

Disease

Progressive familial intrahepatic cholestasis 5^{1, 2, 3}

Gene

NR1H4

Inheritance	AR
Current Treatments	Odevixibat, ursodeoxycholic acid or liver transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	AST, ALT, bilirubin ¹
Disease Frequency (per 100,000)	1.5 ²
Genes Included in Disease Frequency	ATP8B1, ABCB11, ABCB4, TJP2, NR1H4, SLC51A, USP53, KIF12, ZFYV19, MYO5B
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=Progressive%20Familial%20Intrahepatic%20Cholestasis

References

- ¹ <https://omim.org/entry/617049>
- ² <https://pubmed.ncbi.nlm.nih.gov/38192535/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/38405357/>