DIAGNOSTIC ASSESSMENT OF LATE-ONSET CHOLESTASIS

A PFIC diagnosis typically requires a combination of assessments[1-3]

Medical History and Clinical Examination[1,3]

Key symptoms include cholestatic pruritus and jaundice, often accompanied by gastrointestinal symptoms. Any sign of unexplained cholestatic pruritus warrants further investigation.

Laboratory Tests[1,2]

Serum bile acid, ALP, ALT, AST, GGT, and bilirubin are markers of liver function.

Imaging[2]

Abdominal ultrasound and cholangiopancreatography can rule out bile duct blockages outside the liver and identify liver damage.

Histology[2]

A liver biopsy can assess hepatocellular changes and bile duct abnormalities.

Genetics[1,3]

Genetic testing can support a diagnosis and guide treatment decisions, but a negative result does not rule out PFIC.

ALP=alkaline phosphatase; ALT=alanine aminotransferase; AST=aspartate aminotransferase; GGT=gamma-glutamyl transferase; PFIC=progressive familial intrahepatic cholestasis.

References: 1. Thompson RJ, Vilarinho S, Miquel R, Keitel V. Challenges in the diagnosis and treatment of genetic cholestasis in adults. *JHEP Reports*. 2025;8:101625. **2.** Srivastava A. Progressive familial intrahepatic cholestasis. *J Clin Exp Hepatol*. 2014;4(1):25-36. **3.** Thompson RJ, Vilarinho S, Miquel R, Keitel V. Challenges in the diagnosis and treatment of genetic cholestasis in adults. *JHEP Reports*. 2026;8:101625. **4.** Labcorp. Diagnostics—pediatric testing reference ranges. Laboratory Corporation of America® Holdings; 2022. **5.** Mayo Clinic Laboratories. Gamma-glutamyltransferase (GGT), serum. Accessed April 22, 2026. https://pediatric.testcatalog.org/show/GGT