

Biliary atresia is a rare gastrointestinal disorder characterized by destruction or absence of all or a portion of the bile duct that lies outside the liver (extrahepatic bile duct). The bile duct is a tube that allows the passage of bile from the liver into the gall bladder and, eventually, the small intestine. Bile is a liquid secreted by the liver that plays an essential role in carrying waste products from the liver and promoting absorption of fats and vitamins by the intestines. In biliary atresia, absence or destruction of the bile ducts results in the abnormal accumulation of bile in the liver. Affected infants have yellowing of the skin and whites of the eyes (jaundice) and scarring of the liver (fibrosis). In some cases, additional abnormalities may be present, including heart defects and intestinal, spleen and kidney malformations. The exact cause of biliary atresia is unknown. Biliary atresia is a rare disorder with a slight increased frequency in females. It occurs in approximately 1 in 10,000 to 15,000 births in the United States.

Approximately 400-600 new cases of biliary atresia are encountered in the United States each year. According to one estimate, the prevalence of biliary atresia in Europe is approximately 1 in 12,000 births. Biliary atresia is the most common cause of end-stage liver disease and liver transplantation in children. The diagnosis of biliary atresia requires a direct examination of the bile ducts by abdominal surgery (laparotomy) and the microscopic examination of tissue from the liver (liver biopsy). During the surgery special contrast dye is injected into the gallbladder and x-ray films are taken to outline how the dye fills the major bile ducts (intraoperative cholangiogram). These films show the movement (or lack of movement) of the dye through bile ducts and into the small intestine. The physician/surgeon is then able to evaluate the structure of the bile ducts and to determine the site of the blockage (proximal or distal). Blood tests may demonstrate elevated levels of liver enzymes, gamma-glutamyl transpeptidase, and bilirubin and detect viral agents; high blood levels of matrix metalloproteinase-7 has been discovered to be highly specific for biliary atresia. Ultrasound of the liver may show absence of the gall bladder.