Case Report

Carolí’s Disease Presented as a Huge Hepatic Multilobulated Cystic Mass in a Newborn

Pei-Ling Wu, Yung-Ning Yang, Shu-Leei Tey, Chun-Hwa Yang, Chien-Yi Wu

Carolí’s disease is a rare congenital disorder characterized by cystic dilatation of the intrahepatic bile ducts. We reported a newborn with a prenatally detected huge hepatic multilobulated cyst, the diagnosis of which was confirmed by biopsy after birth. However, the hepatic cyst regressed gradually without complication. In addition, there have been totally six patients who were prenatally diagnosed as having Carolí’s disease and the prognosis of Carolí’s disease varies, depending upon the severity of disease.

Key words: Carolí’s disease, hepatic cyst, newborn

Introduction

Carolí’s disease, characterized by cystic dilatations of the intrahepatic bile ducts that communicate with the biliary system, is a rare congenital disorder first described by Carolí in 1958. It is also known as choledochal cyst type V, or type IVA if it is associated with an extrahepatic choledochal cyst. Carolí’s syndrome is defined as congenital bile ductal dilatation associated with features of congenital hepatic fibrosis, whereas Carolí’s disease, which is less common, refers to isolated dilatation of large intrahepatic bile ducts without other apparent hepatic abnormalities.

Carolí’s disease has been reported in adults, adolescents and children, with clinical presentations including abdominal pain, fever, recurrent acute cholangitis or cholangiocarcinoma. There is a limited number of reports on newborns, especially when detected prenatally. Herein, we presented a newborn with Carolí’s disease detected antenatally. The diagnosis was confirmed by pathologic analysis of the surgical specimen after birth. In addition, we reviewed relevant literature on PubMed with search terms including “Carolí’s disease” OR “choledochal cysts type V”, combined with AND “neonate” OR “newborn”.

Case Report

This term female neonate, 38 weeks of...
gestation with a birth weight of 3100 gm, was
born to a Taiwanese non-consanguineous
36-year-old mother who was gravida 2, para 2,
via cesarean section. The mother had Schizo-
phrenia and took the anti-psychotic medication
Haloperidol regularly. The first baby of this
family had died at two years old of undiag-
nosed disease. There was no family history of
liver or renal disease. At 28 weeks of gestation,
polyhydramnios and a complex cystic mass
in the fetal liver were noted on sonographic
examination. There was no evidence of other
structural anomalies in the fetus. After birth,
she had a soft, flat abdomen with hepatomeg-
aly, 7-8 cm below the right costal margin.

Liver function tests revealed that aspar-
tate aminotransferase (AST) and alanine
aminotransferase (ALT) were 34 U/L and 12
U/L, respectively, with normal prothrombin
time and partial prothrombin time. The high-
est total bilirubin level was 6.86 mg/dL, with
direct bilirubin of 0.34 mg/dL on day 7. The
level of alkaline phosphatase (ALK-P) was 169
IU/L, gamma-glutamyl transpeptidase (GGT)
of 119 U/L, and albumin of 3.74 g/dL. Alpha
fetal protein (αFP) level was up to 57817 ng/mL
on the 13th day. Hepatitis B and C titer surveys
were all negative.

Abdominal sonography (Fig. 1A) revealed
a multilobulated intrahepatic cystic mass of
size 6.2 × 4.8 × 5.7 cm within the segment V
of liver. The liver parenchyma and extrahepa-
tic bile ducts were normal. There was neither
splenomegaly nor ascites. The kidneys were
normal in appearance. Magnetic resonance
cholangiopancreatography (MRCP) showed a
multilobulated cystic lesion in segment V of
the liver of size 6.2 × 4.5 × 5.8 cm (Fig. 1B).
No definite imaging evidence of choledochal
cyst or biliary atresia was noted.

At 14th day of age, surgical explora-
tion with laparoscopy was performed and a
huge hepatic cyst connected to the biliary tree
was found. Cholangiogram during operation
showed that the hepatic cyst was communi-
cated with the intrahepatic ducts of the bili-
ary system, and no extrahepatic bile ducts
were involved, compatible with the diagnosis
of Caroli’s disease. There was no obstruction
of bile flow. Thirty-seven milliliters of turbid

![Fig. 1 The image of intrahepatic cyst. (A) A multilobulated cystic mass (6.2 × 4.8 × 5.7 cm) within segment V of the liver from abdominal ultrasonogram performed after birth. (B) A multilobulated cystic lesion in the right upper quadrant of the abdomen was noted, measuring 6.2 × 4.5 × 5.8 cm from T1-weighted image of magnetic resonance cholangiopancreatography (MRCP).]
Caroli’s disease, which is characterized by cystic dilatations of the intrahepatic bile ducts communicating with the biliary tract, is a rare congenital disorder. Reviewing of related literature showed a limited number of reported newborns who were diagnosed as having the disease. To our knowledge, there have only been five reported cases with antenatal diagnosis in the literature (Table 1). We presented a newborn of Caroli’s disease detected antenatally and confirmed after birth. This is also the first patient with prenatally detected Caroli’s disease whose diagnosis was also confirmed postnatally during surgery and after pathological analysis of the surgical specimen.

