CASE REPORT

Congenital Hepatic Fibrosis- A Case Report

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Abstract

CHF is one of the "fibropolycystic diseases" and is a rare disease of children and young adults. Patients usually present with signs of portal hypertension with frequent renal involvement. There is relative preservation of liver function and underlying architecture. Major complaints are due to hepatosplenomegaly and portal hypertension. We present a case of 16 year old male patient who presented with pain in abdomen, splenomegaly and signs of portal hypertension. LFTs were mildly deranged with pancytopenia. CT scan revealed moderate splenomegaly, mild hepatomegaly with renal cortical multiple bilateral cysts. Splenectomy was performed with wedge liver biopsy. Histopathological examination revealed portal tract widening with ductular proliferation along with fibrocongestive splenomegaly. Based on the clinical, radiological and histopathological findings, diagnosis of CHF with fibrocongestive splenomegaly was made.

***Keywords**: Congenital hepatic fibrosis, portal hypertension, splenomegaly, renal cortical cysts*

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Introduction

Congenital hepatic fibrosis is an inherited, congenital malformation that is characterized by fibrotic portal spaces that contain multiple bile ductules which leads to portal hypertension. CHF is one of the "fibropolycystic diseases". This is a multi-organ affecting condition characterized by polycystic liver disease, congenital hepatic fibrosis, caroli's disease, choledocal cysts and renal cysts.[1-3] The disease appears in both sporadic (in many as 56% cases) and familial patterns. Arrest of maturation and the lack of remodelling of the ductal plate engenders persistence of an excess number of immature embryonic duct structures which in turn stimulates the formation of portal fibrous tissue conferring the clinical picture of recurrent cholangitis or portal hypertension and associated symptoms.[4]

Case Report

We report a case of 16 years male child who presented with pain in abdomen and intermittent fever since 6 months. Pain was located in left hypochondirac region. USG abdomen reveal moderated splenomegaly with non-obstructing renal calculi. Upper GI endoscopy showed gastric varieces. CT abdomen showed gross splenomegaly with mild hepatomegaly and bilateral non obstructing renal calculi with multiple cortical cysts. (Figure 1 A & B) On Investigations, hemoglobin was 6.9 g/dl, Total leucocyte count was 24 $\times 10^3$ /µl, Platelet count was 4 lakh/ µl. Liver function tests revealed total bilirubin was 2.2 mg/ dl, AST was 82 U/L, ALT was 59 U/L. Total protein was 5.7 g/dl. serum urea 95 mg/dl and serum creatinine 2.3 mg / dl. Splenectomy with wedge liver biopsy was done. Immediate post opearative period was uneventful.

Figure-1(A). CT Abdomen showing hepatomegaly, with hypodense cystic areas within the kidney

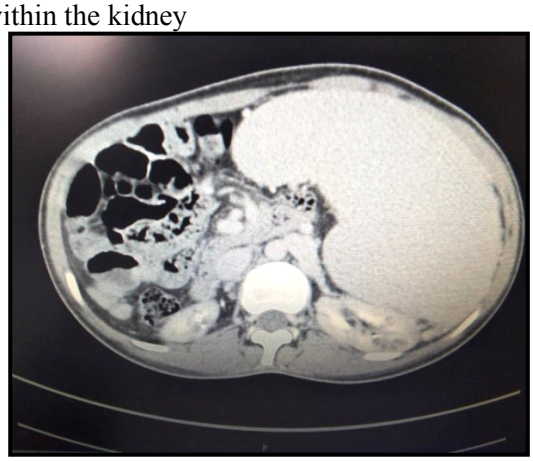


Figure 1(B). CT Abdomen showing hepatosplenomegaly

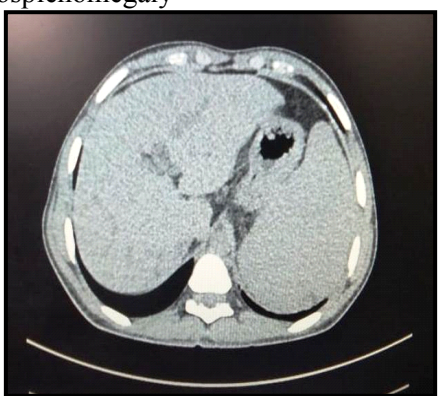


Figure 2(A). Portal area with extensive fibrosis and normal hepatocytes arrangement within nodule. (H & E stain 10x)

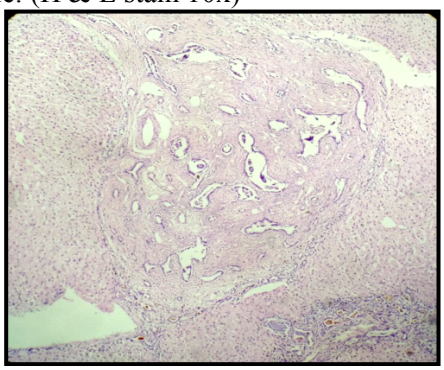
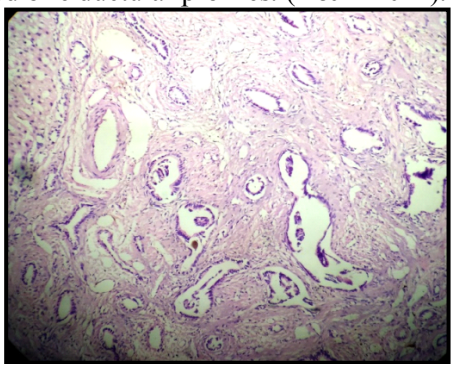


Figure 2(B). Liver biopsy with diagnostic dense fibrosis of portal tracts with numerous marginal dilated bile ductular profiles. (H& E 40 X).



