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Congenital Hepatic Fibrosis - Case Report

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ABSTRACT

Congenital hepatic fibrosis is a heterogenous group of autosomal recessively inherited disorder due to defective remodelling of bile ductal plate. It is often seen in association with hepatorenal ciliopathies and syndromic fibrocystic abnormalities. Age of presentation and symptoms varies among the individuals which plays an important role in prognosis. Liver biopsy is the gold standard for diagnosis, characterized by abnormal excessive small bile duct proliferation with persistence of embryonic bile duct termed as Ductal plate malformation and periportal fibrosis of variable thickness and preserved hepatic lobules. We present a case of Autosomal recessive polycystic kidney disease and Congenital hepatic fibrosis in 11 years male child with growth retardation.

Keywords: Congenital hepatic fibrosis, Ductal Plate Malformation.

INTRODUCTION

Congenital hepatic fibrosis (CHF) is a developmental disorder of the portobiliary system, characterized by ductal plate malformation and diffuse periportal fibrosis. It is a heterogenous group of Autosomal Recessive disorder associated with hepatorenal ciliopathies. Differences exist between children and adults in terms of clinical symptoms, treatment, and outcomes. In children, the most common presentation is portal hypertension. Ascites, splenomegaly, hypersplenism, upper gastrointestinal varices, and immature embryonic bile duct persistence are all caused by excessive fibrous tissue growth in the portal areas. Hepatomegaly is a significant indicator that can be seen in virtually all patients(1). Rarely does CHF occur alone; instead, it frequently coexists with a variety of diseases brought on by different gene abnormalities, such as Caroli syndrome and Autosomal Recessive Polycystic Kidney Disease (ARPKD)(2). CHF is a multiorgan disorder requiring transplantation. Here, we present a case of congenital hepatic fibrosis with autosomal recessive polycystic kidney disease documented by imaging and histopathology.

CASE DESCRIPTION

Our patient is a 11-year-old male child, who was referred to our hospital with complaints of abdominal distension. He was the first-born child of a second-degree consanguineous marriage. He presented with vomiting and loose stools on day 3 of life and evaluated for failure to thrive. On follow up, child developed hypertension and started on regular medication. Radiological investigation CT Abdomen – Both kidneys enlarged in size, bilateral renal parenchyma replaced by multiple cysts of varying size-polycystic kidney, and hepatomegaly (Figure1). Endoscopy performed showed grade 2 esophageal varices for which endoscopic band ligation was performed. Laboratory investigation revealed ALT-17U/L, AST- 33 U /L, creatinine- 1.4mg/dl, urea-59mg/dl, coagulation study

prothrombin time- 23 seconds, plasma fibrinogen- 161 mg%. Peripheral smear Hb- 7.7gm/dl, WBC- 1800cells/cu.mm, platelets- 75000cells/cu.mm. Viral hepatitis work up shows HAV –Negative, HBV, HCV, AntiHBcAb – Non reactive. Mycobacterium Tuberculosis IGRA – Negative. Varicella zoster IgG- Negative. Referred for liver transplantation in view of the portal hypertension. General physical examination revealed hepatosplenomegaly with ascites. CT abdomen shows dilatation of intrahepatic bile duct with evidence of fibrosis and cirrhosis, splenomegaly, both kidneys are enlarged in size with multiple cysts of varying sizes keeping in view the clinical status of autosomal recessive polycystic kidney disease and bilateral pleural effusion seen. Doppler shows evidence of portal hypertension.

Clinical diagnosis -Autosomal Recessive Polycystic Kidney Disease with Congenital Hepatic Fibrosis, hypersplenism and portal hypertension was made. Further proceeded with liver transplantation. Gross finding shows enlarged liver with firm to hard whitish area spanning the entire liver tissue. No focal lesions or nodules identified.

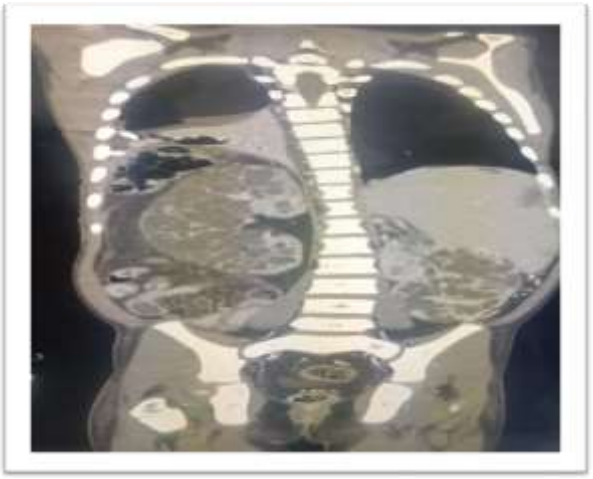

Figure 1: CT abdomen shows dilatation of intrahepatic bile duct with evidence of fibrosis and cirrhosis, splenomegaly, both kidneys are enlarged in size with multiple cysts of varying sizes.

MICROSCOPY: Congenital hepatic fibrosis (Figure 2,3&4).

