic parenchyma. The other associations to be looked for in Caroli’s syndrome and ARPKD are recurrent cholangitis, hepatolithiasis, liver abscess formation, pancreatitis from biliary calculi, and features of hypersplenism.[12]

The characteristic renal imaging findings with one or more of the following are needed for a specific diagnosis of ARPKD.[2,13]

- Biliary ductal ectasia on imaging
- Clinical or laboratory features of portal hypertension
- Histopathologic demonstration of ductal plate abnormality causing congenital hepatic fibrosis
- Absence of typical imaging findings in both parents on high-resolution USG
- Pathologic or genetic diagnosis of ARPKD in a sibling.

However, the diagnosis of ARPKD is established by identification of abnormal alleles of PKHD1 gene by molecular genetic testing in a suspected patient.

In our case, the diagnosis of autosomal recessive polycystic kidney disease was made based on the characteristic imaging features. History of the death of a sibling due to a similar illness, consanguinity of parents, normal USG of both parents, and one normal living sibling also point toward a genetic cause of the disease.

ARPKD should be thought of in a case of pediatric portal hypertension. Apart from symptomatic treatment of the patient, the other siblings should be screened, and the parents should be offered genetic counseling.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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