

Disease

# Crigler-Najjar syndrome<sup>1, 2, 3</sup>

Gene

## UGT1A1

|                                                 |                                                                                                                                                                                                                                                               |
|-------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                            |
| <b>Current Treatments</b>                       | phototherapy, plasmapheresis, enzyme-inducing agents (phenobarbital), bilirubin-binding agents (calcium phosphate, orlistat), choleretics (ursodeoxycholic acid), heme-oxygenase inhibitors (Tin-protoporphyrin, Zinc-protoporphyrin), liver transplantation  |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                                |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                           |
| <b>Non-Molecular Test</b>                       | serum conjugated and unconjugated bilirubin <sup>1</sup>                                                                                                                                                                                                      |
| <b>Disease Frequency (per 100,000)</b>          | 0.117 <sup>4</sup>                                                                                                                                                                                                                                            |
| <b>Genes Included in Disease Frequency</b>      | UGT1A1                                                                                                                                                                                                                                                        |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Crigler-Najjar+Syndrome&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Crigler-Najjar+Syndrome&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

<sup>1</sup> <https://pubmed.ncbi.nlm.nih.gov/32965842/>

<sup>2</sup> <https://omim.org/entry/218800>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/34527915/>

<sup>4</sup> <https://pubmed.ncbi.nlm.nih.gov/31495946/>