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RESEARCH ARTICLE

CAROLI'S SYNDROME- A RARE CASE REPORT

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Abstract

Caroli's disease and caroli's syndrome are two rare congenital diseases of intrahepatic bile ducts. Caroli's syndrome characterized by saccular dilatation of intrahepatic bile ducts associated with congenital hepatic fibrosis. We present a case of 4 year male child who came with gradual distension of abdomen since birth with mild jaundice. Ultrasonography of whole abdomen revealed dilatation of intrahepatic bile ducts. Liver biopsy was done to confirm the diagnosis. On histopathological examination there was cholestatic, fibrotic and dilated intrahepatic bile ducts on H&E and Masson trichome stain. Caroli's syndrome develop due to defect in bile duct differentiation which affect the smaller interlobular ducts and congenital hepatic fibrosis. Caroli's disease confers an approximately 7% risk of malignancy. When disease is localized to one hepatic lobe, hepatectomy relieves the symptoms and appears to remove the risk of malignancy. In diffuse caroli's disease, liver transplantation is treatment of choice.

INTRODUCTION

Caroli's disease and caroli's syndrome are two rare congenital diseases of intrahepatic bile ducts[1]. Caroli's syndrome characterized by saccular dilatation of intrahepatic bile ducts associated with congenital hepatic fibrosis. 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 malformations of small bile ducts and congenital hepatic fibrosis are also present. This process can be either diffuse or segmental and may be limited to one lobe of the liver, more commonly the left lobe[2]. Caroli's disease is sporadic, whereas Caroli's syndrome is generally inherited in an autosomal recessive manner. As with congenital hepatic fibrosis, Caroli's syndrome is often associated with autosomal recessive polycystic kidney disease (ARPKD). A rare association with autosomal dominant polycystic kidney disease (ADPKD) has also been reported.

CASE REPORT

A 4 year-old boy was admitted to the department of pediatrics of our hospital, because of jaundice and gradual distension of abdomen since birth. He was having no known disease before. He was second child from a consanguineous marriage of third degree. On general examination he was having some pallor, icterus. Heart rate was 102/ min, BP was 90/62 mm hg. On systematic examination the child was having hepatosplenomegaly, liver (+6 cm) and spleen (+3 cm) under costal margins (fig-1). The physical examinations of other systems were normal. The laboratory findings showed TLC-16,000, DC-N67, L30, E3. Serum bilirubin was 7 total, 2 direct. Liver enzymes were raised. Chest X-ray showed increased bronchovascular markings in both lungs field. Ultrasound of abdomen

revealed dilatation of intrahepatic bile ducts and hepato-splenomegaly with ascites (fig-2). Liver biopsy was performed to find the cause of dilatation of intrahepatic bile ducts. Histosection shows cholestasis in the intrahepatic bile duct and dilated intrahepatic bile duct with hepatic nodules formation (Figure 3). Histosection shows fibrosis around intrahepatic bile ducts and hepatic nodules formation on Masson trichome and reticulin stain (Figure 4,5,).

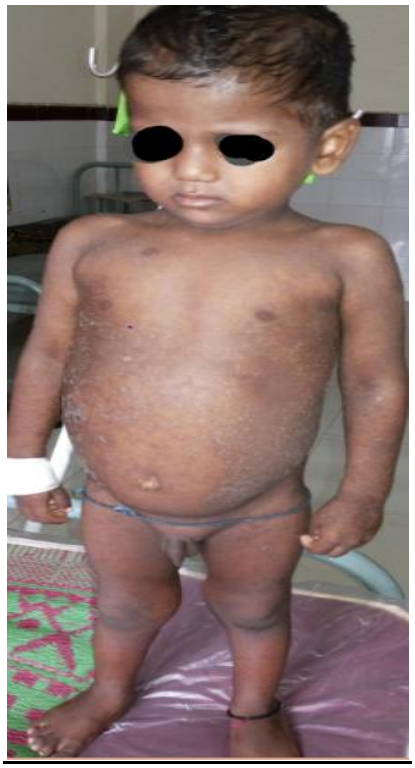
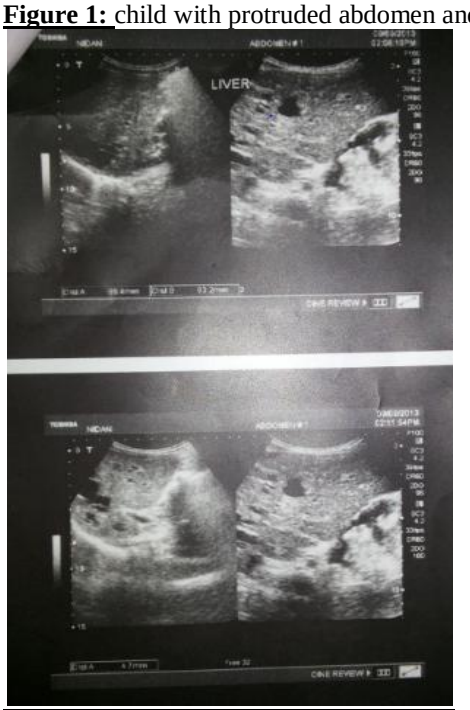


