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A case of congenital hepatic fibrosis: first report from Bangladesh

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Врожденный фиброз печени: первое сообщение из Бангладеш

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Keywords: congenital hepatic fibrosis, hepatosplenomegaly

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 reported [3]. Though frequently confused with liver cirrhosis [2], 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

MA, 19year old 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 five years 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 melena 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

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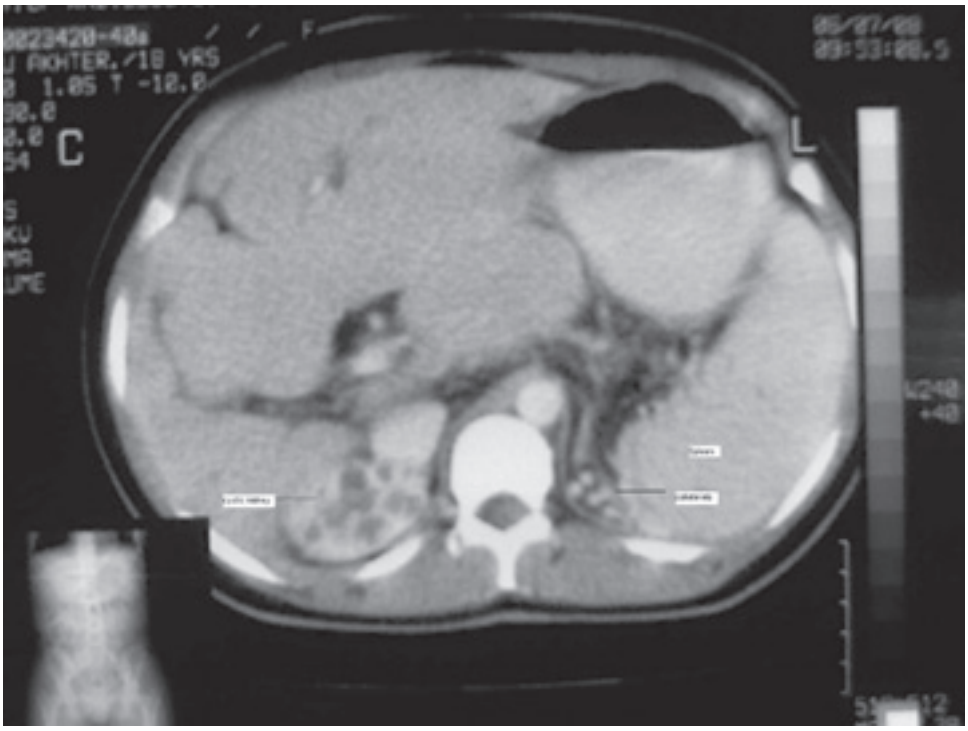

Figure 1: CT scan of upper abdomen of patient

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 firm liver measuring liver span of 14cm which was non tender and had sharp margins. Splenomegaly was also noted about 5 cm below costal margin and no shifting dullness. Examination of other systems was normal.

On investigation her hemoglobin was 9.3 gm%, platelet count of 190,000/mm3, total white cell count of 4,000/mm3 and ESR 80. 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 Kala-azar were negative. CT scan of abdomen revealed bilateral polycystic kidneys with hepatosplenomegaly and multiple splenic collaterals were noted. X-ray of chest were 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 separated hepatic parenchyma into small nodules- findings compatible with congenital hepatic fibrosis.

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 bands contains 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 choledochal 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]. Therefore it can be estimated that 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

