Case Report

Caroli's Syndrome in a 5½-Year-Old Bangladeshi Girl: A Case Report

Nazmul Ahamed[1], Dipanwita Saha[2], AZM Raihanur Rahman[3], Mukesh Khadga[4], Sharmin Akter[5], Md Rukunuzzaman[6]

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Abstract

Caroli's syndrome is a rare inherited disorder characterized by multiple segmental cystic or saccular dilatation of the intrahepatic bile duct associated with congenital hepatic fibrosis. Symptoms of Caroli's syndrome may appear early or late during life and its presentation is highly variable. Portal hypertension followed by development of oesophageal varices is the main consequence of congenital hepatic fibrosis. Up to 60% of Caroli's syndrome patients are associated with renal involvement. The diagnosis of Caroli's syndrome mainly depends on histology and imaging method that can show the communication between bile ducts and saccule. Important complication is cholangitis and later may progress to cholangiocarcinoma. For symptomatic Caroli's syndrome, liver transplantation is the only curative treatment. Here, we report a case of Caroli's syndrome in a 5½ year old girl *admitted in the department of Pediatric Gastroenterology and Nutrition, BSMMU with the complaints of abdominal distension since birth. Her CT scan report showed type V choledochal cyst with large cyst in right lobe of liver and polycystic kidney disease. Endoscopy of upper GIT revealed grade II oesophageal varices and MRCP also suggested Caroli's disease (Type V choledochal cyst). Finally, she was diagnosed as a rare case of Caroli's syndrome (Type V choledochal cyst with grade II oesophageal varices with polycystic kidney disease).*

Key words: *Caroli's syndrome; Congenital hepatic fibrosis; Cholangitis*

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Introduction

A rare congenital condition Caroli's disease was first described by Jacques Caroli in 1958 characterized by multiple segmental saccular or cystic dilatation of the intrahepatic bile ducts. It is also classified

1. *Resident, Department of Pediatric Gastroenterology and Nutrition, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh*
2. *Resident, Department of Pediatric Gastroenterology and Nutrition, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh*
3. *Resident, Department of Pediatric Gastroenterology and Nutrition, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh*
4. *Resident, Department of Pediatric Gastroenterology and Nutrition, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh*
5. *Resident, Department of Pediatric Gastroenterology and Nutrition, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh*
6. *Professor and head, Department of Pediatric Gastroenterology and Nutrition, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh.*

Correspondence *Nazmul Ahamed dr.nazmulahamed1985@gmail.com*

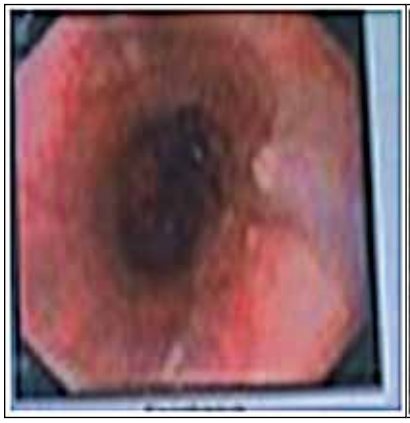
as type V choledochal cysts according to Todani's classification.[1,2] Caroli's disease when associated with congenital hepatic fibrosis, it is referred as Caroli's syndrome.[3,4] Caroli's syndrome is generally autosomal recessive. The prevalence of CS is approximately one in 6,000– 40,000 newborns.[4]

