

What is the most common form of familial intrahepatic cholestasis?

- a. Alagille syndrome
- b. Dubin-Johnson syndrome
- c. Benign recurrent cholestasis
- d. Stauffer's syndrome

ANSWER: A

Alagille syndrome or arteriohepatic dysplasia is the most common form of familial intrahepatic cholestasis, which is characterized by cholestasis, paucity of interlobular bile ducts and other congenital malformations. Alagille syndrome is an autosomal dominant disorder, with mutations in JAGGED1 (JAG1) gene found in 70% of these patients. The metabolic defect in both Rotor's syndrome and Dubin-Johnson syndrome is related to impaired canalicular export of conjugated bilirubin. Benign recurrent cholestasis is an autosomal recessive disorder associated with mutations in transport proteins, and leads to minimal histologic abnormalities despite recurrent attacks of fatigue, pruritis and jaundice. Stauffer's syndrome relates to jaundice resulting from intrahepatic cholestasis as a paraneoplastic phenomenon, and would therefore not be familial.