

Atypical presentation of caroli's syndrome: A case report

Rahul Tiotia¹, Monika Sharma², Vani Narayani², Shikha Singh¹, Vivek Dewan³, Shivani Deswal⁴, Abhinandan Tiotia¹, Sudhir Baghel¹

¹PG Student, ²Senior Resident, ³Professor, ⁴Associate Professor, Department of Pediatrics, Post Graduate Institute of Medical Education and Research and Dr. Ram Manohar Lohia Hospital, New Delhi, India

Correspondence to: Dr. Monika Sharma, Department of Pediatrics, Post Graduate Institute of Medical Education and Research and Dr. Ram Manohar Lohia Hospital, New Delhi. E-mail: monikash789@gmail.com

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ABSTRACT

Caroli's disease and Caroli's syndrome are rare congenital disorders. Caroli's disease is characterized by multiple sequential cystic or saccular dilatations of the large intrahepatic biliary ducts while Caroli's syndrome has small bile duct involvement and congenital hepatic fibrosis. The incidence of Caroli's disease is as low as 1/1,000,000 people. The average age of presentation is early adolescence. Magnetic resonance cholangiopancreatography is a most valuable investigation in diagnosis. Here, we report the case of Caroli's Type II without renal involvement as late as 6 years of age with severe portal hypertension and hypersplenism. The child had no history of jaundice or recurrent abdominal pain in the past.

Key words: Autosomal recessive, Caroli's syndrome, Congenital hepatic fibrosis, Kidney disorder

Caroli's disease is characterized by an abnormal widening of the intrahepatic biliary ducts. It is also classified as Type V choledochal cysts according to Todani's classification [1,2]. In India, Caroli's disease attributes to 5% of all causes of chronic hepatobiliary diseases [3].

Caroli's disease Type I (simple type) is most often diagnosed at an average age of 20 years. The simple type presents with recurrent right upper quadrant pain and recurrent attacks of cholangitis. Type II Caroli's disease is also known as Caroli's syndrome. It is usually associated with autosomal recessive polycystic kidney disease (ARPKD) in which the patients may have various degrees of renal cysts, interstitial fibrosis, and renal failure and presents with features suggestive of portal hypertension [4]. We herein report a child who presented at 6 years of age with hematemesis.

CASE REPORT

A 6-year-old boy presented with complaints of 10–12 episodes of hematemesis and melena. There was no previous history of jaundice, abdominal distension, recurrent fever with the right upper quadrant abdominal pain, or similar episodes in the past.

On examination, the child had severe tachycardia (heart rate of 182/min), hypotension (blood pressure 80/40 mmHg), feeble peripheral pulses, and severe pallor (hemoglobin –3.6 g/dl). There were no icterus, cyanosis, clubbing, or significant lymphadenopathy. On abdominal examination, the abdomen was distended. The right lobe of the liver was palpable 1 cm below the right costal margin, and the left lobe was palpable 5 cm below the xiphisternum with a smooth surface, sharp leafy margins, and firm consistency. Spleen (splenic notch) was palpable 5 cm below the left costal margin in

midclavicular line with a smooth surface, sharp margins, and firm consistency. The liver span was 8 cm on percussion. Kidneys were not palpable. No other abdominal masses were palpable. No shifting dullness or evidence of free fluid was present on percussion. Other systems examination was not remarkable.

On investigations, hemoglobin was 3.6 g/dl, total leukocyte count was 3200/mm³, and platelets were 57,000/mm³. Peripheral smear for malarial parasite was negative, and red blood cells were normocytic and normochromic with leukopenia and thrombocytopenia. Investigative work-up for pancytopenia was done and the patient was found to have both iron deficiency (serum iron - 22 mg/dL and transferrin saturation - 5%) and Vitamin B12 (serum B12 levels - 58 ng/mL) deficiency. Liver function tests revealed total serum bilirubin as 0.6 mg/dl with direct fraction - 0.3 mg/dl and indirect - 0.3 mg/dl, serum glutamic-oxaloacetic transaminase - 130 U/dl, serum glutamic pyruvic transaminase - 59 U/dl, alkaline phosphatase - 109 IU/L, international normalized ratio (INR) - 2.14 and 1.15 after administration of Vitamin K and three units fresh frozen plasma, and gamma-glutamyltransferase - 21 U/dl. Kidney function tests were normal. Viral markers and metabolic workup were negative.