We received liver biopsy measuring 2x1.5x1cm, firm in consistency and cut surface showed nodular appearance. Spleen was weighing 720 gms and measuring 20x12x5 cm. Capsule was thickened and splenic parenchyma was congested. On histopathological examination revealed liver parenchyma partially effaced by pseudolobules formation and widening of portal tracts with increase porto-portal fibrosis. The portal tracts showed irregular proliferation of large and small ducts and ductules. Some of the ducts and ductules showed bile thrombi and were dilated. Portal tract showed sparse mononuclear cell infiltration. Hepatocytes showed mild degenerative changes. (Figure 2A & B).

Spleen showed effaced architecture. Capsule was thickened and infiltrated with chronic inflammatory infiltrate. White pulp showed atrophy and red pulp showed fibroblastic proliferation with dilation of sinusoids.

With this clinical, radiological and histomorphological features the diagnosis of congenital hepatic fibrosis with fibrocongestive splenomegaly was made.

Patient was under close follow up. Then, patient developed difficulty in breathing and was intubated for the same. After few days, ascites developed. Subsequently patient's condition was deteriorated and patient died of respiratory failure.

Discussion

Bile duct proliferation is thought to be a form of biliary dysgenesis arising from arrested biliary development.[2,5,6] The disproportionate overgrowth of biliary epithelia and their supporting connective tissue might be the cause of proliferated and ecstatic bile ductal changes.[7] The primary disorder of congenital hepatic fibrosis is most likely to be bile ductular proliferation, with fibrosis being secondarily induced by multiple bile ductules.[7] This supports the hypothesis that congenital hepatic fibrosis, Caroli disease, choledocal cyst, and multiple cystic lesions of the liver and kidney may be closely related entities representing different level of involvement of the same basic congenital defect.[5]

Congenital hepatic fibrosis (CHF) is a rare, autosomal recessive disorder, clinically characterized by hepatic fibrosis and portal hypertension. CHF results from ductal plate malformation (DPM) of the intrahepatic bile ducts. Four clinical forms can be observed: portal hypertensive, cholangitic, mixed and latent. CHF is one of the "fibropolycystic diseases" which also include several conditions with a variety of intrahepatic bile duct dilatation and associated periportal fibrosis such as Caroli disease, autosomal recessive and dominant polycystic kidney disease (ARPKD or

ADPKD), Ivemark, Jeune, Joubert, Bardet-Biedl, Meckel-Gruber and Arima syndromes. Most of them are accompanied by progressive cystic degeneration of the kidneys.[8]

In congenital hepatic fibrosis, diverse clinical manifestations, such as portal hypertension, renal failure, cholangitis or mixed form can be shown with or without associated anomaly.[3,10-12] .Portal hypertension may develop relatively early in congenital hepatic fibrosis and may progress to a fatal termination by bleeding esophageal varices.[9-11] Portal hypertension in congenital hepatic fibrosis is presinusoidal and the liver function is usually preserved.[10] The long term outcome for patients with the portal hypertension is good.[12] In the present case, patient presented with signs and symptoms of the portal hypertension like ascites and splenomegaly but there was no history of jaundice. There was no similar history in family members.

Sepsis is the complication of bile stasis and intrahepatic duct stones.[6] Congenital hepatic fibrosis may present in the perinatal or neonatal period.[10]

The younger patients show more renal tubular involvement and death occurs due to renal failure. In the childhood and adult type, only a few tubules are involved and the presentation is of hepatomegaly or portal hypertension.[13, 14]

Congenital hepatic fibrosis is characterized pathologically by diffuse portal and perilobular fibrosis and multiple irregular bizarre shaped bile ductules in the area of fibrosis.[7, 14]

Congenital hepatic fibrosis may appear either sporadically or in several members of family whose parents are normal.[10] It is likely to be an autosomal recessive disease that affects both sexes.[2, 3] In the present case the exact inheritance pattern could not be assessed as other family members did not show any signs of liver or kidney involvement and were also not screened due to low socio-economic status.

Conclusion

Congenital hepatic fibrosis is an unusual congenital disorder of the liver having a heredofamilial tendency and presenting within first two decades of life. The treatment basically is for portal hypertension with secondary esophageal and gastric varises. Portosystemic shunt along with splenectomy surgery is the treatment of choice in most of the cases. Liver transplantation is considered in cases of liver failure.

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