Congenital Hepatic Fibrosis

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Congenital hepatic fibrosis (CHF) is an unusual condition in which portal hypertension (PH) occurs without significant hepatic or renal functional impairement(1). CHF is a subtype of group of congenital disorders described as fibropolycystic disease with a wide clinical spectrum depending upon the time of presentation and degree of hepatic involvement. We report a patient with CHF.

Case Report

An 8-year-old boy was admitted with slowly progressive distension of abdomen and fullness in upper abdomen of 9 months duration, and history of minor episodes of epistaxis for 7 months. There was no history of pain abdomen, jaundice, hematemesis, malena or any skin bleeds or hyperpigmentation. No umbilical catheterisation was done in the neonatal period. On tracing the pedigree no other family member was known to be affected. The boy weighed 20 kg (5th percentile of NCHS), with a height of 123 cm (25th percentile of NCHS). General examination revealed pallor and conjunctival xerosis without any signs of liver cell failure or icterus. Temperature, pulse and BP were normal. On abdominal examination spleen measured 12 cm below costal margin with tip below umbilicus, liver span was 6 cm with no evidence of free fluid in the abdomen. Kidneys were not palpable and other systemic examination was normal.

On investigations, hemoglobin was 6.8 g/dl, total leukocyte count was 4600/mm^3 with P_{67} L_{25} E_5 M_3, platelet count was 63000/mm^3, MCV was 85.2/L and peripheral blood smear revealed thrombocytopenia, leukopenia, normal erythrocytes and no malarial parasite (MP). Liver function tests revealed total biliuibin of 0.7 mg/dl (direct 0.4 mg/dl and indirect 0.3 mg/dl), serum aspartate transaminase was 73 IU/L, serum alanine transaminase was 77 IU/L and alkaline phosphatase was 1218 IU/L. Bone marrow aspiration showed erythroid hyperplasia with normoblastic reaction and no abnormal cells, LD bodies or MP. HBsAg and serology for both malaria and kala azar were negative. Ultrasound abdomen showed normal liver, kidneys and normal calibre of portal vein with no evidence of ascites. Upper GI endoscopy revealed grade II esophageal varices. Slit lamp examination of eyes was normal. Prothrombin time and PTTK were also within normal limits.

Liver biopsy (Fig. 2) showed intersections of white thick bands of collagenous connective tissue septae partially encircling liver in periportal areas. Liver lobular structure was maintained. Periportal connective tissue enclosed numerous interlobular type of ductules many of which were seen as solid cords of ductal epithelial cells without central cannalisation. There was focal mild dilatation of interlobular ductules. These histological features con-

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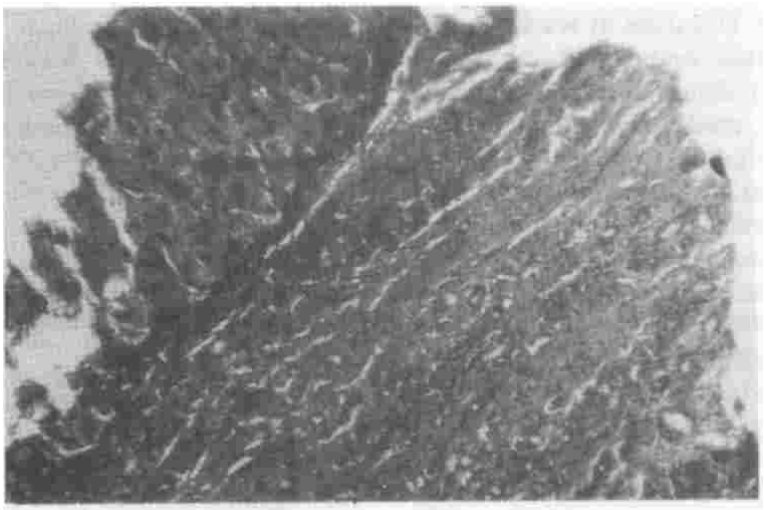

Fig. 1. Microphotograph of liver biopsy.

firmed the diagnosis of CHF and hematological picture suggested hypersplenism. During the hospital stay, the patient remained asymptomatic and had no evidence of active bleeds.

Discussion

Congenital hepatic fibrosis is a term first coined by Kerr in 1961, clinically reserved for a condition in which PH occurs without significant impairement of liver or kidney function(l). Cases have been reported from all over the world(2-7) but the exact incidence of the disease is not known. An unusual association of CHF has been reported with congenital hypoplasia of depressor anguli oris muscle in one patient from India(6). CHF has usually autosomal recessive inheritance and initial presentation may be at around 3-6 months. The presentation ranges between 1.8-14 years(8), PH is a usual accompaniment and renal involvement is seen with < 10% tubules being affected. Classically affected patients are asymptomatic until the age of 5 or 7 years when manifestations of PH or cholangitis lead to the diagnosis. Several clinical forms are described which depend on the variable predominance of PH and cholangitis. Cholangitis form of CHF is more severe and usually occurs in late childhood and adult life(9). Blyth and Ockenden(10) have divided their patients into 4 groups called perinatal, neonatal, infantile and juvenile in accordance with the age at clinical presentation. Renal involvement is maximal in perinatal group and minimal in juvenile group. Our patient had presented with PH, with no clinical or histological evidence of cholangitis and belongs to juvenile groups of this classification.

The usual presentation of CHF is with abdominal distension(4), hematemesis or melena, failure to thrive, jaundice, anemia, hepatomegaly and splenomegaly(1,8). The other features of CHF are abdominal pain (splenic infarction), fever (cholangitis in dilated ductules), ascites, etc.(2-5). CHF is particularly associated with infantile polycystic kidney disease or intrahepatic bile duct dilatation (Caroli's disease)(l). The diagnosis is based on liver functions which are well preserved, features of

hypersplenism, elevation in levels of alkaline phosphatase and gamma glutamyl transferase(1,5). Other associated disorders with CHF are medullary sponge kidney, Ivemark's Familial Dysplasia, Meckels syndrome, vaginal atresia and rarely adult type polycystic kidney disease or nephronopthises, Jenunne's syndrome, tuberous sclerosis, *etc.* These conditions were ruled out in our case by absence of other clinical features/malformations associated with these conditions and relevant investigations. Biliary hemartomas (von Meyenberg complexes) are frequently associated with CHF and are detected on histology and by imaging.

Hallmark of diagnosis is liver biopsy which shows bands of fibrous tissues often containing linear or circular spaces lined by cuboidal epthelium. There is diffuse portal and perilobular fibrosis varying in thickness but it does not distort lobular structures. The limiting plate is intact and parenchyma is separated by islands of fibrosis. There are no inflammatory changes and regenerative nodules are absent or few(8). The cholangitis form of CHF is difficult to differentiate from Caroli's disease characterized by nonobstructive dilatation of intrahepatic bile ducts occurring as an isolated abnormality without portal fibrosis. This suggests a spectrum of congenital biliary tree disease with portal fibrosis and normal calibre ducts at one end and multiple intrahepatic, even extrahepatic, dilatations without fibrosis at the other end. Overlapping of CHF and Caroli's disease has been confirmed by histological studies(9).

The management and prognosis of CHF is dependent on alimentary bleeding secondary to PH. In late childhood abdominal pain, cholangitis and features of hypersplenism complicate the problem. However, prognosis may be greatly improved by shunt surgery but survival in some patients may be limited by degree of renal failure(l).

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