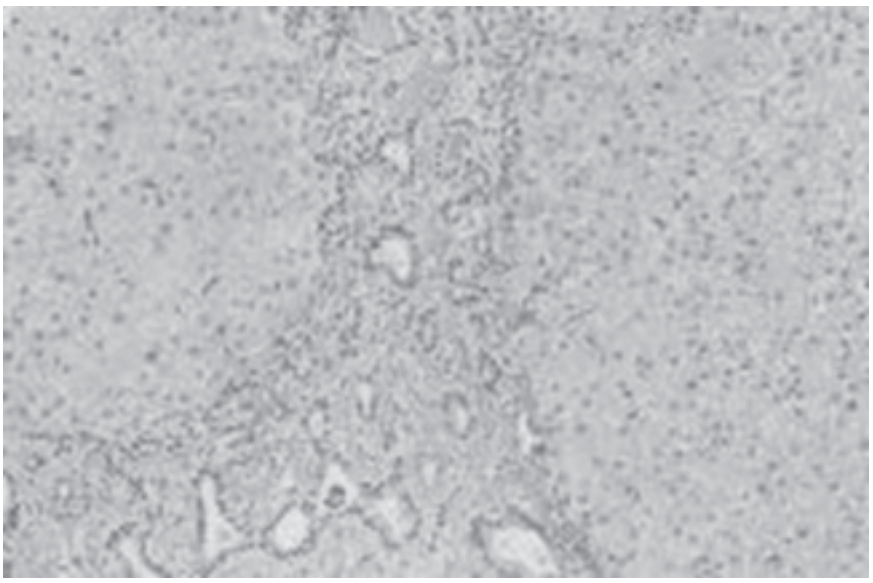

Figure 2: Liver histopathology of patient

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 longstanding 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 individuals [8].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 with cholangitic presentation is also recommended by some authors [9]. It may improve bile salt dependent bile flow. It may also have some role in preventing biliary stone formation and so was prescribed in this case. Further management plans in this case include regular surveillance of varices and management of cholangitic episodes along with prophylaxis for biliary stone formation.

Conclusion

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

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Врожденный фиброз печени: первое сообщение из Бангладеш

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Вступление. Врожденный фиброз печени является редким состоянием, которое характеризуется обширным фиброзом печени, но с сохраненной нормальной лобулярной архитектурой, которое наследуется по аутосомно-рецессивному признаку.

История болезни: мы сообщаем о 19-летней женщине, которая обратилась в медицинский университет Bangabandhu Sheikh Mujib с жалобами на комок в верхней части живота последние 13 лет, эпизоды лихорадки и боли в животе той же продолжительности. Проведено гистологическое исследование печени и поставлен диагноз.

Обсуждение: Врожденный фиброз печени является редкой причиной портальной гипертензии, которая проявляется в детстве. Прогноз врожденного фиброза печени хороший. Опасные для жизни события у этих больных связаны с варикозным кровотечением и эпизодами холангита. Благодаря относительно хорошей функции печени эти пациенты переносят операции портосистемного шунтирования достаточно хорошо.

Заключение: Хотя врожденный фиброз печени встречается редко, данное заболевание должно быть включено в дифференциальную диагностику портальной гипертензии на ранних годах жизни.

Ключевые слова: врожденный фиброз печени, гепатоспленомегалия

Вроджений фіброз печінки: перше повідомлення з Бангладеш

Мамун-Ал-Махтаб, Шеикх Мохаммад Фазле Акбар, Камал, Салимур Рахман

Вступ. Вроджений фіброз печінки є рідкісним станом, який характеризується обширним фіброзом печінки, але зі збереженою нормальною лобулярною архітектурою, який успадковується за аутосомно-рецесивною ознакою.

Історія хвороби: ми повідомляємо про 19-річну жінку, яка звернулася до медичного університету Bangabandhu Sheikh Mujib зі скаргами на клубок у верхній частині живота останні 13 років, епізоди лихоманки і болю в животі тієї ж тривалості. Проведено гістологічне дослідження печінки і поставлений діагноз.
Обговорення: Вроджений фіброз печінки є рідкісною причиною портальної гіпертензії, яка проявляється в дитинстві. Прогноз вродженого фіброзу печінки хороший. Небезпечні для життя події у цих хворих пов›язані з варикозною кровотечею і епізодами холангіту. Завдяки відносно хорошій функції печінки ці пацієнти переносять операції портосистемного шунтування досить добре.

Висновок: Хоча вроджений фіброз печінки зустрічається рідко, дане захворювання має бути включено в диференціальну діагностику портальної гіпертензії на ранніх роках життя.

Ключові слова: вроджений фіброз печінки, гепатоспленомегалія

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Mamun-Al-Mahtab, Sheikh Mohammad Fazle Akbar, Md. Kamal, Salimur Rahman

Introduction: Congenital hepatic fibrosis is a rare condition characterized by extensive fibrosis of liver but with preserved normal lobular architecture inherited as autosomal recessive trait.

Case report: 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.

Discussion: 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.

Conclusion: Though rare, congenital hepatic fibrosis should be included in the differential diagnosis of portal hypertension in early life.

Keywords: congenital hepatic fibrosis, hepatosplenomegaly