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ADVANCES IN PEDIATRICS

Biliary Atresia

Epidemiology, Genetics, Clinical Update, and Public Health Perspective

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Keywords

• Biliary atresia • Obstructive jaundice • Portoenterostomy • Liver cirrhosis

Key points

- Biliary atresia is a rare birth defect, with prevalence of 0.5 to 0.8 per 10,000 births in developed nations, and remains a surgical disease with good outcomes with the Kasai portoenterostomy, although age at operation plays a key determination in outcome.
- Although some genetic factors and associations have been identified, biliary atresia appears to have multifactorial etiologies, which remain unclear.
- Timely work-up of obstructive jaundice is of key importance to performing portoenterostomy before 90 days of life, should biliary atresia be the diagnosis.
- For those who do not have successful drainage after operation, liver transplant remains the only option.
- Universal screening, using stool color cards or other methods, may improve age at diagnosis and treatment, leading to improved outcomes for infants with biliary atresia.

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Biliary atresia is a rare destructive, inflammatory condition in which progressive fibrosis of the biliary tree in an infant leads to bile duct obstruction and consequent liver cirrhosis [1]. If left untreated, progressive liver cirrhosis leads to death by age 2 [2]. Biliary atresia can be classified into 3 categories, all of which are dependent on the level most proximal to the biliary obstruction (Fig. 1). Type I involves obstruction of the common bile duct, characterized with luminal patency down to the common bile duct, and accounts for approximately 5% of cases. Type II, characterized with patency to the level of the common hepatic duct, accounts for approximately 2% of cases. In both types, there is some preservation of the intrahepatic ducts, although they may morphologically still be abnormal and irregular. Given this abnormality in structure, they typically do not dilate, despite the presence of an obstruction. Finally, type III involves obstruction at the level of the porta hepatis [3]. In most populations, type III accounts for more than 90% of cases and is characterized by most of the proximal part of the extrahepatic biliary tract within the porta hepatis entirely solid [2]. On abdominal exploration, a dense inflammatory proximal remnant is noted at the porta hepatis. Distal ducts may be well preserved, atrophic, or even absent. In this type, the intrahepatic ducts are grossly abnormal. In addition, it is important to distinguish this type of biliary atresia from a choledochal cyst, because there may be an associated extrahepatic cyst formation in type III biliary atresia. The key to distinguishing

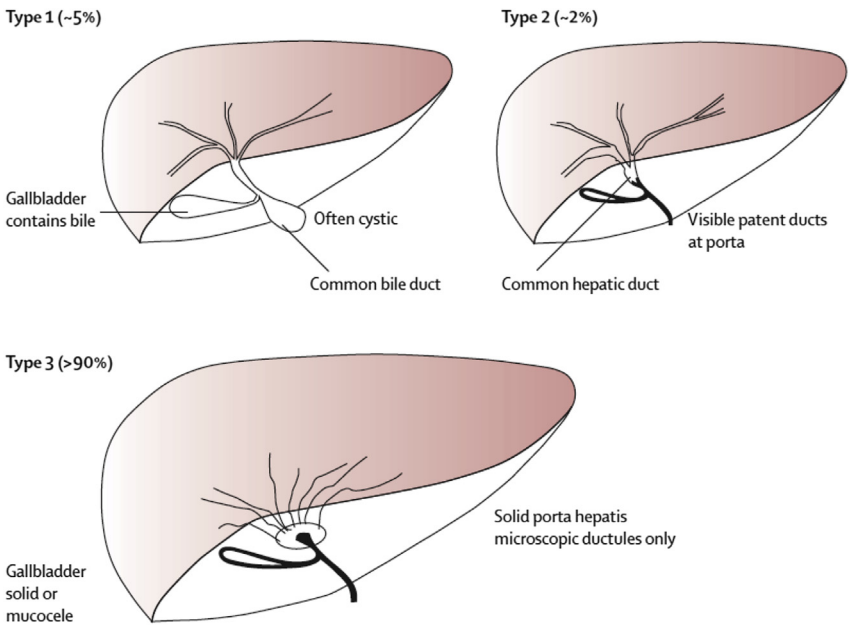

Fig. 1. Schematic illustration of classification of biliary atresia. (*From* Hartley LJ, Davenport M, Kelly DA. Biliary atresia. Lancet 2009;374(9702):1705.)