

Case Report

Its Congenital Hepatic Fibrosis; Not Cirrhosis At All

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ABSTRACT

Congenital hepatic fibrosis is a rare condition characterized by extensive fibrosis of liver but with preserved normal lobular architecture inherited as autosomal recessive trait. We report a 19 year old female admitted to Bangabandhu Sheikh Mujib Medical University with the complaints of lump in upper abdomen since last 13 years and episodes of fever and abdominal pain for same duration. She was diagnosed with hepatic TB on hepatic histology. Congenital hepatic fibrosis is a rare cause of portal hypertension that presents during childhood. Prognosis of congenital hepatic fibrosis is good. Life threatening events in these patients are related with variceal bleeding and episodes of cholangitis. Owing to relatively good liver function these patients tolerate portosystemic shunt surgeries quite well. Though rare, congenital hepatic fibrosis should be included in the differential diagnosis of portal hypertension in early life.

Key words: Fibrosis; Ductal plate malformation; Portal hypertension

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INTRODUCTION

Congenital hepatic fibrosis is a rare condition characterized by extensive fibrosis of liver but with preserved normal lobular architecture, often occurring in association with cystic lesions of the kidney. It is inherited as autosomal recessive trait. The underlying abnormality lies in the development of biliary ductules, classically described as "Ductal Plate Malformation".^{1,2} It commonly presents in childhood with features of portal hypertension nevertheless incidental diagnosis in adulthood or even autopsy diagnosis has been reported³. Though frequently confused with liver cirrhosis², congenital hepatic fibrosis do not present with feature of hepatocellular insufficiency.¹ Absence of

signs of liver failure may rather indicate possibility of congenital hepatic fibrosis in those who are mistakenly considered to have liver cirrhosis. Here we report a case of congenital hepatic fibrosis that was misdiagnosed and treated as a case of liver cirrhosis for 13 years.

CASE REPORT

A, 19 year patient was admitted to Bangabandhu Sheikh Mujib

Medical University with the complaints of lump in upper abdomen since last 13 years and episodes of fever and abdominal pain for same duration. Abdominal lump was located in the right upper quadrant and did not increased in size over time. She complained of pain over the lump which on most occasions was accompanied by fever that used to occur one to two times a year and lasted over four to fifteen days. Though on occasions the fever was associated with chills and rigors, she did not ever have jaundice or itching, dysuria or hematuria. She was diagnosed as a case of chronic liver disease since the age of 5 yrs on the basis of firm hepatomegaly and presence of oesophageal varices grade 2 and she was put on propranolol therapy since then. She never had features of decompensation, hematemesis and malena or altered sensorium. She had apparently good appetite and did not have loss of weight or inability to gain weight. Her parents had consanguineal marriage and she was the only live child, four of her siblings did not thrive enough to stay alive beyond first few days of delivery. Her family history was negative and she had normal menarche and menstrual history. On examination, she was of average built and nutrition, mildly anaemic but did not have any stigma of liver disease. Her vitals were normal and on abdominal examination, she had palpable hard liver measuring liver span of 14cm which was non tender and had sharp margins. Splenomegaly was also noted about 5 cm below costal margin but no shifting dullness. Examinations of other systems were normal.

On investigation her hemoglobin was 9.3gm%, platelet count of 1, 90,000/mm³, total white cell count of 4,000/mm³ and ESR 80mm/hr. Blood film showed microcytic hypochromic anaemia. Her bilirubin serum transaminases, alkaline phosphatase were normal. Her prothrombin time and serum albumin were within normal range. Viral markers for hepatitis C and hepatitis B virus including Anti HBc (Total) were negative. No KF ring was noted in slit lamp examination of cornea and copper studies were normal. Her Anti nuclear antibody was positive but with titre of 18.5 U/ml (normal <10 U/ml) but AMA and Anti Sm antibodies were negative. Her urine examination was normal and her creatinine clearance was within normal range. Hemoglobin electrophoresis showed normal hemoglobin pattern. Direct agglutination and immunochromatography for Kalaazar were negative. CT scan of abdomen revealed bilateral polycystic kidneys with hepatosplenomegaly and multiple splenic collaterals were noted (fig.1). X-ray of chest was normal. Liver biopsy was done which showed broad fibrous band containing many bile ductules, blood vessels and small number of chronic inflammatory cells. Wide fibrotic bands were seen joining portal tracts but no central to portal fibrosis noted. Lobular architecture was maintained (fig.2). Findings were compatible with congenital hepatic fibrosis. She remained in apparently good health till the date of submission of this report (two years since her diagnosis).

