Choledochal Cysts: Presentation, Clinical Differentiation, and Management

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Choledochal cysts (CC) are a rare congenital cystic dilation of the biliary tract, first described by Vater and Ezler in 1723.[1] They present primarily in female infants and young children and are more prevalent in East Asian populations. Although benign, CC can be associated with serious complications including malignant transformation, cholangitis, pancreatitis, and cholelithiasis.[2] We herein provide a state-of-the-art, evidence-based review of CC with particular emphasis on clinical differentiation and approach to management. A search of the available electronic databases, including MEDLINE/Pubmed, using the term *choledochal cyst* as well as under the MeSH database subheading *choledochal cyst,* was performed. Criteria for inclusion included English articles (Fig. 1).

Incidence and epidemiology

Approximately 80% of CC are diagnosed in infants and young children within the first decade of life.[3,4] The incidence of CC ranges from 1 in 100,000 to 1 in 150,000 individuals in Western countries[5] to 1 in 13,000 individuals in Japan.[6] Choledochal cysts are 4 times more common in females.[2,7,8] Although the exact etiology is unknown, anomalous pancreaticobiliary duct union (APBDU) is seen in 30% to 70% of all CC where the common bile duct (CBD) and pancreatic duct junction occurs outside the duodenum, allowing reflux of pancreatic fluid into the biliary tree.[9-13] The exposure of biliary epithelium to digestive and caustic pancreatic enzymes may contribute to CC formation. In 1969, Babbitt[14] initially described APBDU, and it is believed to be secondary to arrest in migration of the choledochopancreatic junction into the duodenal wall, leading to a long common channel (Fig. 2).[15] A long common channel is defined as insertion of the CBD farther than 15 mm from the ampulla of Vater.[16] It occurs in less than 2% of the population,[16] although it is more commonly seen in pediatric CC patients. Eighty percent to 96% of pediatric CC are associated with APBDU.[2,13,17] In one series of 2,885 patients undergoing ERCP, nearly 90% of patients diagnosed with an APBDU had a CC.[16] Animal studies have given credence to this theory since iatrogenic APBDU in murine models demonstrated cystic dilatation of the CBD.[18,19] Amylase levels in the fluid contained in the gallbladder and CC are typically elevated in patients with APBDU.[13] Other pathophysiologic mechanistic hypotheses for CC include a weak bile duct wall, sustained increased intrabiliary pressure, inadequate autonomic innervations, sphincter of Oddi dysfunction, and distal obstruction of the CBD.[5,20,21]

Classification

Alonso-Lej and colleagues[20] proposed the first CC classification in 1959. Komi and associates[11] later proposed a new CC classification according to the type of APBDU based on 2 unique features: a long common channel and the angle of the junction between the pancreatic duct and distal CBD as they converge on the sphincter of Oddi.[5] However, the most widely accepted classification was reported by Todani and colleagues[22] in 1977, derived from the original Alonso-Lej classification and based on the site of cystic change (Fig. 3). Five types of CC are described and classified: type I (80% to 90% of all CC), type II, type III, type IV (15% to 20% of all CC) and type V or Caroli's disease.[2,7,22-24]

Type I cysts typically appear as anechoic cystic lesions, which communicate with the biliary tract. A type I cyst can be associated with mild enlargement of the intrahepatic bile ducts secondary to biliary stasis (Fig. 4).[7] Further differentiation of type I cysts (1A, 1B, or 1C) is accomplished using ultrasound and cholangiography to evaluate the gallbladder relationship and cystic duct location. In type IA CC, the gallbladder arises from the choledochal cyst and a dilated extrahepatic biliary tree

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Abbreviations and Acronyms

APBDU = anomalous pancreaticobiliary duct union
BA = biliary atresia
CBA = cystic biliary atresia
CBD = common bile duct
CC = choledochal cyst
CHD = common hepatic duct
MRCP = magnetic resonance cholangiopancreatography
OLT = orthotopic liver transplant
RYHJ = Roux en Y hepaticojejunostomy

is seen while the intrahepatic ducts are normal in size and appearance.[7] Type IB CCs contain a mostly normal appearing extrahepatic biliary tree with an isolated dilatation of the most distal aspect of the CBD, with no evidence of pancreaticobiliary malunion.[5] A smooth fusiform dilatation of the common hepatic duct (CHD) and CBD along with pancreaticobiliary malunion is classified as type 1C CC.[25]

