Atypical presentation of a case of Caroli's disease

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Caroli's disease (CD) is a rare congenital diseases of intrahepatic bile ducts, characterized by saccular dilatations of intrahepatic bile ducts associated with congenital hepatic fibrosis. Present study describes a case of a 06 months old baby girl with complaints of gradual distension of abdomen since birth and recent discovery of a lump in right upper abdomen by her mother. Patient was referred to National Institute of Nuclear Medicine & Allied Sciences (NINMAS) for an abdominal ultrasonography, which revealed mild hepatomegaly with gross fusiform dilatation of common bile duct (CBD) and common hepatic duct (CHD) producing sac like structures. Intra-hepatic biliary channels were also found grossly dilated, producing multiple sac like structures. Magnetic Resonance Cholangio Pancreatogram (MRCP) revealed significant fusiform dilatation of CBD and CHD. Intrahepatic biliary trees were also grossly dilated. Liver biopsy was done to confirm the diagnosis after excision of choledochal cyst with Roux-en-Y hepaticojejunostomy. On histopathological examination, there was cholestasis, hepatocellular damage, parenchymal inflammation, bile duct proliferation, inflammation and central vein dilatation.

Keywords: Caroli's disease, Ultrasonography, Common bile duct, Common hepatic duct, Magnetic Resonance Cholangio Pancreatogram

Caroli's disease is a rare congenital disorder characterized by intrahepatic bile ducts dilatation and congenital hepatic fibrosis (1). Caroli's disease may represent single disorder or distinguished by congenital hepatic fibrosis. Incidence of females to male ratio is 4:1(2). The term Caroli's disease is applied if the disease is limited to ectasia or segmental dilatation of the larger intrahepatic ducts. This form is less common than Caroli's syndrome, in which congenital malformations of small bile ducts and congenital hepatic fibrosis are also present. This process can be either diffuse or segmental or may be limited to one lobe of the liver, more commonly the left lobe (3). Caroli's disease is sporadic, whereas Caroli's syndrome is generally inherited in an autosomal recessive manner. Caroli's syndrome is often associated with autosomal recessive polycystic kidney disease (ARPKD).

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CASE REPORT

A baby girl of 6 months was admitted in the department of pediatrics, Bangabandhu Sheikh Mujib Medical University (BSMMU), with the complaints of gradual distension of abdomen since birth. She was the first child from a consanguineous marriage of third degree. On examination, she was oriented, afebrile and having normal vital signs with distended abdomen. Abdominal palpation revealed hepatomegaly with the liver extending 2 cm below the costal margins (Figure-1) and other systems was normal. Laboratory findings showed, Hemoglobin 10.1 g/dl (normal range 13.5±1.3 gm/dl), ESR- 65 mm/1st hour (normal range 1-10 mm/1st hour), RBC- 3.43 X 10^12/L (normal range 4.3 ±0.5 X 10^12/L), WBC- 18.50 X 10^9/L (normal range 7 ± 3 X 10^9/L). Liver enzymes were raised (AST – 48 U/L, ALT- 70 U/L) with normal serum bilirubin. Gamma Glutamyl Transferase (GGT) is 400 IU/l (normal range between 20 and 360 IU/l) and C reactive protein levels 30 mg/L (normal range 1.5–20 mg/L). Chest X-ray was suggestive of right sided pneumonitis. Abdominal ultrasound revealed mild hepatomegaly with grossly dilated CBD and CHD forming fusiform pattern. dilatation of CBD

and CHD which producing sac like structures. Intra-hepatic biliary channels are also grossly dilated and producing multiple sacs like structures (Figures 1, 2, 3).

MRCP revealed gross fusiform dilatation of CBD and CHD (Figures 4, 5). Intrahepatic biliary trees are also grossly dilated. Further diagnostic procedures were not performed because the Ultrasound imaging and computed tomography (CT) were considered sufficient. Liver biopsy was done to confirm the diagnosis. On histopathological examination there was cholestasis, hepatocellular damage, parenchymal inflammation, bile duct proliferation and inflammation with central vein dilatation.

