

Choledochal Cyst

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Abstract

Choledochal cysts (CDCs) are a rare condition characterized by abnormal dilation of the biliary tree. They are most diagnosed during infancy or childhood. There does not seem to be a single cause that explains the various types of CDCs. Children may present with the classic triad of abdominal pain, jaundice, and an abdominal mass. While abdominal US is the preferred initial diagnostic tool, magnetic resonance cholangiopancreatography and endoscopic retrograde cholangiopancreatography provide more accurate results. Common complications include stone formation and malignant transformation. CDCs generally have a good prognosis after surgical resection. However, patients require long-term surveillance.

Keywords: hepatobiliary, congenital, neonatal

Case Summary

A newborn was referred for pediatric hepatology consultation to follow up for prenatal US showing an abdominal cyst. The patient had an uneventful delivery.

Imaging Findings

Abdominal US (Figure 1) showed a cystic configuration of the common bile duct (CBD). Subsequent magnetic resonance cholangiopancreatography (MRCP) (Figure 2) showed diffuse enlargement of the CBD. Intrahepatic bile ducts were not dilated.

Diagnosis

Type 1 choledochal cyst (CDC).

The differential diagnosis of a congenital cystic mass in the right upper quadrant includes a foregut duplication cyst and a duplicated gallbladder.

Discussion

A CDC is a rare congenital condition characterized by the abnormal dilation of the biliary tree.¹ The Todani classification system divides CDCs into 5 classes based on anatomical location and morphology.² Type 1 cysts are the most common, representing 80-90% of cases.^{1,2} This subtype is characterized by fusiform or cystic dilation of the CBD. Other types of cysts are less common. Their appearance is summarized in Table 1.¹⁻³ Type 1 and 4 cysts are most likely to be associated with later development of cholangiocarcinoma that increases in incidence with increasing age.² The cumulative incidence is 1.6%, 3.9%, and 11.3% at 15, 20, and 25 years if the cyst is not resected.³ In contrast, it is only 0.7-5.4% after cyst resection.⁴

CDCs are considered rare, with an estimated occurrence of approximately 1 in 13,000-15,000 individuals in Western populations. However, in Asia, the incidence is higher, affecting 1 in 1000 infants. Notably, two-thirds of these cases

are reported in Japan.⁵ Females are more commonly affected, accounting for 70-84% of all cases (except for type 4 cysts, where the incidence is similar between the sexes).^{1,3,5} CDCs are most diagnosed in children <10 years old. However, the diagnosis is occasionally made in adults. Presenting symptoms depend on the cyst subtype and severity. The classic clinical triad of a CDC in children includes abdominal pain, jaundice, and an abdominal mass. More than 80% of children will present with 2 or more of these classic symptoms, while only 23% of adults will report any symptoms. Infants most commonly present with hepatomegaly, abdominal mass, and jaundice but rarely present with symptoms that could be attributed to abdominal pain.^{1,6} Other symptoms in children include intermittent fever and vomiting, which may be related to pancreatitis or cholangitis.⁷ These symptoms may coincide with the release and reflux of bile and pancreatic enzymes.

Multiple theories have attempted to explain the pathogenesis of CDCs. Potential causes include anatomical, environmental, and genetic factors.^{1-3,5,8} The most accepted theory postulates that CDCs are caused by an anomalous pancreaticobiliary union.^{2,8} The normal pancreaticobiliary duct forms when the CBD and pancreatic duct join

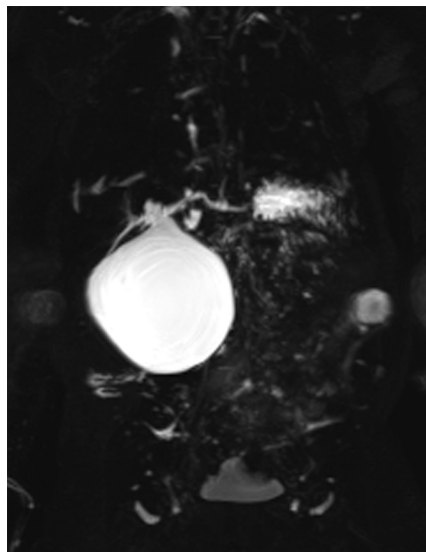
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Figure 1. Transverse grayscale US image showing cystic dilation of the common bile duct. A small portion of the proximal bile duct (arrowhead) is seen in the cyst.



Figure 2. Coronal T2-weighted maximum intensity projection from magnetic resonance cholangiopancreatography showing cystic dilation (arrow) of the common bile duct consistent with a type 1 choledochal cyst.



at the sphincter of Oddi.^{1,8} An anomalous pancreaticobiliary union occurs when the CBD and pancreatic duct join more outside of the duodenal wall (typically > 1 cm proximal to the sphincter of Oddi). This results in an elongated common channel and an abnormal angle that differs from the anatomically normal acute angle typically seen. These structural defects allow for bile and activate pancreatic