Type II cysts are true diverticula of the CBD and represent 2% of reported cases.[26] Type II cysts appear as anechoic cysts juxtaposed to the CBD with a normal appearing gallbladder and CHD (Fig. 5). Cholangiography demonstrates opacification of a true diverticulum arising from the CBD[7] and can resemble gallbladder duplication.[5,27]

Type III cysts, or choledochoceles, were initially described by Wheeler[28] in 1940. Type III cysts comprise 1% to 4% of CC and are characterized by their intraduodenal location at the pancreaticobiliary junction.[5,7,26,29] Although CC have a female predominance, choledochoceles are more evenly distributed between the sexes.[17,30] Type III cysts are also more likely to be diagnosed using ERCP and are managed primarily with endoscopic therapy.[17,31] Pancreatitis is commonly seen and biliary tract symptoms are less common.[17,32,33] Type III cysts are associated with a much lower incidence of malignant transformation (2.5%).[29,34,35] Additionally, APBDU is less commonly seen in choledochoceles in comparison with other types of CC, and patients are more likely to have undergone a previous cholecystectomy at the time of diagnosis.[10,17,36] In fact, given the distinct differences in presentation, clinical course, diagnosis, and pathophysiology, some authors argue that choledochoceles represent a different disease entity.[17,30,33]

Type IV CC can include both intrahepatic and extrahepatic duct involvement. Type IV CC are subclassified into type IVA and type IVB. Type IVA CC dilatation extends from the CBD and CHD into the intrahepatic biliary tree (Fig. 6). Additionally, primary ductal stricture around the hepatic hilum is commonly seen.[5,25] Although intrahepatic biliary dilatation most commonly presents with bilobar involvement, dilatation of the left lobe is the second most common presentation.[37,38] Isolated dilatation of the right lobe is rarely seen.[38] By contrast, type IVB CC consists of multiple dilations of the extrahepatic biliary tree, classically described as a "string of beads," with an uninvolved intrahepatic biliary tree.[5]

Finally, type V CC, or Caroli's disease, demonstrates intrahepatic saccular or fusiform dilatation with no underlying obstruction or extrahepatic biliary tree involvement (Fig. 7A, B).[7] Type V CCs are thought to arise from ductal plate malformation[15] and be associated with polycystic kidney disease,[39] an autosomal recessive inherited condition associated with mutation in PKD1 gene (Fig. 7C).[40] When type V CCs are accompanied with congenital hepatic fibrosis, it is termed Caroli's syndrome.[15] The enhancement of the portal vein surrounded by dilated intrahepatic bile ducts, or "central dot sign," is highly suggestive of Caroli's disease and can easily be seen on magnetic resonance cholangiopancreatography (MRCP) or contrast-enhanced CT.[7,41,15] Contrast filling in well-defined intrahepatic cystic dilatations is pathognomic.[15]

Visser and colleagues[27] have challenged the modified Todani classification, stating that it combines multiple and different disease entities. In support of this, the investigators note the different clinical courses, management, and complication rates of the 5 types of CC. Specifically, Visser and colleagues[27] note the distinction of types I and IVA CC as arbitrary given that there is generally some intrahepatic duct involvement in both classes of CC. The authors further state that gallbladder-like diverticula, choledochoceles, and Caroli's disease are completely unrelated to CC and therefore propose abandoning the Todani classification and instead using descriptive terminology.

Clinical presentation

Choledochal cysts are usually diagnosed in childhood, although in utero and adult diagnosis is also common.[7,42] Common presentations include abdominal pain, jaundice, and right upper quadrant mass and are most commonly seen in pediatric patients.[24,43] Cholangitis, pancreatitis, portal hypertension, and liver function test abnormalities are common and are thought to be a result of ABPDU or stone obstruction.[23,24,43-47] Biliary amylase levels can be elevated in CC patients, and clinical features correlate with degree of elevation.[48-51] The classic triad of abdominal pain, right upper quadrant mass, and obstructive jaundice is mainly seen in the pediatric population, although still rare.[26,50,52]