Pediatric Oncall Journal (Oct-Dec 2022)
Volume 19, Issue 4:141-142
DOI: https://doi.org/10.7199/ped.oncall.2022.50

PEDIATRIC ONCALL
CHILD HEALTH CARE

TEACHING FILES (GRAND ROUNDS)

CAROLI'S DISEASE IN INFANCY - A REPORT OF 2 CASES

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KEYWORDS
Caroli's disease, infancy

ARTICLE HISTORY
Received 25 July 2021
Accepted 5 August 2021

Clinical Problem

A 9-month-old boy born of non consanguineous marriage presented with progressive abdominal distension with prominent abdominal veins since 2 months of age. He was born at full term and had achieved milestones appropriately for age. On examination, he was malnourished (weight = 5 kg, less than 5th centile; height = 64 cm, less than 5th centile), had tachycardia (heart rate = 140/min) respiratory rate of 42/min and blood pressure of 92/60 mm of Hg. He had pallor, palmar erythema and splenohepatomegaly with dilated abdominal veins. Investigations showed hemoglobin of 8.1 gm%, WBC count of 25,400/cumm and platelet count of 4,05,000/cumm. Liver function tests are depicted in Table 1. USG abdomen showed multiple round hypoechoic areas in the liver with collaterals around liver capsule and splenomegaly. A CT abdomen showed polycystic kidneys with dilated cysts in liver (Figure 1). Serum creatinine and urine examination was normal. Thus the child was diagnosed as Caroli's syndrome with portal hypertension and polycystic kidney disease.

Table 1. Liver function tests in the patient.

Lab parameters	Patient values
Bilirubin (mg/dl)	1
Direct bilirubin (mg/dl)	0.5
SGOT (IU/L)	100
SGPT (IU/L)	46
Total protein (gm/dl)	6.5
Albumin (gm/dl)	3.5
Alkaline phosphatase (IU/dl)	2550
Prothrombin Time (sec)	32.4
Partial thromboplastin Time (sec)	46.0

How common is Caroli's syndrome in infancy? How to manage this case?

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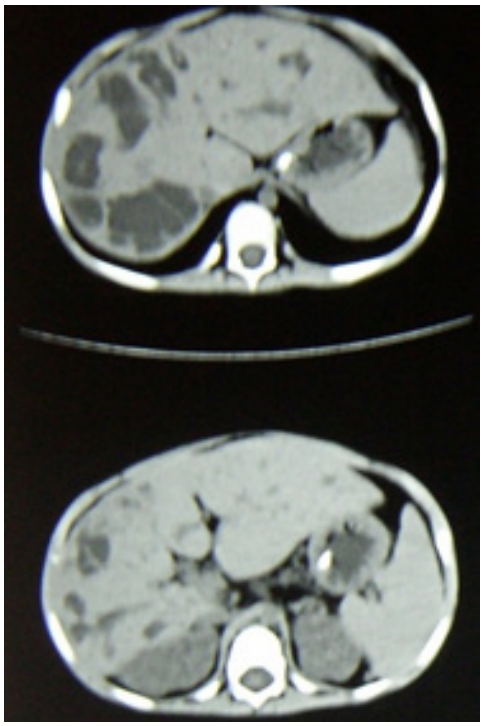
Figure 1. CT abdomen showing cysts in liver and bilateral kidney suggestive of Caroli's syndrome.



Discussion

Caroli's disease and Caroli's syndrome are a rare group of congenital disorders of the biliary tree involving ectasia of the bile ducts.[1] The usual age of presentation of this spectrum is in the adolescent age group.[2] It rarely presents in infancy and there have been very few reports of its diagnosis before one year of age.[2] According to the Todani classification, Caroli's disease falls under Type V of choledochal cysts.[3] It presents in 2 forms. The first is a 'pure form' called Caroli's disease and the second is Caroli's syndrome, differentiated by the presence of a component of congenital hepatic fibrosis in the latter. The disease is less common than the syndromic presentation.[4] An association with autosomal recessive polycystic kidney disease has been found.[5] Our patient had Caroli's syndrome. Clinically, Caroli's spectrum shows a male preponderance.[4] Our patient was also a male. Mean age when the spectrum is diagnosed is 25 (range 1-60) years.[2] Neonatal presentation has rarely been

described.[1,6] Prenatal diagnosis has also been described.[2] Patients typically give a history of intermittent abdominal pain and symptoms suggestive of recurrent cholangitis and hepatolithiasis. Congenital hepatic fibrosis leads to the development of portal hypertension and its associated manifestations in Caroli's syndrome. Renal involvement may be in the form of renal cysts, renal tubular dilatation, autosomal recessive polycystic kidney disease and rarely autosomal dominant kidney disease.[3] Our patient with Caroli's syndrome presented with chronic liver disease and portal hypertension. There is an increased risk of developiong cholangiocarcinoma in patients with Caroli's spectrum.[7]

A chromosomal loss of 3p and gain of 8q is involved in the pathogenesis of Caroli's disease.[1] The disease results from an arrest of remodeling of the ductal plate, which is responsible for the formation of the biliary tree. The malformation in Caroli's disease is associated with the large bile ducts, whereas Caroli's syndrome involves the distal, interlobular ducts.[3] The disease may be diffuse or have focal involvement. The association with autosomal recessive polycystic kidney disease (ARPKD) is explained by a mutation in the PKHD1 gene. This gene codes for a protein called fibrocystin, which is expressed on the biliary tree and kidneys, giving rise to the amalgamation of clinical features.[4] We could not do the genetic tests in our patient due to unaffordability.

Ultrasound today remains the first line of investigation among imaging studies in Caroli's spectrum. Dilatation of large intrahepatic bile ducts are visualized, along with multiple hypoechoic areas in the liver. Other modalities for diagnosis include computed tomography, endoscopic retrograde cholangiopancreatography, radionuclide hepatobiliary imaging and intraoperative cholangiography.[1] Renal function tests to rule out renal involvement are commonly done. Other close differential diagnosis of this spectrum includes Von Mayenburg complexes and primary sclerosing cholangitis.[8]

Treatment of the condition is essentially supportive, with a role of antibiotics to prevent occurrence of sepsis following cholangitis. Ursodeoxycholic acid can be given for hepatolithiasis.[8] Depending on the expanse of area involved, surgical resection may be tried for focal involvement, whereas diffuse involvement heralds the need for a liver transplantation.[9] The prognosis varies depending on the structures involved. Recurrent cholangitis in patients of Caroli's disease and spectrum is very commonly the cause of death. Cholangiocarcinoma occurs more commonly in these patients. Renal involvement in infancy shows a higher likelihood of the patient going into renal failure.

Compliance with ethical standards

Funding: None
Conflict of Interest: None

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