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Cholecysto-appendicostomy as partial internal biliary drainage in Progressive Familial Intrahepatic Cholestasis Type 1: A case report and review of literature

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ABSTRACT

Intractable pruritus secondary to bile salts retention in Progressive Familial Intrahepatic Cholestasis (PFIC) can be relieved surgically by diverting bile drainage from ileum to reduce bile salts reabsorption into entero-hepatic circulation. We are reporting on the successful biliary diversion in a child with PFIC, with the use of the appendix as a conduit to drain bile from gallbladder to the colon (cholecysto-appendicostomy).

Progressive Familial Intrahepatic Cholestasis (PFIC) is an autosomal recessive disorder, typically presented during infancy with jaundice, pruritus and failure to thrive [1]. It is characterized by impaired bile acid metabolism and excretion causing intrahepatic cholestasis [1,2]. The concentrated biliary compounds lead to disabling pruritus and hepatocellular damage, subsequently progresses to liver failure and death in early childhood or adolescence [2,3]. Mutation in the *ATP8B1* gene, a regulator of intestinal bile acid absorption, has been identified in PFIC Type 1, also known as Byler disease [4]. Before 1990, liver transplant (LTX) was the only treatment option for PFIC [2]. However, in the past 2 decades, surgical intervention with biliary diversion (BD) had been considered for symptomatic relief and was found to curtail liver impairment, and delaying the need for LTX [2,3]. In this report, we share our experience of cholecysto-appendicostomy (CA, a partial internal biliary drainage, PIBD) in a child with PFIC and reviewed the literature of BD surgery.

1. Case report

A 7-year-old girl presented with jaundice and failure to thrive since the age of 10 months old. She was born at term with a birth weight of 2.25 kg. She had neonatal jaundice but did not require admission or phototherapy. Her parents were healthy and unrelated, and there was no family history of liver disease. She was admitted at the age of 10 months old for acute gastroenteritis but found to be jaundiced and her weight was 4.6 kg (below 3rd centile). Liver function test (LFT) showed raised levels of Aspartate Aminotransferase (AST) and Alkaline Phosphatase (ALP) but with normal level of Gamma-Glutamyl Transpeptidase (GGT, 5 U/L). She had conjugated hyperbilirubinemia (82 umol/L). Hepatobiliary ultrasound showed no evidence of biliary obstruction or focal liver disease. Additionally, screenings of her thyroid function, immunology and inborn error of metabolism were all normal. Percutaneous liver biopsy was reported with paucity of intrahepatic bile ducts. Subsequent assessments for Alagille syndrome included

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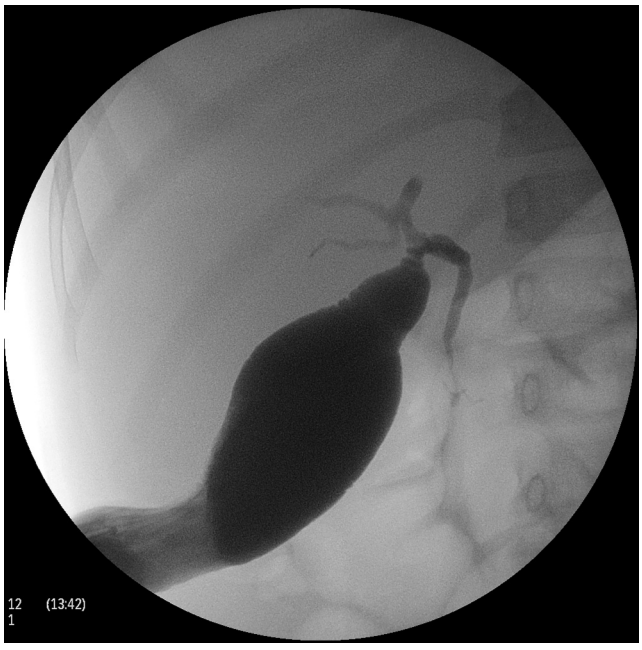

Fig. 1. Operative cholangiogram showed a distended, elongated gallbladder and patent biliary tree.

spine x-rays, echocardiogram and ophthalmology examination were normal. Importantly, genetic study found mutation in *ATP8B1* gene, diagnostic for PFIC Type 1.

