Deficiency	Mutated gene	Typical clinical characteristics	Characteristic histology at diagnosis	Typical clinical outcomes
FIC1	*ATP8B1*	Multisystem disease Normal γGT Only modest elevation of transaminases	Bland canalicular cholestasis Coarsely granular canalicular bile	Moderate rate of progression Post-transplant hepatic steatosis and diarrhea
BSEP	*ABCB11*	Normal γGT High risk of HCC High incidence of gallstones	Giant cell transformation	Moderate to rapid progression Allo-antibody formation after transplant in some
MDR3	*ABCB4*	Progressive cholangiopathy Elevated γGT	Cholangiolytic changes	Highly variable rate of progression
TJP2	*TJP2*	Some extra hepatic features Near normal γGT	Bland cholestasis	Rapid progression
FXR	*NR1H4*	Early onset coagulopathy Normal γGT Markedly elevated AFP	Intralobular cholestasis Ductular reaction Giant cell transformation	Very rapid progression Post-transplant hepatic steatosis
MYO5B	*MYO5B*	Normal γGT Variable degree of intestinal involvement	Giant cell change Hepatocellular and canalicular cholestasis	Slow progression

Table 1. Summary of the typical features of progressive familial intrahepatic cholestasis associated with different genetic etiologies.

γ-glutamyltranspeptidase (γGT); hepatocellular carcinoma (HCC); α-fetoprotein (AFP)