Case report

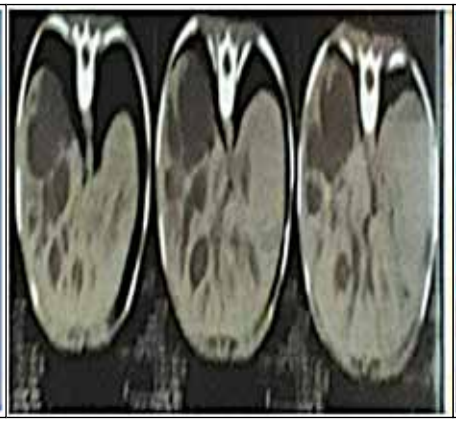
A 5½-year-old girl, 2nd issue of non-consanguineous parents, got admitted in the department of Pediatric Gastroenterology and Nutrition, BSMMU with the complaints of gradual abdominal distension since birth and low-grade intermittent fever, occasionally associated with chill and rigor, highest recorded temperature 101°F for last five days. She was suffering from similar type of illness for one year of her age and admitted in several private hospitals, but her condition did not improve. There was no remarkable past history. On examination she was ill-looking, mildly pale, anicteric, afebrile, BCG mark present, all vitals were in normal limit, anthropometrically well-thrived. Her liver edge was 6 cm from right costal margin in mid clavicular line, consistency was firm and non-tender. Spleen was 7 cm enlarged along its long axis, ascites was present evidenced by shifting dullness. Other physical sign was unremarkable. Laboratory investigations showed reduced haemoglobin (Hb 9.4 g/dL), platelet count 1,00,000/cmm, ESR slightly high (30 mm in 1st hr.), liver function test was normal. USG of whole abdomen showed hepatic cyst in both lobes and bigger both kidneys with high cortical echogenicity and loss of renal architecture, renal function test was also normal and CT scan report showed hepatosplenomegaly, type V choledochal cyst with large cyst in right lobe of liver and polycystic kidney disease.

Endoscopy of upper GIT showed grade II oesophageal varices which were a result of portal hypertension due to development of hepatic fibrosis. Diagnosis was confirmed by MRCP that suggested Caroli's disease (Type V choledochal cyst) with large hepatic cyst in right lobe of liver.

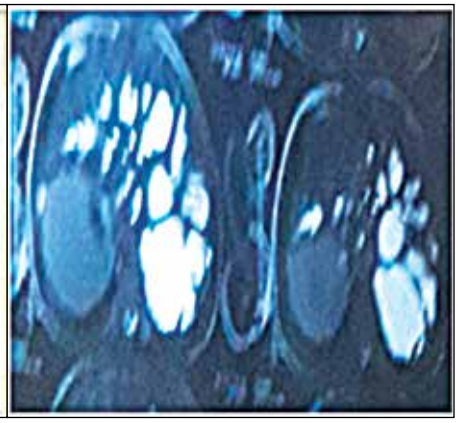
Both cholangiography and liver biopsy are helpful to establish the final diagnosis, but not possible due to financial problem. The child was treated conservatively with broad spectrum antibiotics. Oral supplementations with vitamin A, D, E, K and hematinics were given. Fever resolved within two days of admission. By this time, consultation was taken from Pediatric Surgery Department, BSMMU and advice for definitive treatment liver transplantation was given. The parents were counseled regarding the disease progression with prognosis and advised to come for regular follow-up.



Endoscopy of upper GIT (Grade II oesophageal varices)



CT scan of the abdomen showing hepatosplenomegaly, areas of focal intrahepatic biliary radicals dilatation type V choledochal cyst with large cyst in right lobe of liver and polycystic kidney disease



MRCP report suggested Caroli's disease (Type V choledochal cyst) with large hepatic cyst in right lobe of liver

Discussion

Caroli's syndrome is a rare form of congenital disease that represents ectasia of intrahepatic bile duct and congenital hepatic fibrosis. In 1958, Jacques Caroli first reported a case with a distinct clinical entity known as Caroli's disease. Caroli described two forms of this disease: the so called "pure form" of Caroli's disease which occurs in a focal or diffuse manner, characterized by saccular, communicating intrahepatic bile duct dilatation and the second form, termed as Caroli's syndrome having relatively less bile duct dilatation, but associated with hepatic fibrosis that results in portal hypertension and terminal liver failure.[3] The ductal plate malformation (DPM), a developmental abnormality of the portobiliary system, is the basis of the liver disease in CS. The severity of DPM and the level of the affected portobiliary tree results in a spectrum of abnormalities including congenital hepatic fibrosis (CHF) (microscopic bile ducts), CS (microscopic and medium size bile ducts) and CD (medium and large bile).[3,4,5] Many authors believe that CD and CS are actually different stages of the same disease characterized by peri-portal fibrosis and ductal dilatation.[6] Depending on whether duct dilatation or portal hypertension is the predominant pathology, patients present with recurrent cholangitis or hematemesis respectively. Associated cystic dilatation of kidneys is seen in 60−80% of the cases (renal tubular ectasia, medullary sponge kidney, cortical cyst, recessive polycystic kidney disease or rarely autosomal dominant polycystic kidney disease). These patients are usually asymptomatic (as far as renal disease is concerned) but may develop renal stone disease and infections.[3]

