

Disease

# Progressive familial intrahepatic cholestasis 5<sup>1, 2, 3</sup>

Gene

## NR1H4

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

References

<sup>1</sup> <https://omim.org/entry/617049>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/38192535/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/38405357/>