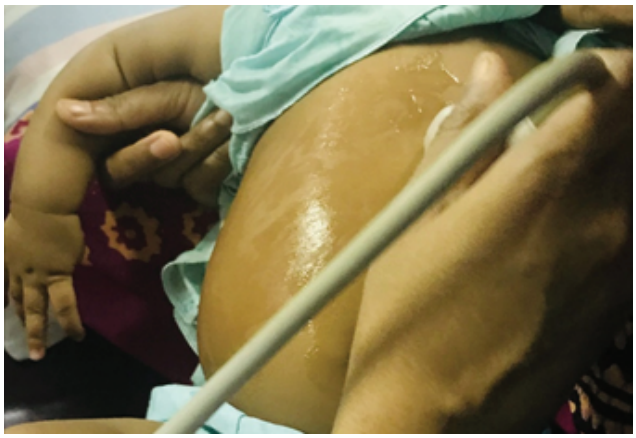

Figure 1: Visibly distended abdomen of the baby during ultrasound examination

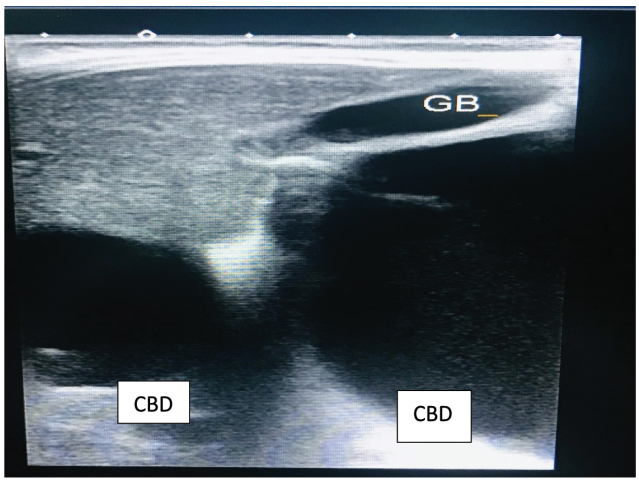

Figure 2: Oblique image of the liver showing normal appearing gallbladder and gross fusiform dilatation of CBD and CHD which producing sac like structures

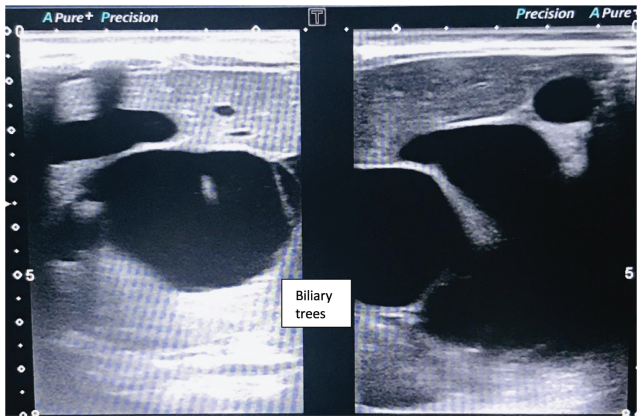

Figure 3: Oblique image of the liver showing gross fusiform dilatation intra-hepatic biliary channels and producing multiple sac like structures

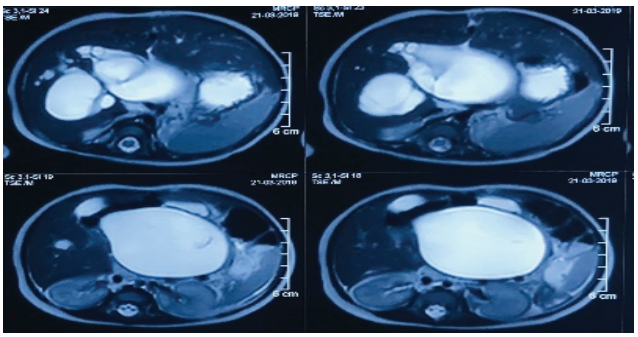

Figure 4: MRCP image showing a cluster of serpingineous hypodensities in the posterior segment of the right lobe, suggesting that these represent dilated, tortuous CBD and CHD.