Table 1. Choledochal Cyst Classification

Choledochal Cyst Type	Description
1	Cystic or fusiform dilation of the extrahepatic bile duct
1A	Cystic dilation of the entire extrahepatic biliary tree
1B	Focal, segmental dilation of the extrahepatic biliary tree
1C	Fusiform, diffuse dilation of the entire extrahepatic bile duct
2	A true diverticulum, or outpouching, from the extrahepatic bile duct
3	A choledochocele, which is a dilation of the distal common bile duct within the duodenal wall
4	Multiple cysts
4A	Cysts involve both the intrahepatic and extrahepatic biliary ducts
4B	Multiple cysts are confined to the extrahepatic bile ducts
5	Caroli disease, characterized by multiple cystic dilatations within the intrahepatic bile ducts

enzymes to reflux into the biliary tree. The reflux of pancreatic secretion is thought to lead to inflammation and ultimately dilation of the CBD.^{1,5,8} While an anomalous pancreatobiliary junction can explain type 1 and 4 cysts, it does not explain other types. Type 2 cysts are thought to be caused by a congenital weakness or acquired injury in the wall of the CBD, while type 3 cysts represent a congenital or acquired structural defect near the ampulla of Vater.^{1,3,8} Type 5 CDC (Caroli disease) is thought to be an abnormality of remodeling of the ductal plates as is associated with biliary atresia.

Diagnosis of CDC is typically achieved via imaging.² US is the preferred initial imaging modality in children. It has a diagnostic sensitivity between 71% and 91%.^{1,2,5,6} Sonography can identify the level of bile duct dilation.¹ At times, it is difficult to distinguish a type 1 cyst from other causes of CBD dilation, such as a choledocholithiasis. In these instances, correlation with clinical symptoms and repeat imaging after the acute symptoms resolve may be useful.

CT is occasionally used in the patients with acute abdominal pain. It can also identify dilated bile ducts. However, CT is typically not able to demonstrate the pancreatobiliary junction, especially if the ducts are not dilated.^{2,5} The technetium-99 Hepatobiliary Iminodiacetic Acid scan has 100% sensitivity in diagnosing type 1 CDCs. It is also useful in differentiating cystic biliary atresia and CDCs in neonates.^{5,9}

Distinguishing features of HIDA scans in neonates with biliary atresia usually demonstrate no activity in the duodenum on delayed imaging, while those with CDCs do show gallbladder activity. US is also a valuable tool. In addition, neonates with CDCs are more likely to show intrahepatic biliary dilation, a normal-sized gallbladder (> 1.5 cm) or distended gallbladder, and a larger cyst size in patients with CDCs. Cystic biliary atresia is more likely if there is a triangular cord sign and absence of gallstones and sludge within the cyst at the hepatic hilum.

MRCP has become the preferred method of evaluation of the pancreatobiliary junction. It provides excellent evaluation of intra- and extrahepatic bile ducts, as well as the pancreatic duct. Because of this, MRCP has a diagnostic accuracy of 82-100% of pediatric CDCs, with a sensitivity of 96-100% and specificity of 90%.^{5,7} At times, endoscopic retrograde cholangiopancreatography (ERCP) is needed. Notably, ERCP is currently the gold standard for the identification of anomalous pancreatobiliary junction.^{1,5,6}

The most common complication of CDCs is stone formation, followed by malignant transformation to cholangiocarcinoma.^{1,4} The risk for cholangiocarcinoma is related to bile stasis and pancreatic enzyme reflux into the biliary tree, leading to chronic irritation and metaplasia. Malignancy is most common in patients with type 1 and 4 CDCs, with an associated prevalence of 7.5%.^{1,5,10}

Neonates are also prone to cystic rupture, which may result in bile peritonitis. Pancreatitis is a complication of CDCs commonly caused by an obstructive stone or bile reflux into the pancreatic duct.¹ Children with CDCs may get pancreatitis in 35-53% of cases. The incidence is high if pancreaticobiliary maljunction is present.

The management of CDCs can vary depending on size, location, and type. Type 1 and 4 CDCs are treated with surgical excision and Roux-en-Y hepaticojejunostomy.^{1,5,6} Laparoscopic resection in pediatric populations has resulted in decreased postoperative complications and shorter hospitalizations. Incomplete resection can increase risk for ascending cholangitis, cystolithiasis, and malignant transformation. Type 2 and 3 cysts have a low risk for malignancy. A type 2 cyst is managed by diverticulectomy, while small choledochoceles are treated with endoscopic sphincterotomy. Type 5 cysts are managed with liver resection (if the cysts are focal) or liver transplant (if the cysts are diffuse).^{1,5,11}

If resected in childhood, patients with CDCs have an excellent prognosis.^{1,5,6,11} Pediatric resection is associated with an 89% event-free rate and a 5-year survival rate of >90%.¹¹ The risk for biliary malignancy remains high, >15 years after excision, and, thus, long-term surveillance is highly recommended.

Conclusion

CDCs are a rare condition characterized by abnormal dilation of the biliary tree. It is most diagnosed during infancy or childhood. There does not seem to be a single cause that explains the various types of CDC. Children may present with the classic triad of abdominal pain, jaundice, and an abdominal mass. While abdominal US is the preferred initial diagnostic tool, MRCP and ERCP provide more accurate results. Common complications include stone formation and malignant transformation. CDCs generally have a good prognosis after surgical resection. However, patients require long-term surveillance.

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