She is under close follow-ups by the paediatrics gastroenterology and hepatology team, together with the dietician. She has persistent hyperbilirubinaemia with deranged liver enzymes (raised ALT and ALP). During her disease course, she developed severe skin pruritus, disturbing her social activity and sleep, and her skin became lichenified due to persistent scratching. Despite treatment with antihistamines, ursodeoxycholic acid and cholestyramine, her pruritus persisted and surgical referral was made for consideration of BD at the age of 7 years old.

Laparotomy was performed via a right upper transverse muscle-cutting incision. Operative cholangiogram showed patent biliary tree with grossly distended and elongated gallbladder (Fig. 1). The appendix was identified, mobilized and brought up close to the gallbladder via a small mesenteric defect created medial to caecum/ascending colon to facilitate a tension-free, non-kinking anastomosis (Fig. 2). Liver biopsy was obtained. The surgery was uneventful and her postoperative recovery was excellent.

At recent follow-ups, she reported markedly reduced pruritus, with improved quality of life. Moreover, her parents observed less intense scratching at home and she had better quality of sleep. Clinically, she was less jaundice and her skin was less lichenified. Although her liver enzymes recorded minimal improvement but her bilirubin level has markedly reduced (Fig. 3). Her liver biopsy only showed evidence of chronic hepatitis with fibrosis. She remained well 8 months after surgery with no symptoms to suggest progression of liver impairment.

2. Discussion

Intractable pruritus is one of the most disturbing and disabling symptom of PFIC [2,5], with persistent scratching leading to social and sleep disturbances, and skin lichenification. Medical therapy with cholestyramine, phenobarbital, rifampin or ursodeoxycholic acid may relieve symptoms in 60% of patients [4,6]. LTX was the only surgical intervention until the late 1980s when surgery for BD was introduced and considered. BD involves diversion of biliary drainage, bypassing the ileum to reduce bile salts reabsorption into entero-hepatic circulation and thus increases the elimination of bile acids from the body [2].

Various techniques of BD have been reported (Table 1). In 1988, Whitington and Whitington introduced partial external biliary drainage (PEBD): jejunum was used as a conduit to drain bile from gallbladder to an external stoma (cholecysto-jejuno-cutaneostomy), with very encouraging short-term outcomes [7]. Successful PEBDs were further reported but issues related to stoma or external biliary fistula, such as stoma prolapse and high output stoma losses were documented, respectively [5,9]. In 2005, Metzelder et al.

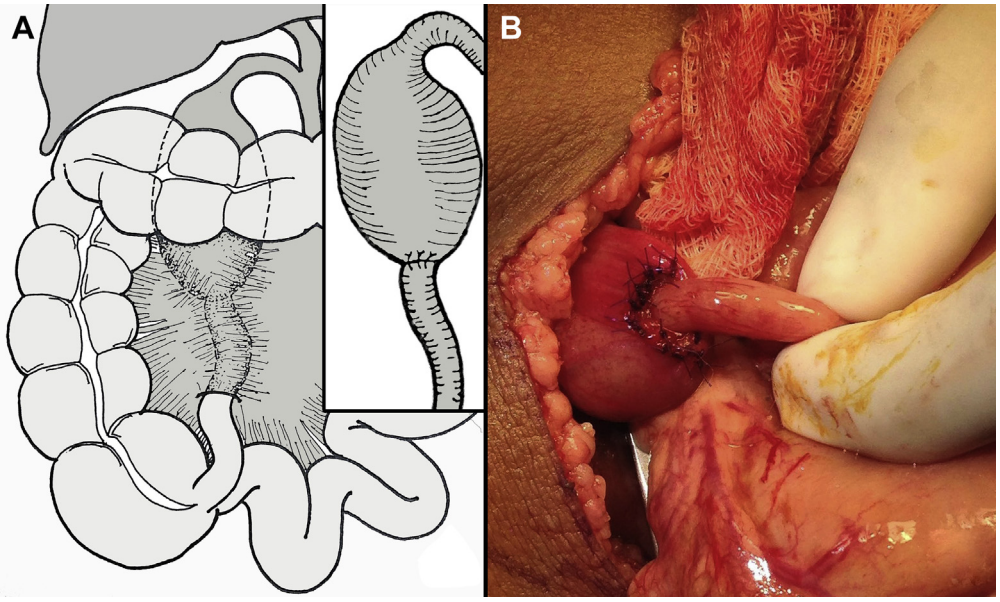

Fig. 2. A – Illustrative diagram of cholecysto-appendicostomy. B – Intra-operative image showed end-to-end anastomosis with interrupted sutures (cholecysto-appendicostomy).