

CLINICAL MANIFESTATIONS OF PFIC IN ADULTS

◀ PFIC should be suspected in adults with ▶



Cholestatic pruritus¹



Serum bile acid levels
>ULN^{1,2}



Gallstone disease
(especially when present at
a young age)^{1,3}



Jaundice^{1,2}



Elevated liver enzymes
and/or GGT^{1,2}



Episodic cholestasis^{1,4-7}



Cholestasis arising after infection, hormonal changes in pregnancy, or use with a hepatically metabolized medication^{1,4-7}

◀ Symptoms overlap with other conditions, such as⁷ ▶

- Intrahepatic cholestasis of pregnancy
- Cholesterol gallstone disease
- Drug-induced cholestasis or liver injury
- Adult idiopathic/cryptogenic cirrhosis

GGT=gamma-glutamyl transferase; PFIC=progressive familial intrahepatic cholestasis; ULN=upper limit of normal.

References: 1. Progressive familial intrahepatic cholestasis in adulthood: genetics, diagnosis, treatment, and further research. *EMJ Hepatol.* 2024;12(suppl 2):2-7. doi:10.33590/emjhepatol/HFCN9816 2. Dröge C, Götze T, Behrendt A, Gohlke H, Keitel V. Diagnostic workup of suspected hereditary cholestasis in adults: a case report. *Explor Dig Dis.* 2023;2:34-43. doi:10.37349/edd.2023.00016 3. Vitale G, Sciveres M, Mandato C, d'Adamo AP, Di Giorgio AD. Genotypes and different clinical variants between children and adults in progressive familial intrahepatic cholestasis: a state-of-the-art review. *Orphanet J Rare Dis.* 2025;20(1):80. doi:10.1186/s13023-025-03599-2 4. Hilscher MB, Kamath PS, Eaton JE. Cholestatic liver diseases: a primer for generalists and subspecialists. *Mayo Clin Proc.* 2020;95(10):2263-2279. doi:10.1016/j.mayocp.2020.01.015 5. Nayagam JS, Miquel R, Thompson RJ, Joshi D. Genetic cholestasis in children and adults. *J Hepatol.* 2024;80(4):670-672. doi:10.1016/j.jhep.2023.11.028 6. Zu Y, Yang J, Zhang C, Liu D. The pathological mechanisms of estrogen-induced cholestasis: current perspectives. *Front Pharmacol.* 2021;12:761255. doi:10.3389/fphar.2021.761255 7. Stättermayer AF, Halilbasic E, Wrba F, Ferenci P, Trauner M. Variants in *ABCB4* (MDR3) across the spectrum of cholestatic liver diseases in adults. *J Hepatol.* 2020;73(3):651-663. doi:10.1016/j.jhep.2020.04.036