Prenatal sonographic appearance of congenital bile duct dilatation associated with renal-hepatic-pancreatic dysplasia

S. BOOPATHY VIJAYARAGHAVAN*, M. KAMALAM† and M. L. RAMAN‡
*Sonoscan, Ultrasonic Scan Centre and ‡G.K.N.M. Hospital, Coimbatore and †Prabu Polyclinic, Pollachi, India

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ABSTRACT
We report the prenatal sonographic features of congenital bile duct dilatation associated with renal-hepatic-pancreatic dysplasia. The condition was seen at 22 weeks of gestation and led to termination of pregnancy. This is the first description of congenital bile duct dilatation using prenatal sonography. It is also the first report of a case in which the features of dysplasia were evident in all three of the organs which may be affected, the kidneys, liver and pancreas. Copyright © 2004 ISUOG. Published by John Wiley & Sons, Ltd.

INTRODUCTION
The combination of renal, hepatic and pancreatic dysplasia (RHPD), also known as Ivemark II syndrome, is very rare and uniformly fatal. We describe the prenatal sonographic appearance of congenital bile duct dilatation associated with this syndrome.

CASE REPORT
A 25-year-old woman, gravida 2 para 1, was referred for sonography at 22 weeks' gestation because the fetus was small-for-gestational age. Her first pregnancy had resulted in miscarriage at around 12 weeks' amenorrhea. There was a history of consanguinity. Sonography was performed using a Philips HDI 5000 (Philips Medical Systems, Bothell, WA, USA) ultrasound machine, which revealed severe oligohydramnios. The head circumference corresponded with a gestational age of 22 weeks, but all the long bones were short, falling below the 5th percentile, and the chest was narrow. Both kidneys showed multiple cysts (Figure 1). An irregular cyst, 12 × 7 mm in diameter, was observed in the upper abdomen anterior to the aorta and inferior vena cava, with its long axis oriented along the transverse plane (Figure 2). The transversely placed irregular cyst was suggestive of a pancreatic cyst. The gall bladder was seen anterior to the cyst and appeared normal. The transverse scan of the liver revealed a tubular structure anterior to the right and left branches of the portal vein, suggestive of dilated right and left hepatic ducts (Figure 3). There was no suggestion of a dilated common duct, and the main portal vein and the hepatic artery could be seen.

The parents chose to undergo termination of pregnancy. The fetus had short limbs, post-axial polydactyly of all four limbs, and a narrow chest. Autopsy revealed multicystic dysplasia of both kidneys (Figure 4) and there was an irregular cyst in the head of the pancreas (Figure 5). A needle was placed in the gall bladder and contrast medium was injected under image intensifier.
control. This revealed a normal gall bladder and common duct. The right and left hepatic ducts and intrahepatic biliary radicles were grossly dilated (Figure 6). This confirmed the sonographic features of dilated hepatic ducts. Histologically, the kidneys showed features of multicystic renal dysplasia, with abortive glomerular, tubular and mesenchymal collarettes around cystically dilated tubules. The pancreas showed dilated pancreatic ducts, forming cystic spaces within the pancreas. The liver showed dilated bile ducts and proliferated ductules with portal-to-portal bridging by fibrous tracts.

DISCUSSION

In 1959 Ivemark et al.\textsuperscript{1} described a syndrome of RHPD in two siblings; their kidneys showed tubular and glomerular dysplasia with small cysts. Their liver and pancreas contained embryonal connective tissue with numerous dilated ducts. In 1978 Crawfurd et al.\textsuperscript{2} described a similar syndrome in two siblings, one of whom had splenic agenesis and cardiac transposition. One year later, Strayer and Kissane\textsuperscript{3} published a sporadic case. In 1987 Bernstein

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**Figure 2** Transverse (a) and longitudinal (b) ultrasound images of the upper abdomen showing the pancreatic cyst (C). A, aorta; GB, gall bladder; I, inferior vena cava.

**Figure 3** Transverse ultrasound image of the liver showing dilated right and left hepatic ducts (arrow) anterior to the right and left branches of the portal vein.

**Figure 4** Autopsy photograph of the bilateral multicystic kidneys.
Bile duct dilatation in renal-hepatic-pancreatic dysplasia

et al.\textsuperscript{4} reviewed this syndrome and reported five unrelated cases of RHPD all of which had bilateral renal cystic dysplasia, biliary dysgenesis, pancreatic fibrosis and cyst formation. They reported biliary ductal dilatation as a feature in four of the five cases. They also emphasized that the patterns of renal, hepatic and pancreatic involvement were indistinguishable from those reported in several other syndromes, including Jeune asphyxiating thoracic dystrophy (JATD) and Ellis–van Creveld syndrome (EVC). They concluded that the diagnosis of RHPD should be one of exclusion and that even when all identifiable syndromes have been excluded, the remaining cases may not constitute a homogenous group.

In our case, prenatal sonography of the fetus showed dilated hepatic ducts anterior to the right and left branches of the portal vein. This finding was confirmed by contrast cholangiography at autopsy. Sonography also showed that the common duct and gall bladder were not dilated. On histology there were no remarkable findings around the junction of dilated and non-dilated ducts. The fetus also showed bilateral multicystic kidneys. There was a transversely placed irregular cyst in the upper abdomen anterior to the aorta and inferior vena cava with brightly echogenic tissue around it, suggestive of a pancreatic cyst. A choledochal cyst can mimic this appearance, but the transverse orientation and irregularity of the pancreatic cyst distinguished it from choledochal cyst. The fetus also showed short long bones, narrow chest and post-axial polydactyly of all four limbs.

To our knowledge this is the first report of the prenatal sonographic features of bile duct dilatation. It is also the first sonographic description of a case in which the features of dysplasia were evident in all three of the organs which may be affected in RHPD, the kidneys, liver and pancreas.

**REFERENCES**