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Case Report

Diagnosis of Caroli's Disease: a Rare Case Report

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Abstract

Caroli's disease is a rare disease caused by Ductal Plate malformation. Patients may present with history of intermittent abdominal pain, pruritus and/ or symptoms of cholangitis. Cholangitis, liver cirrhosis, and cholangiocarcinoma are potential complications. It is rarely diagnosed in childhood. A six (06) years old was presented with intermittent abdominal pain, fever, pruritus, and hepatomegaly at the Department of Paediatric Gastroenterology, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh. After evaluating history, signs/ symptoms, physical examination, biochemical, ultrasonographic and endoscopic findings; this case was provisionally diagnosed as a compensated chronic liver disease with portal hypertension due to cause of cholestasis. Finally magnetic resonance cholangiopancreatography (MRCP) was done for further evaluation and the diagnostic findings of MRCP was suggestive of Caroli's disease. After consultation the MRCP report with Pediatric Surgery Department of BSMMU the patient was advised for liver transplantation due to multifocal involvement of liver. Lastly patient was discharged with supportive management.

Keyword: *Caroli's disease; cholangitis, MRCP*

INTRODUCTION

Caroli's disease is a rare cystic malformation of the liver caused by Ductal plate malformation (DPM).[1] It is transmitted by autosomal recessive inheritance but autosomal dominant inheritance is also reported. Usually, it is present in childhood and early adulthood. But neonatal presentation also occurs. Intermittent abdominal pain due to cholangitis and hepatomegaly are common clinical presentations.

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CASE DESCRIPTION

An 8-year-old immunized boy 2nd issue of consanguineous parents, presented with jaundice off and on since birth. He also developed pruritus since 6 months of age and gradual abdominal distension since 3 years of age. He had no history of fever, recurrent abdominal pain, vomiting out of blood, the passage of black tarry stool, any other bleeding manifestation or blood transfusion. He had a history of passage of dark urine and clay-colored stool. His family history is significant. He had a history of sib death who also suffered from jaundice since birth and died at 7 months of age. On physical examination, he was pale, deeply icteric, non-edematous, clubbing, leukonychia, and thenar and hypothenar muscle wasting were present, weight, and height were below 3rd centile. The abdomen was distended and the flanks were not full. Hepato-splenomegaly was present and ascites were absent. Other systemic examinations revealed normal findings. We provisionally diagnosed this case as a compensated chronic liver disease with portal hypertension due to cholestasis cause.

CASE MANAGEMENT

On admission, the patient was managed symptomatically. Investigation report revealed low hemoglobin % (Hb% 7.3 gm/dl), raised ALT (102 U/L), raised AST (197 U/L), raised gamma-GT (100U/L), raised ALP (495 U/L), direct hyperbilirubinemia (Total S. bilirubin 7.5 mg/dl and direct bilirubin 6.23 mg/dl) with normal S. albumin (35 gm/L) and prothrombin time (14.10 sec). Ultrasonography of the whole abdomen revealed coarse hepatic parenchyma and splenomegaly. Endoscopy of upper GI revealed Grade II esophageal varices.

After evaluating history, physical examination, and biochemical findings, the decision was made to do MRCP. MRCP report (Figure 1) revealed multiple hyperintense almost rounded areas of multifocal cystic dilatation of segmental intrahepatic bile ducts having continuity with the central biliary tree. Finding suggestive of Caroli's disease. Finally, the patient was diagnosed as Caroli's disease.