Ultrasonography revealed liver of normal size with coarse echotexture and perihepatic free fluid. There was ectasia of intrahepatic biliary radicals and gallbladder was contracted with thickened irregular walls, moderate splenomegaly, and mild ascites. Upper gastrointestinal endoscopy revealed esophageal varices of Grade III (Paquet classification) [5] one column and two columns of Grade II, and mild antral gastritis. Magnetic resonance cholangiopancreatography (MRCP) findings were coarse and shrunken liver with nodular surface, distorted architecture with relative caudate, and the left lobe

hypertrophy suggestive of changes of chronic liver disease, spleen was moderately enlarged in size 18 cm, mild free fluid seen in pelvic cavity, multiple portal peripancreatic, pre-paraortic, aortocaval, and retrocaval mesenteric lymphadenopathy with largest measuring approximately 20 mm in size (Fig. 1). Bilobar intrahepatic biliary radicals are moderately dilated with patent confluence with cystic dilatation of the right lobar intrahepatic biliary radical; however, common bile duct was not found to be dilated (Fig. 2). Gallbladder was found to be contracted with peri-gallbladder and periportal fibrosis. Both kidneys were normal.

Liver biopsy findings showed distorted liver architecture with the presence of multiple jigsaws shaped nodules separated by a marked fibrous expansion of portal tracts with portal tracts showing multiple proliferating small irregular bile ducts few of which are dilated (ductal plate malformation) along with mild focal chronic inflammation with lymphoid aggregate formation. On reticulin stain, single-cord pattern is seen with the final impression as congenital hepatic fibrosis.

The patient was given supportive care with packed red blood cells and fresh frozen plasma transfusion, Vitamin K, and

other fat-soluble vitamins were given along with proton-pump inhibitors and ursodeoxycholic acid. Endoscopic band ligation of esophageal varices was done. Patient's attendants were explained about the prognosis and referred for liver transplantation as definitive management. Propranolol prophylaxis was started.

DISCUSSION

Caroli's disease is a rare autosomal recessive congenital disorder characterized by non-obstructive, saccular or fusiform, multifocal, segmental dilatation of the intrahepatic bile ducts. Sometimes, the extrahepatic duct involvement has also been reported. ARPKD and Caroli's syndrome share the same genetic defect, i.e., PHKD1 mutation on chromosome 6p21 [6,7].

The simple (Type I) Caroli's disease presents with recurrent cholangitis and cholelithiasis while type II presents with cirrhosis and portal hypertension. There is an increased risk of cholangiocarcinoma in Type II (2.5–17%) and the prognosis is poor. The case we have discussed had Type II Caroli's disease. Liver biopsy and MRCP confirmed associated hepatic fibrosis in the patient. For localized disease, lobectomy may be considered while generalized disease needs liver transplantation. The case we discussed had diffuse fibrosis, so the liver transplant was advised. In a case series of 104 Caroli's disease patients, 96 patients underwent liver transplantation alone and eight underwent combined liver/kidney transplantation.

The patient survival and graft survival were analyzed by Kaplan–Meier survival analysis, and the risk of death and graft loss were analyzed by Cox proportional hazards regression. The overall 1-, 3-, and 5-year graft (79.9%, 72.4%, and 72.4%) and patient (86.3%, 78.4%, and 77%) survival rates were excellent for patients after liver transplantation. For combined liver/kidney transplantation ($n = 8$), the 1-year patient survival and graft survival were 100%. Proportional hazards analysis identified Asian ethnicity, elevated bilirubin, requirement of life support or hospitalization before transplantation, and a cold ischemia time >12 h as associated with increased risk of both graft loss and death. A history of prior transplant or prior abdominal surgery was also associated with an increased risk of graft loss [8].