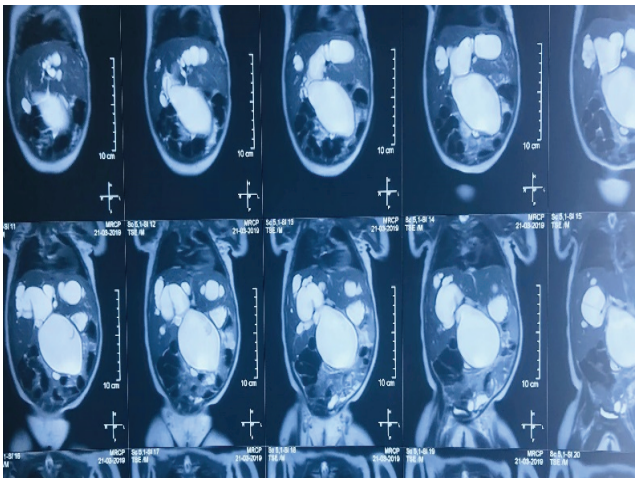

Figure 5: MRCP image shows a cluster of serpingineous hypodensities in the posterior segment of the right lobe, suggesting that these represent dilated, tortuous CBD and CHD

DISCUSSION

Caroli's disease is characterized by multiple segmental cystic or saccular dilatations of intrahepatic bile ducts. The clinical features of this syndrome reflect both the characteristics of congenital hepatic fibrosis such as portal hypertension and that of Caroli's disease named as recurrent cholangitis and cholelithiasis. The diagnosis depends on both histology and imaging methods which can show the communication between the saccules and the bile ducts. Treatment consists of symptomatic treatment of cholangitis attacks by antibiotics, some endoscopic, radiological and surgical drainage procedures and surgery. Liver transplantation seems the ultimate treatment for this disease. Prognosis is fairly good unless recurrent cholangitis and renal failure develops. Patients with caroli's disease and syndrome suffering from relapsing episodes of cholangitis with the severe danger of bacteremia and sepsis. It is rarely present in childhood and the diagnosis usually made at an advanced age (4,5,6), although there are rare case reports published of even neonatal presentation of the disease (7). Caroli's disease results from an arrest in ductal plate remodeling at the level of the larger intrahepatic bile ducts. In contrast, caroli's syndrome develops when the full spectrum of bile duct differentiation is affected, such that smaller interlobular ducts are involved and congenital hepatic fibrosis develops. Polycystic liver disease could be a possible diagnosis, but in this condition although large cysts developed from the biliary tree, they did not obstructed the bile ducts. In most patients, kidneys are similarly affected with cysts, which may cause high blood pressure and kidney failure. The tendency to form the cysts is probably present at birth, but usually do not enlarge and cause problems until adulthood (8). Our patient had normal blood pressure and his kidneys were normal in shape and size, with normal function. Von meyenberg complex could be another possible diagnosis, which is a rare condition characterized by multiple small hepatic cysts within the parenchyma at a distance from peri-portal regions and no communications with bile ducts (9). In this reported case, dilatation of intrahepatic bile ducts and hepatomegaly were the prominent findings. The mode of transmission is generally claimed to be autosomal recessive although another study suggests an autosomal dominant inheritance (10).

Modern noninvasive imaging techniques allow the diagnosis to be made more easily and without invasive imaging of the biliary tree (11). Images taken by USG, CT scan, X-ray, or MRCP show dilated intrahepatic (in the liver) bile ducts. On a CT scan, CD can be observed by noting the many fluid-filled, tubular structures extending to the liver (12). Using an ultrasound, tubular dilation of the bile ducts can be seen. When the intrahepatic bile duct wall has protrusions, it is clearly seen as central dots or a linear streak. CD is commonly diagnosed after this "central dot sign" is detected on ultrasound or CT scan (13).

CONCLUSION

The clinical presentation of CD is variable and outcome depends on the extension of the disease and rapid diagnosis of complications, with frequent ultrasound follow-ups and liver function tests. Bedside ultrasonography plays major role in meticulous diagnosis of atypical cases.

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