DISCUSSION

Congenital hepatic fibrosis is a rare cause of portal hypertension that presents during childhood. Consequently, the manifestation is mainly that of portal hypertension and is devoid of features of hepatocellular failure. Primary defect lies in the involution of ductal plates that forms bile ducts resulting into persistence of the ductal plate with an increase in duct elements and an increase in portal fibrous tissue called as Ductal Plate Malformation.⁴ Though the underlying pathogenesis mainly involves ductal system, the final result is diffuse fibrotic reaction involving the portal areas separating the remaining liver parenchyma. Of note, the fibrotic

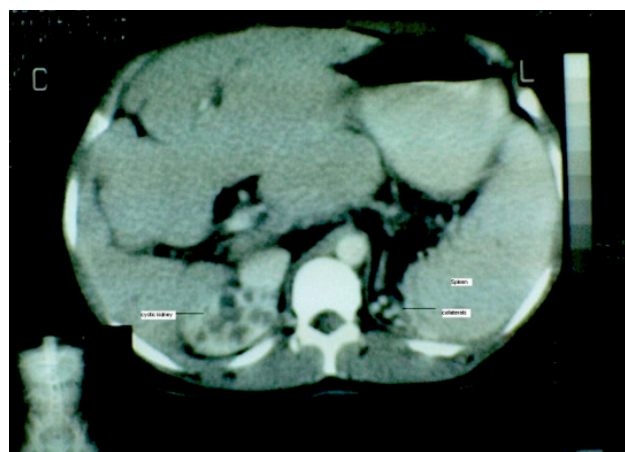


Figure 1: CT scan of upper abdomen of patient showing enlarged liver and spleen with polycystic kidneys. There are multiple collaterals in left paravertebral region

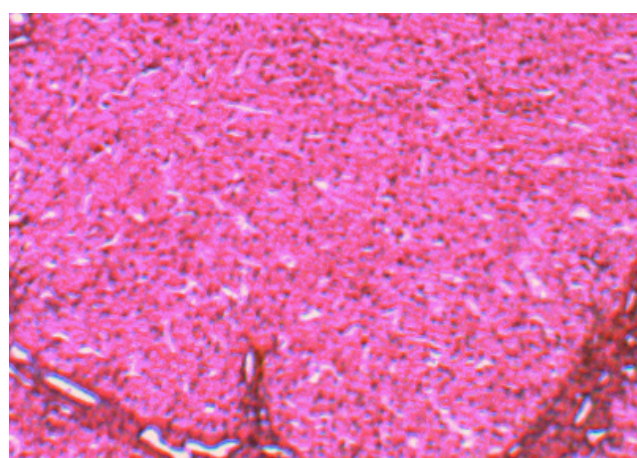


Figure 2: Liver biopsy showing ectatic bile ducts, portal to portal fibrosis but intact lobular architecture and central vein (HE stain X 100).

bands contain abnormal or ectatic bile ducts and numerous portal vessels commonly called as Von Meyer's Complexes.²

Seventy seven percent of patients with congenital hepatic fibrosis have esophageal varices at presentation.⁵ Rupture of esophageal varices and subsequent bleeding is the commonest presentation. However other presentations like cholangitic and mixed cholangitic and portal hypertensive varieties also occur. Association with intra hepatic choledocal cyst has been seen and such cases are termed as Caroli's syndrome.² Medullary sponge kidney and polycystic kidney disease are the most common associated abnormalities. Presentation in such association is further complicated by occurrence of hypertension, renal disease, pulmonary emphysema, cerebellar hemangioma and berry aneurysms of the arterial system of brain.¹ Congenital hepatic fibrosis can have a wide variety of manifestations and presentations.

