Prenatal Diagnosis of Caroli Disease Associated With Autosomal Recessive Polycystic Kidney Disease by 3-D Ultrasound and Magnetic Resonance Imaging

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Abstract

Background: Caroli disease is a very rare congenital anomaly characterized by non-obstructive saccular or fusiform dilatation of the intrahepatic bile ducts. It is associated with bile stagnation and hepatolithiasis, which explain the recurrent cholangitis and portal hypertension as a consequence of congenital liver fibrosis. Although there are several reports of diagnosis in childhood and adult life, the prenatal diagnosis using conventional 2-D ultrasound is rare, with few reports in the literature.

Case: We present a case of a 26-year-old primigravid woman at 24 weeks of gestation which 3-D ultrasound in the rendering mode clearly revealed the enlarged fetal kidneys and the increased abdominal volume, confirming the diagnosis of autosomal recessive polycystic kidney disease. The MRI was essential to the prenatal diagnosis of Caroli disease, identifying the congenital saccular dilations of intrahepatic bile ducts.

Résumé

Background : La maladie de Caroli est une anomalie congénitale très rare caractérisée par une dilatation sacciforme ou fusiforme non obstructive des voies biliaires intra-hépatiques. Elle est associée à une stagnation de la bile et à une lithiase intra-hépatique, qui expliquent la cholangite récurrente et l'hypertension portale secondaires à la fibrose hépatique congénitale. De nombreux cas ont été diagnostiqués chez des enfants et des adultes, mais le diagnostic prénatal au moyen d'une échographie traditionnelle 2D est rare, d'où le faible nombre de cas documentés.

Case : Nous présentons le cas d'une femme primigeste de 26 ans chez qui une échographie 3D en mode rendu réalisée à 24 semaines de grossesse a permis de détecter, sans équivoque, une hypertrophie des reins fœtaux et une augmentation du volume abdominal fœtal et de confirmer le diagnostic de maladie polykystique des reins autosomique récessive. Une IRM a permis de repérer les dilatations sacciformes congénitales des voies biliaires intra-hépatiques, ce qui a été essentiel au diagnostic prénatal de la maladie de Caroli.

Key Words: Caroli disease, autosomal recessive polycystic kidney disease, 3-D ultrasound, MRI

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INTRODUCTION

In 1958, Jacques Caroli was the first to describe a rare congenital malformation characterized by non-obstructive saccular or fusiform dilatation of the intrahepatic bile ducts.[1] Although there are several reports of diagnosis during the neonatal period and several of others diagnosed in childhood and adult life, the incidence of this malformation is 6,000-40,000 newborns.[2–4] The clinical presentation can range from neonatal renal dysfunction to the onset of recurrent cholangitis in adults.[5] However, although the symptoms are dynamic and progressive, the clinical presentation and progression are highly variable.[6]

The prenatal diagnosis of Caroli disease and polycystic kidney disease was first reported in 1991 by Hussman et al.[7] when liver cysts and polycystic kidney disease were considered a spectrum of the same disorder with a possible genetic relation. In 1997, Torra et al.[8] indicated the presence of an association between ductal plate malformation

and autosomal recessive polycystic kidney disease (ARPKD) during embryogenesis related to a mutation in the PKD1 gene.

To the best of our knowledge, there are no reports of the prenatal diagnosis of Caroli disease using 3-D ultrasound or MRI. We present a prenatal diagnosis of Caroli disease associated with ARPKD using 3-D ultrasound and MRI in a pregnant woman at 24 weeks of gestation.

THE CASE

A 26-year-old primigravid woman at 24 weeks of gestation was referred to our service with the prenatal diagnosis of hepatosplenomegaly and oligohydramnios. During the fetal morphological study by 2-D ultrasound examination, an enlarged liver was noted (Figure 1). Saccular dilatations were observed in the liver parenchyma toward the porta hepatis. Color Doppler did not show flow into the lesions. The fetal kidneys were enlarged and echogenic with multiple cystic lesions (Figure 2). The 3-D ultrasound in the rendering mode clearly revealed the enlarged fetal kidneys and the increased abdominal volume (Figure 3). MRI study was performed on the same day and T2-weighted sequences revealed the heterogeneous fetal liver as a consequence of congenital saccular dilations (Figure 4). The fetal kidneys were bilaterally enlarged with multiple cystic lesions with hypersignal in the T2 sequence (Figure 5). The fetal lungs presented low signal intensity, suggesting pulmonary hypoplasia. The bladder was not visible, and the amount of amniotic fluid was reduced for the gestational age. No other malformations were found, including any in the pancreas, and the diagnosis of Caroli disease associated with ARPKD was established. The fetus died in utero 2 weeks later, and the parents declined postmortem anatomopathological studies.

Figure 1. A 2-D ultrasound in the axial view showing heterogeneous fetal liver (*arrows*).

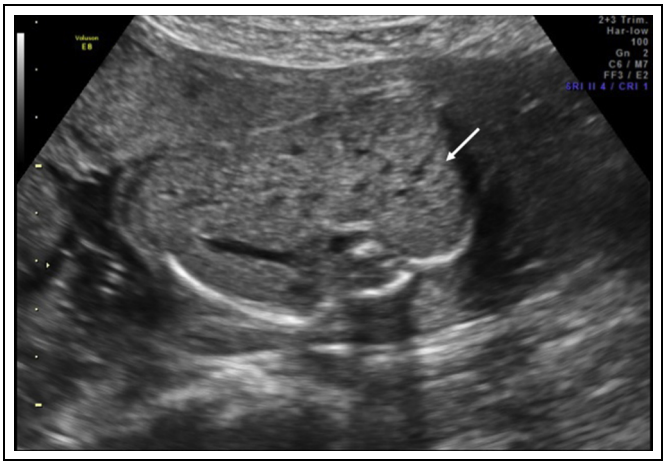
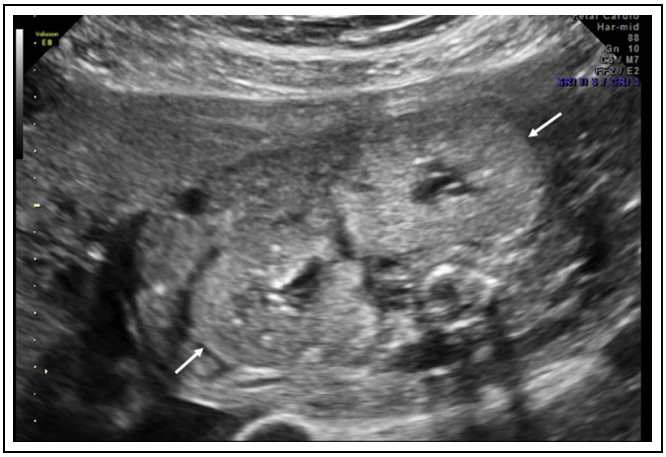


Figure 2. A 2-D ultrasound in the axial view showing enlarged and echogenic fetal kidneys (*arrows*).



DISCUSSION

Caroli disease is a consequence of ductal plate malformation at different levels of the intrahepatic biliary tree. Clinical presentation is characterized by bile stagnation and hepatolithiasis, which explain the recurrent cholangitis, and portal hypertension as a consequence of congenital liver fibrosis.[5] There are two forms of the disease: type I, the

Figure 3. A 3-D ultrasound in the rendering mode showing enlarged fetal kidneys.