A Case Report of Caroli's Disease in an Infant With Review of Literature

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The authors report an infant with bilobar Caroli's disease, a rare congenital disorder with multiple cystic dilatations of the intrahepatic bile ducts. This disease entity presents a diagnostic as well as therapeutic dilemma. Various investigations used for diagnosis include ultrasound scan, hepatobiliary scan and percutaneous transhepatic cholangiography. Treatment of this rare disorder poses a challenging problem because various conservative surgical procedures are bound to fail. Hepatic resection in case of unilobar disease, and liver transplantation for biolobar disease are the only definitive curative procedures for this problem which is otherwise associated with a high morbidity and mortality.
Key words: Congenital biliary dilatation, Caroli's disease

Caroli's disease (CD) is characterised by uni or bilobar berry-like cysts in continuity with the primary brances of main intrahepatic bile ducts[1].This rare entity was first reported by Caroli and his colleagues in 1958 who described two types of the disease[2]. A rare form which is not associated with cirrhosis and portal hypertension, and presents with recurrent pyrexial illness from cholangitis. The second more common type of CD is associated with hepatic fibrosis and may present with hepatosplenomegaly and oesophageal varices. CD may coexist with dilatation of the common bile duct or may exist independently.

The diagnosis and management of this rare disease is always a challenging problem. We report an infant with CD, along with a brief literature review.

Case Report

A 45 days old girl presented to Sir Ganga Ram Hospital for management of diarrhoea. On physical examination her liver was found to be slightly enlarged. Liver function tests showed normal bilirubin, alkaline phosphatase within upper normal limits and markedly elevated transaminases. Ultrasound scan demonstrated multiple communicating cystic spaces in both lobes of the liver (Fig. 1). The porta hepatis was normal. No dilatation of the extrahepatic bile ducts was found. The portal vein was normal. A tentative diagnosis of CD was made. HIDA scan demonstrated multiple filling defects in the liver, with delayed (24 hours) films showing progressively increasing radioactivity in these defects (Fig. 2). The gall bladder was visualisd. The scan was labelled as "inconclusive". To make a definitive diagnosis, a percutaneous transhepatic cholangiogram (PTC) was performed that showed dilated communicating tortuous intrahepatic biliary ducts distributed equally in both the lobes (Fig. 3). Delayed films showed opacification of the gallbladder and faint contrast in the gut. A diagnosis of Caroli's disease was made and a follow-up arranged with Paediatric Medical and Surgical Units. However, the patient moved to Karachi with her parents, where she died of some undetermined illness a few months later.

Discussion

Caroli's disease blongs to the group of fibrocystic disorders of the liver. It often coexists with congenital hepatic fibrosis, and, fibrocystic disease of the kidneys may also be associated to a variable extent[3]. CD is usually classfied as type V congenital biliary dilation[4], but Miyano graded it as type III C[5]. Recently Guntz et al[6] have further described four sub-types of CD: Type 1, racemose; type 2, digitiform; type 3, large cystic, and type 4, intrahepatic disease in association with choledochal cyst. The pathognomonic lesion of CD is non-obstructive biliary duct dilatation, resembling a chain of lakes. These structures contain bile and communicate with the biliary tree as well as with each other[7]. This is in contrast to polycystic disease of the liver, in which the cysts do not communicate with the biliary tract, and contain mucus rather than bile[8]. Inheritance of CD is autosomal recessive[3]. The basic anomaly may be developmental defect in the supportive structure of the ductal wall[7]. It is suggested that condition is a link between congenital cystic dilatation of the common bile duct and liver cysts[9]. The dilatation leads to bile stasis, predisposes to bacterial growth, and by compression of parenchymal cells impairs liver function[7]. CD is often complicated by gallstone formation within the saccular dilatations, recurrent bouts of cholangitis and abscess formation[10]. Although the disease may manifest at any age, patients usually present as children or as young adults. In older age, the patient may present with complications of portal hypertension like oesophageal varices, or hepatic carcinoma. During acute episodes the patient may present with fever, pruritus, mild jaundice, slightly enlarged tedner liver, and moderately elevated serum bilirubin, transaminases and alkaline phosphatase[1]. In our patient, the condition was brought to attention when a moderately enlarged liver was found on physical examination during hospital admission for diarrhoea.

In the past, preoperative diagnosis was considerably difficult, and often the diagnosis was revealed by operative cholangiography. However, in recent years, with the development of new imaging techniques, a correct

preoperative diagnosis can be made in almost all instances. Ultrasound scan (US) is a precise screening tool in experienced hands. The typical pattern is characterised by the presence of sonolucent spaces within liver converging towards the porta hepatis[7,11]. US can differentiate CD from cysts in polycystic disease which appear spherical or egg-shaped, and do not communicate with one another or with the biliary tree[7]. Another advantage of US is that once the diagnosis is established, the patient may be followed with ultrasonographic examinations for early detection of stone formation and carcinoma[7]. CT and MRI scans may also be used. Though these procedures give better resolution, they usually do not give any more information than can be obtained from sonography. Hepatobiliary scan shows cold areas in the early phase which become hot at a later phase of imaging[12] as was observed in our patient. The diagnosis can further be confirmed by percutaneous transhepatic cholangiography (PTC) which is an invasive procedure but it may be extremely useful in patients with CD when the anatomy cannot be clearly outlined by other means[11]. The definitive diagnosis was ultimately confirmed by PTC in our patient.

Management of CD is extremely challenging. Long-term antibiotic therapy is usually required to combat cholangitis. The infecting organisms may be cultured from bile obtained by percutaneous aspiration to direct antimicrobial therapy[13]. Excision of any associated choledochal cyst, is the first step in surgical treatment[14]. Surgery is also indicated for obstructing stones, abscess, or portal hypertension. However, drainage procedures like hepatico-enterostomies are disappointing with regards to the problems of recurrent cholangitis, and in general, the long-term prognosis is poor after surgical drainage[15]. Conservative procedures like removal of accessible affected canaliculi, aspriations, sclerotherapy, marsupialisation, and fenestrations have all been tried for intrahepatic dystic malformations, but the outcome has been uniformly disappointing[15]. Recently, Ros et al reported successful treatment of hepatolithiasis in CD by litholytic therapy employing ursodeoxycholic acid[16].

Hepatic resection is the method of choice when feasible. It is ideally suited for single-lobe CD if the contralateral lobe is normal, and when resection will not injure the bile duct of the remaining liver[6].Apart from curative intentions liver resection also represents a prophylactic method since the risk of malignant transformation is considerably high[17]. The generalised from of the disease presents a dilemma in terms of treatment. Orthotopic liver transplantation may represent the only effective and valid therapeutic option. Prognosis of residual or unresectable CD is guarded, and fatalities due to liver abscess, sepsis, cholangiocarcinomas, and amyloidosis have been reproted[16].

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