Review of literature revealed totally 6 patients whose live cysts were detected prenatally with confirmation of the diagnosis of Caroli’s disease including the present case (Table 1). The overall survival was 66.7% in the absence of other comorbidities. Two patients (33.3%) showed gradual regression on sonographic follow-up. There have been no reports of cholangiocarcinoma till now.

Caroli’s disease exists with a heterogeneity of clinical presentations ranging from asymptomatic in neonate to adult-onset recurrent cholangitis or liver cirrhosis. The onset of symptoms can be as early as the neonatal period, especially in cases associated with congenital hepatic fibrosis. The majority of adult patients present with abdominal pain, fever, acute cholangitis, recurrent cholangitis, cholangiocarcinoma and symptoms of portal hypertension such as ascites and esophageal varices hemorrhage.

The diagnosis of Caroli’s disease and...
Caroli’s syndrome mainly relies on imaging studies that demonstrate continuity between the cystic lesions and the biliary tree. Although endoscopic retrograde cholangiopancreatography (ERCP) or percutaneous transhepatic cholangiography (PTC) have been considered the “gold standards” to demonstrate biliary anatomy,9 Magnetic resonance cholangiopancreatography (MRCP), hepato-iminodiacetic acid (HIDA) scan, or intraoperative cholangiogram might be better choices in newborn for anatomical delineation of the biliary tract.10 In our presented patient, imaging studies did not find evidence of communication between the cyst and bile ducts, making the diagnosis more difficult. Therefore, we relied on intraoperative cholangiogram to make a definite diagnosis.

The treatment of Caroli’s disease depends on the clinical features and the location of the biliary abnormalities. In spite of acceptable conservative management of complication of Caroli’s disease such as acute cholangitis and intrahepatic cholelithiasis, surgery with hepatectomy offers a definite therapy for localized Caroli’s disease. Symptomatic relief after hepatectomy which removed the whole lesion is often complete and permanent. However, most patients of Caroli’s disease were not suitable for total removal of the lesion. These patients should be considered for liver transplantation if they develop recurrent cholangitis or serious complications of portal hypertension. Liver transplantation is an optimal option for patients with advanced Caroli’s disease and should be considered in a timely fashion to prevent worsening complications including refractory cholangitis and cholangiocarcinoma.

The treatment of newborn diagnosed as having Caroli’s disease prenatally has so far been conservative. It seems that possibility exists regarding regression of the cystic lesion. Although the natural history of Caroli’s disease diagnosed in utero is still unclear, a period of observation appears warranted in asymptomatic patients8 for symptoms and signs of cholangitis, cirrhosis, portal hypertension and cholangiocarcinoma.

The prognosis of Caroli’s disease varies, depending upon the severity of disease and the presence of coexisting renal dysfunction. Recurrent infections and other complications related to biliary lithiasis can be associated with significant morbidity. Since Caroli’s disease is a rare and possibly genetically heterogeneous condition, it remains unclear if cases presenting prenatally or neonatally have poorer prognosis compared with those diagnosed as having the disease as adults.9

In conclusion, we presented a newborn with Caroli’s disease detected antenatally with

Table 1. Reported cases of Caroli’s disease detected prenatally and neonatal presentation

<table>
<thead>
<tr>
<th>Case number</th>
<th>Gender</th>
<th>Age at detection</th>
<th>Image findings of liver from fetus</th>
<th>Renal involved</th>
<th>Operation/biopsy</th>
<th>Outcome</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Un</td>
<td>36th weeks of gestational age</td>
<td>Two large hepatic cysts</td>
<td>Y (ARPKD)</td>
<td>N</td>
<td>Died at birth due to pulmonary hypoplasia</td>
<td>4</td>
</tr>
<tr>
<td>2</td>
<td>Un</td>
<td>28th weeks of gestational age</td>
<td>Un</td>
<td>N</td>
<td>N</td>
<td>Survived at 3 years old</td>
<td>5</td>
</tr>
<tr>
<td>3</td>
<td>F</td>
<td>25th weeks of gestational age</td>
<td>Several intrahepatic cysts</td>
<td>N</td>
<td>N</td>
<td>Survived at 1 years old</td>
<td>6</td>
</tr>
<tr>
<td>4</td>
<td>F</td>
<td>23th weeks of gestational age</td>
<td>Multiple cystic dilations in the liver</td>
<td>N</td>
<td>N</td>
<td>Died at 11 weeks of age due to sepsis</td>
<td>7</td>
</tr>
<tr>
<td>5</td>
<td>M</td>
<td>33rd weeks of gestational age</td>
<td>A cystic liver mass</td>
<td>Y (ARPKD)</td>
<td>N</td>
<td>Survived at 3 months old</td>
<td>8</td>
</tr>
<tr>
<td>6</td>
<td>F</td>
<td>28th weeks of gestational age</td>
<td>A huge intrahepatic multiloculated cyst</td>
<td>N</td>
<td>Y</td>
<td>Survived at 3 months old</td>
<td></td>
</tr>
</tbody>
</table>
unique presentations, whose diagnosis was confirmed by surgical pathology after birth. This is the first case of prenatally detected Caroli’s disease confirmed postnatally. Our case illustrates the unique presentation of Caroli’s disease in a newborn and the need for considering Caroli’s disease in the differential diagnosis of prenatally detected cystic liver lesions.

References