Liver transplantation is an excellent treatment 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.

CONCLUSION

The classical clinical presentation of recurrent abdominal pain and jaundice is not always present in patients with Caroli's syndrome. A high index of clinical suspicion is needed for the diagnosis.

REFERENCES

1. Todani T, Watanabe Y, Narusue M, Tabuchi K, Okajima K. Congenital bile duct cysts: Classification, operative procedures, and review of thirty-seven cases including cancer arising from choledochal cyst. *Am J Surg* 1977; 134:263-9.

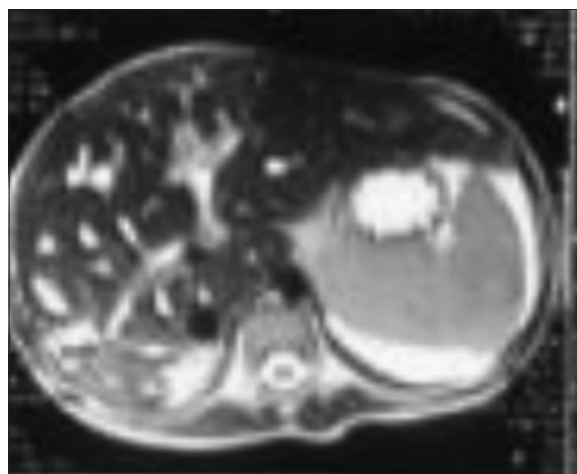


Figure 1: Magnetic resonance cholangiopancreatography - axial T2-weighted image showing caudate lobe hypertrophy and splenomegaly

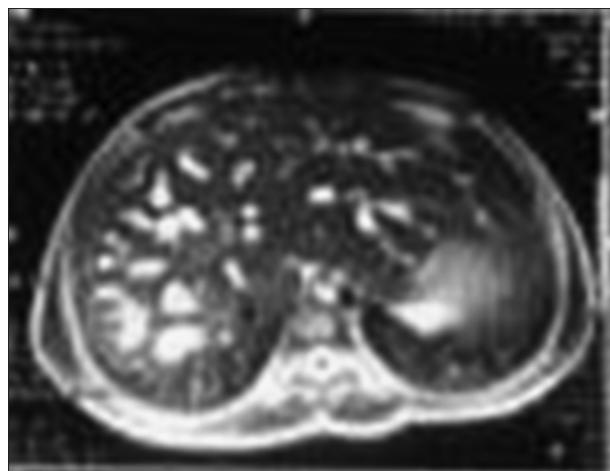


Figure 2: Magnetic resonance cholangiopancreatography - axial T2-weighted image showing multifocal cystic dilatation of intrahepatic biliary radical

2. Biliary Cysts. Available from: <http://www.uptodate.com>. [Last accessed on 2018 Dec 25].
3. Shah I, Bhatnagar S. Clinical profile of chronic hepatobiliary disorders in children: Experience from tertiary referral centre in Western India. *Trop Gastroenterol* 2010;31:108-10.
4. Liang JJ, Kamath PS. Caroli syndrome. *Mayo Clin Proc* 2013;88:e59.
5. Paquet KJ. Prophylactic endoscopic sclerosing treatment of the esophageal wall in varices-a prospective controlled randomized trial. *Endoscopy* 1982; 14:4-5.
6. Baris T, Ocak I, Daryanani K, Font-Montgomery E, Lukose L, Bryant J, *et al.* Autosomal recessive polycystic kidney disease and congenital hepatic fibrosis (ARPKD/CHF). *Pediatr Radiol* 2009;39:100-11.
7. Gael JL, Rice RR, Suarez ES. Autosomal recessive polycystic kidney disease: Radiologic-pathologic correlation. *Radiographics* 2000;20:837-55.
8. Millwala F, Segev DL, Thuluvath PJ. Caroli's disease and outcomes after liver transplantation. *Liver Transpl* 2008;14:11-7.

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