Prognosis of congenital hepatic fibrosis is seen to be good.⁶ Life threatening events in these patients are related with variceal bleeding and episodes of cholangitis.⁷ Owing to relatively good liver function these patients tolerate portosystemic shunt surgeries quite well. But reports of development of nodular transformation later in life cannot be neglected.⁷ Other factors that may guard prognosis are associated renal disease and their complications.

In our case, the suspicion of congenital hepatic fibrosis was

raised when polycystic kidneys were noted in association with long standing portal hypertension and relatively preserved liver function. Repeated episodes of abdominal pain and fever in our case were later related with cholangitis. The reason she did not bleed from varices may be that she was prescribed oral propranolol since detection of her varices and this might have masked the presentation. The case we report here was mixed cholangitic and portal hypertensive type. Her variceal size was medium sized and no associated red weal markings were noted. In background of preserved liver function her variceal characteristics appeared benign and were controlled by propranolol alone. ERCP to visualize biliary tree can sometimes detect biliary abnormalities that are inapparent on other imaging techniques. Cholangiogram however is also known to be an inciting event that may trigger overt manifestations and complications in other wise asymptomatic individuals⁸ This fact together with lack of consent for the procedure, ERCP was not done in our case. Recurrent cholangitis in this patient was also benign and patient was doing well with oral antibiotics (ciprofloxacin) during such episodes. Repeated episodes of cholangitis and ectatic biliary tree may precipitate biliary stones. Chenodeoxycholate have been recommended for

preventing biliary stones.¹ Treatment with ursodeoxycholic acid (UDCA) for cholangitic variety of congenital hepatic fibrosis is also recommended by some authors.⁹ It may also improve bile salt dependent bile flow and prevention of biliary stone formation. Further management plans in this case include regular surveillance of varices and management of cholangitic episodes along with prophylaxis for biliary stone formation.

CONCLUSIONS

Though rare, congenital hepatic fibrosis should be included in the differential diagnosis of portal hypertension in childhood. Relatively preserved liver function and hard non-tender hepatomegaly, splenomegaly along with ballotable kidneys should further obviate the need to consider this diagnosis.

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REFERENCES

1. Witzleben CL, Ruchelli E. Hepatology: a text book of liver disease, 4th ed. Philadelphia: Saunders; 2003. p.1461-80.
2. Sherlock S, Dooley J, editor. Diseases of the Liver and Biliary system, 11th ed. Oxford: Blackwell Science; 2002. p.583-96.
3. Fauvert R, Benhamou J. The Liver and Its Diseases. New York: Intercontinental Medical Book; 1974. p.283.
4. Desmet VJ. Congenital diseases of intrahepatic bile ducts: variations on the theme "ductal plate malformation". Hepatology. 1992;16(4):1069-83. [Crossref](#)
5. Alvarez F, Bernard O, Brunelle F, Hadchouel M, Leblanc A, Odievre M et al. Congenital hepatic fibrosis in children. J Pediatr 1981;99:370-5. [Crossref](#)
6. Kerr DNS, Okonkwo S, Choa RG. Congenital hepatic fibrosis: The long-term prognosis. Gut 1978;19:514-20. [Crossref](#)
7. Benhamou JP, Menu Y. Oxford Textbook of Clinical Hepatology, 2nd ed. Newyork: Oxford University Press; 1999. p.817-23.
8. Lam SK, Wong KP, Chan PK, Ngan H, Todd D, Ong GB. Fatal cholangitis after endoscopic retrograde cholangiopancreatography in congenital hepatic fibrosis. Aust N Z J Surg 1978;48(2):199-202. [Crossref](#)
9. Nazer H. Congenital Hepatic Fibrosis. emedicine. (Accessed on Aug. 6, 2017). [Crossref](#)