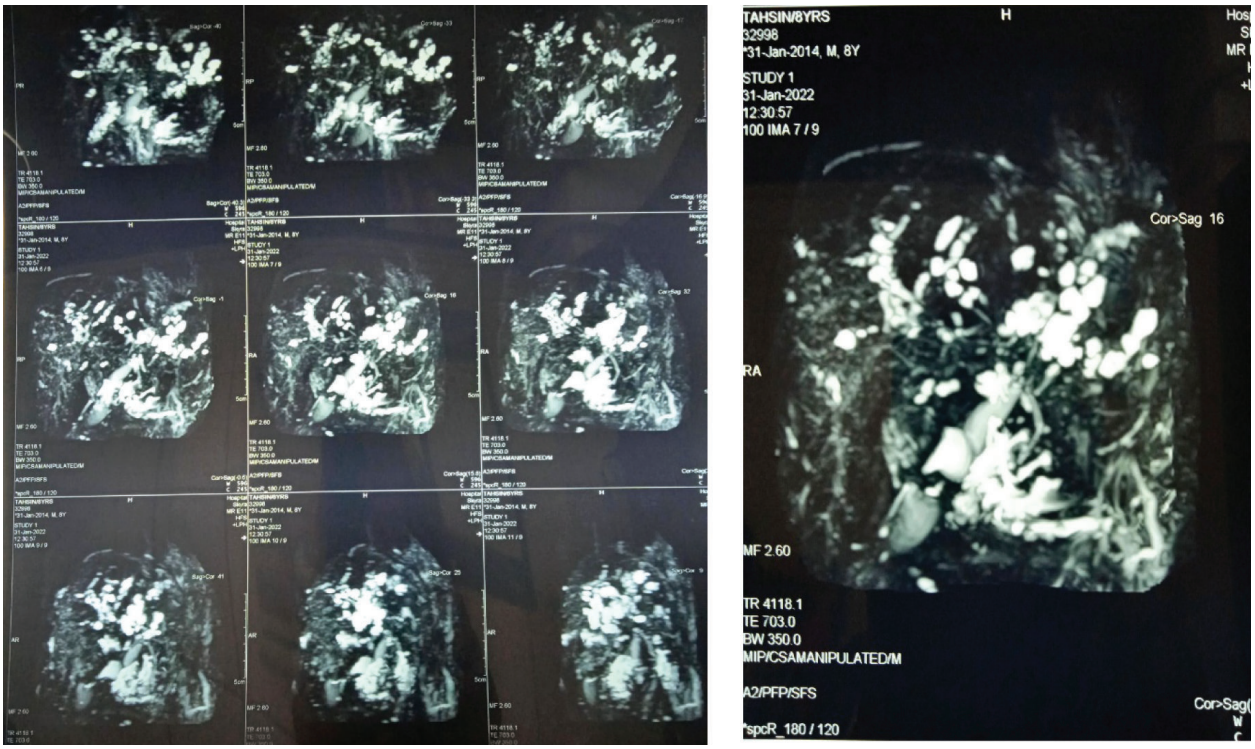

Figure- 1: *MRCP film shows Multiple hyperintense almost rounded areas of multifocal cystic dilatation of segmental intrahepatic bile ducts having continuity with the central biliary tree.*

After getting the MRCP report, we counseled with pediatric surgery department. However, due to multifocal involvement, the patient was advised for Liver transplantation. We discharged this patient with supportive management.

DISCUSSIONS

Caroli's disease is a rare congenital disease of the liver characterized by non-obstructed multiple segmental or saccular dilatations of the intrahepatic bile duct.[2] It was first described by French physician Jaques Caroli in 1958. When it is associated with hepatic fibrosis it is termed Caroli's syndrome (CS), which is more complex and is linked with portal hypertension.[3] Caroli's disease is also classified as a type V choledochal cyst. Usually, it affects both lobes of the liver in a diffuse manner. When Mono lobar involvement occurs, the left lobe is more commonly involved.[4] The main embryonic basis is Ductal plate malformation (DPM). The anatomic manifestation of cystic disease of the liver is determined by the level of involvement of the biliary tract.[5] Caroli's disease is characterized by DPM of the larger intrahepatic bile ducts, whereas malformations at the level of the interlobular bile ducts lead to congenital hepatic fibrosis.[1]

The most common presentation is intermittent abdominal pain and hepatomegaly. The patient may present with pruritus and jaundice.[6] The patient may also present with the features of complications like Cholangitis, cholelithiasis, portal hypertension, biliary abscess, septicemia, liver cirrhosis, and cholangiocarcinoma. On physical examination, hepatomegaly is usually present; splenomegaly can be present if portal hypertension develops. Palpable kidney indicates renal involvement.[7]

Duct ectasia and irregular, cystic dilation of the large proximal intrahepatic bile ducts with a normal common bile duct are the imaging findings of Caroli's disease.[8] These findings can be visualized with ultrasonography, endoscopic retrograde cholangiopancreatography, and magnetic resonance cholangiography. Direct visualization of the biliary tract with endoscopic retrograde cholangiopancreatography (ERCP) or percutaneous transhepatic cholangiography (PTC) is the gold standard diagnostic modality for Caroli's disease. The emerging diagnostic modality of choice for Caroli's disease is magnetic resonance cholangiopancreatography (MRCP). The "string of beads" pattern of the ectatic intrahepatic bile ducts is the characteristic MRI appearance of Caroli's disease.[9] The characteristic CT imaging and

ultrasonographic findings of caroli's disease is a central "dot-sign" appearance.[10] Nuclear scintigraphy is another diagnostic modality in which the characteristic beaded appearance of the intrahepatic bile ducts can be visualized.

The treatment option for Caroli's disease depends on its clinical features and the location of the biliary abnormalities. Ursodeoxycholic acid (UDCA) is the litholytic therapy for primary hepatolithiasis. Cholangitis can be treated with appropriate antibiotics and UDCA to prevent further episodes. Surgical resection (partial hepatectomy; lobectomy) remains a good option for segmental or unilobar involvement. For diffuse involvement in the liver, Orthotropic liver transplantation (OLT) is the only modality of treatment.[11] There is no definite indication or timing of OLT for patients with Caroli's disease. The primary indications of OLT which have been reported in different studies are signs of hepatic decompensation, defined as ascites, encephalopathy, coagulopathy, portal hypertension, jaundice, prolonged prothrombin time, or decreased albumin and recurrent cholangitis.[12]

CONCLUSIONS

Caroli's disease is a rare congenital malformation of the intrahepatic bile duct. Though it is rare, clinicians should it keep in mind as a differential diagnosis of recurrent abdominal pain and cholangitis. Radiological investigation such as MRCP is helpful in diagnosing this condition. Usually, the patient is managed symptomatically. However, surgical treatment may be required in specific circumstances.

Author's contribution:

Conception and design: Lubana Akram

Manuscript drafting and revising it critically: Lubana Akram, Rubaiyat Alam, Md.Rukunuzzaman

Approval of the final version of manuscript: Lubana Akram, Rubaiyat Alam, Md.Rukunuzzaman

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Data availability statement: Share upon request

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