Caroli's syndrome is a combination of Caroli's disease (bouts of cholangitis, gall bladder stones and hepatolithiasis) and congenital hepatic fibrosis (portal hypertension). Our patient presented with abdominal distension since birth with recurrent history of fever suggesting as cholangitis. On examination, patient had hepatomegaly and splenomegaly that also indicate developing hepatic fibrosis followed by portal hypertension. The main consequence of congenital hepatic fibrosis are portal hypertension and the development of oesophageal varices.[6] In the majority of patients, portal hypertension will not be present or will appear only later in the disease evolution.[7] The late appearance of these symptoms in patients with Caroli's disease suggests that congenital hepatic fibrosis is dynamic and progressive.[8]

As hepatic fibrosis in CS is dynamic and progressive, portal hypertension is usually a late feature. Also, hepatocellular function of the liver is relatively well preserved in the early stages, with liver enzymes being either normal or mildly elevated.[5,6] Thrombocytopenia and leukopenia are present with portal hypertension and hypersplenism. The elevated erythrocyte sedimentation rate and leukocytosis may indicate cholangitis. Renal function tests can be deranged in CS having associated renal involvement, as in our patient.[6] Laboratory findings of our patient showed anaemia, platelet count 1,00,000/cmm, slightly high ESR, and normal liver function tests.

Definitive diagnosis of CS is confirmed with imaging studies such as ultrasonography, endoscopic/ magnetic retrograde cholangiopancreatography, computed tomography (central dot sign), radionuclide hepatobiliary imaging, intraoperative cholangiography and percutaneous transhepatic cholangiography.[1,5,6,9] Ultrasonographyis the initial investigation of choice. CT scan is an invaluable adjunct that complements ultrasound. It can identify cholangiocarcinoma and hepatic masses not identified by ultrasound. The diagnosis is more difficult to establish in the case of fusiform dilatations of the biliary tracts and endoscopic retrograde cholangiopancreatography (ERCP) is the gold standard in this situation. ERCP shows communication between the sacculi and bile ducts and diverticulum-like sacculi of the intra-hepatic biliary tree. In our case, ERCP was not required. US of our patient showed hepatic cyst in both lobes and bigger both kidneys with high cortical echogenicity and loss of renal architecture but renal function test was also normal. CT scan revealed hepatosplenomegaly, Type V choledochal cyst with large cyst in right lobe of liver and polycystic kidney disease.

Endoscopy of upper GIT showed grade II oesophageal varices and diagnosis was confirmed by MRCP that suggested Caroli's disease (Type V choledochal cyst)

with large hepatic cyst in right lobe of liver.

Though cholangiography and liver biopsy are helpful to establish the diagnosis, it was not done in our case due to financial crisis.

The treatment is primarily aimed at managing the associated complications of recurrent cholangitis, hepatic abscesses, biliary calculi and carcinoma. Recurrent cholangitis requires drainage which can be done by open surgery, positioning of an open stent or by percutaneous drainage. However, this treatment is just palliative. If the disease is confined to one lobe, partial lobectomy is the surgical treatment of choice.[10] In case of diffuse involvement of liver, treatment options include conservative management, endoscopic therapy (sphincterotomy for clearance of intra-hepatic stone), internal biliary bypass procedures and liver transplantation.[11] Our patient got supportive treatment along with antibiotics for suspected cholangitis and advice was given for definitive treatment (liver transplantation).

Because of silent and slow progression with bouts of cholangitis of this rare disease, Caroli's syndrome should be considered as one of the important differential diagnoses.

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