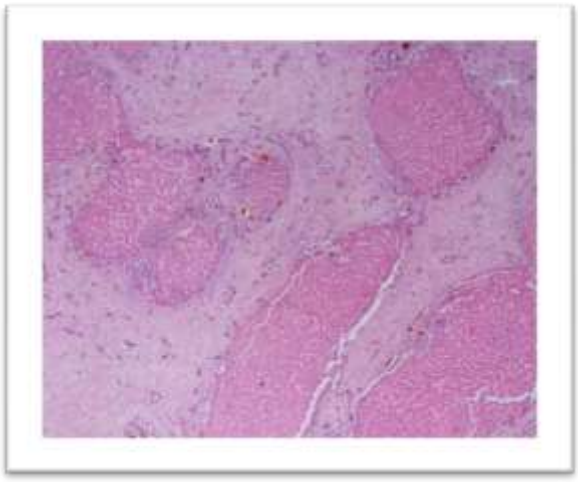

Figure 2: Low power on H&E shows diffuse portal fibrous expansion with broad fibrous septae containing spindled fibroblastic cells separating hepatocytes into multiple small nodules

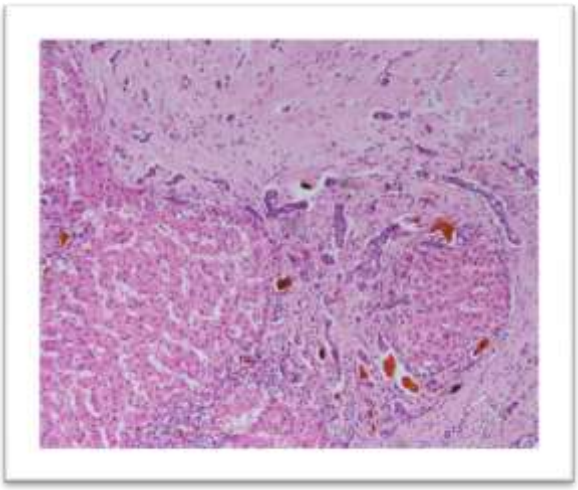

Figure 3: High power on H&E shows fibrous septae contains brisk ductular proliferation in the periphery of the lobules composed of cuboidal cells with vacuolated cytoplasm and bile plugs. Some loss of portal vein and prominent portal arteries noted. Single hepatic plate thickness with no evidence of regeneration.

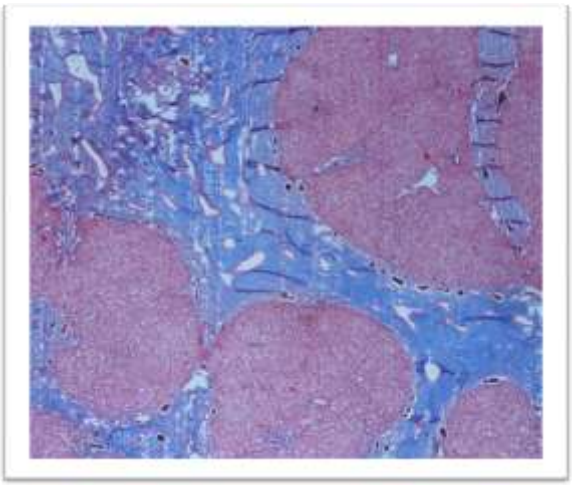

Figure 4: Low power- Masson trichrome stain confirms the above findings

DISCUSSION

CHF is a rare autosomal recessive disease described in detail by Kerr et al in 1961(3). Our patient has autosomal recessive polycystic kidney disease. Depending on the age at presentation and renal involvement ARPKD is separated into perinatal, neonatal, infantile and juvenile, of which perinatal form is severe type. Congenital hepatic fibrosis is considered as a juvenile variant of Autosomal recessive polycystic kidney disease. Patients most commonly presents with signs and symptoms of portal hypertension which include splenomegaly and variceal bleeding(4). Congenital hepatic fibrosis is usually diagnosed in early infancy or during childhood (5). However, some of these patients can be asymptomatic for many years, leading to an unexpected diagnosis of CHF in adulthood(6).The major aetiology of CHF seems to be a distortion of the ductal plate as a result of aberrant biliary system remodelling. Some cases are associated with phosphomannose isomerase deficiency leading to hypoglycosylation which affects the remodeling of bile ductal plate(7) The stage of biliary abnormalities during fetal development greatly influences the clinical and pathologic characteristics . In Caroli disease, however, the larger intrahepatic bile ducts are impacted. It is feasible to explain the high prevalence of Caroli disease and CHF cases by the fact that different intrahepatic bile duct segments are affected by ductal plate distortion. The connections between CHF along with related ductal plate malformation diseases, such as choledochal cysts, Von Meyenburg complex (bile duct hamartoma), and Caroli disease, have been well reported in the literature.(8). Anks6 knock-out mice study explained that the Hippo-YAP/TAZ angiogenesis pathway was active during the development of the bile duct(9) suggested a finely tuned interaction between the portal veins, ductal plates, and the intervening mesenchyme during ductal plate remodelling provided a nice illustration of the molecular mechanisms underlying this "vein-duct" interaction. Congenital hepatic fibrosis can only be definitively diagnosed through histological analysis. Although hepatic fibrosis is a dynamic, reversible process, many researchers are still looking for medications that could stop the disease's progression, which is ineffective in humans(10). The primary goal of CHF therapy is to alleviate the challenges the condition causes(11). Liver transplantation is a curative treatment of choice as performed in our case(12). Few studies show liver transplantation will improve the renal function(13).

CONCLUSION

Congenital hepatic fibrosis in an isolated form is very rare, and always occur in association with other fibrocystic disease especially polycystic kidney disease. A multidisciplinary correlation and systematic evaluation are important to make the correct diagnosis. Liver biopsy is the gold standard for diagnosis. CHF should be differentiated from other causes portal hypertension and cirrhosis.

Declaration by Authors

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Conflict of Interest: The authors declare no conflict of interest.

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