Figure 1: child with protruded abdomen and short stature



(fig-2 : USG showing hepatomegaly and intrahepatic cyst)

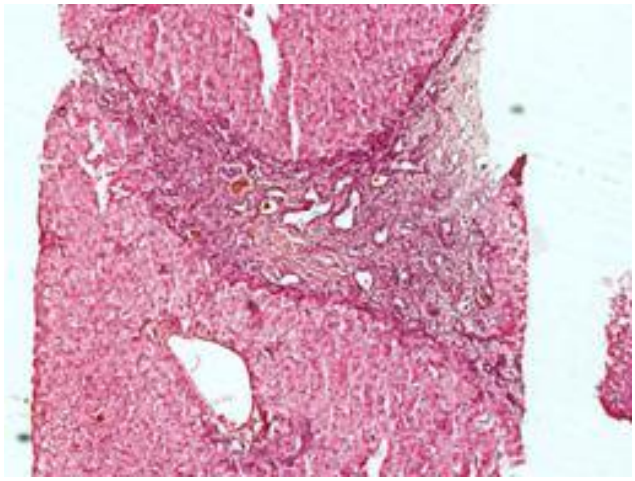


Figure 3: (Photomicrograph) cholestasis in intrahepatic bile duct and dilated intrahepatic bile duct (10X H/E)

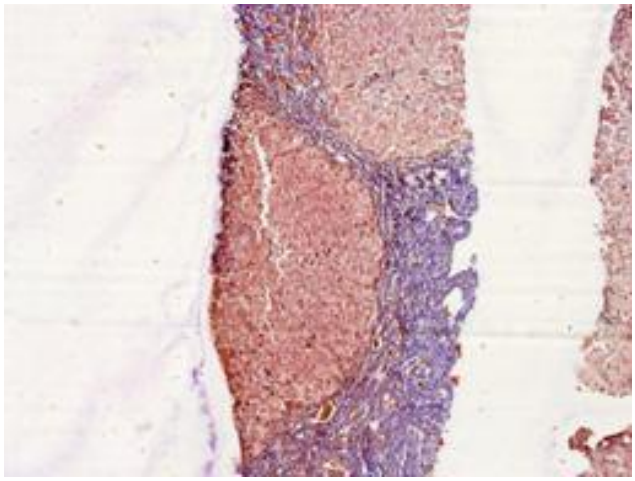


Figure4: (Photomicrograph) cholestasis and hepatic nodules formation(40X Masson trichome)

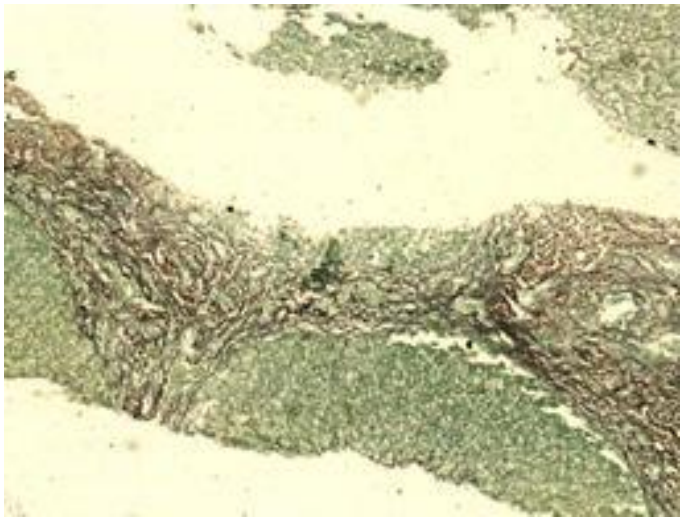


Figure 5: (Photomicrograph) fibrosis in intrahepatic septa and hepatic nodules(40X reticulin stain).

DISCUSSION

Patients with caroli's disease/syndrome suffer 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 obstruct 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 peribiliary portal regions and no communications with bile ducts[9]. In our patient dilatation of intrahepatic bile ducts and hepato-splenomegaly with ascites was the prominent finding.

The mode of transmission is generally claimed to be autosomal recessive[10], although one study suggests an autosomal dominant inheritance[6]. The process of congenital hepatic fibrosis in Caroli's syndrome patients is dynamic and progressive. Caroli's syndrome may be associated with autosomal recessive polycystic kidney disease, renal failure or cardiac disease. Autosomal recessive polycystic kidney disease is characterized by nonobstructive fusiform dilatation of the renal collecting ducts, ductal plate malformation of the intrahepatic bile ducts and fibrosis of both liver and kidneys[11,12]. Caroli's disease confers an approximately 7% risk of malignancy. The treatment depends on the clinical features and the location of the biliary abnormality. When disease is localized to one hepatic lobe, hepatectomy relieves the symptoms and appears to remove the risk of malignancy. In diffuse caroli's disease, liver transplantation is treatment of choice. The usual complications of caroli's disease/syndrome are cholestasis, cholangitis, choledocholithiasis and cholangiocarcinoma[13].

CONCLUSION

In patients with caroli's disease and syndrome, the long-term prognosis is determined by mainly by the frequency and the gravity of the episodes of cholangitis that can lead to sepsis and death or creation of hepatic abscesses. Hepatic insufficiency can develop and transplantation of liver may be required. Malignancies are also possible complications of caroli's disease/syndrome as cholangiocarcinoma risk is 100-fold increase in comparison with general population[14]. Amyloidosis is also described as a complication of caroli's disease/syndrome[15]. The clinical presentation and the outcome of the disease is variable and depends upon extension of the disease and rapid confrontation of complications, with frequent follow-up of examination of liver function and U/S control for the existence of stones and the early recognition of the probable associated conditions.

REFERENCES:

1. Caroli, J., et al. "La dilatation polykystique congénitale des voies biliaires intrahépatiques: essai de classification." *Sem Hop Paris* 34.2 (1958): 2128-2135.

2. Taylor, A. C., & Palmer, K. R. (1998). Caroli's disease. *European journal of gastroenterology & hepatology*, *10*(2), 105-108.

3. Yoshizawa, Kaname, et al. "Caroli's disease in three siblings."*Gastroenterologia Japonica* 27.6 (1992): 780-784.

4.Tsuchida, Y., et al. "Evaluation of long-term results of Caroli's disease: 21 years' observation of a family with autosomal" dominant" inheritance, and review of the literatu re." *Hepato-gastroenterology* 42.2 (1995): 175-181.

5. Keramidas, D. C., G. P. Kapouleas, and G. Sakellaris. "Isolated Caroli's disease presenting as an exophytic mass in the liver." *Pediatric surgery international* 13.2-3 (1998): 177-179

6. Fagundes-Neto, Ulysses, et al. "Caroli's disease in childhood: report of two new cases." *Journal of pediatric gastroenterology and nutrition* 2.4 (1983): 708-711.

7.Keane, F., et al. "Neonatal presentation of Caroli's disease." *Archives of Disease in Childhood-Fetal and Neonatal Edition* 77.2 (1997): F145-F146.

8. Itai, Yuji, et al. "Hepatobiliary cysts in patients with autosomal dominant polycystic kidney disease: prevalence and CT findings." *AJR. American journal of roentgenology* 164.2 (1995): 339-342.

9. Maher, M. M., et al. "Bile duct hamartomas (von Meyenburg complexes): value of MR imaging in diagnosis." *Abdominal imaging* 24.2 (1999): 171-173.

1o. Mortelé, Koenraad J., and Pablo R. Ros. "Cystic Focal Liver Lesions in the Adult: Differential CT and MR

Imaging Features 1." *Radiographics* 21.4 (2001): 895-910.
11. Turkbey, Baris, et al. "Autosomal recessive polycystic kidney disease and congenital hepatic fibrosis (ARPKD/CHF)." *Pediatric radiology* 39.2 (2009): 100-111.
12. Lonergan, Gael J., Roy R. Rice, and Eric S. Suarez. "Autosomal Recessive Polycystic Kidney Disease: Radiologic-Pathologic Correlation 1: (CME ava ilable in print version and on RSNA Link)." *Radiographics* 20.3 (2000): 837-855.
13. Hussain, Sunny Zaheed, David A. Bloom, and Vasundhara Tolia. "Caroli's disease diagnosed in a child by MRCP." *Clinical imaging* 24.5 (2000): 289-291.
14. Taylor, A. C., & Palmer, K. R. (1998). Caroli's disease. *European journal of gastroenterology & hepatology*, *10*(2), 105-108.
15. Gupta, Ashish K., et al. "Caroli's disease." *The Indian Journal of Pediatrics*73.3 (2006): 233-235.