

Disease

# Hyperphenylalaninemia due to dihydropteridine reductase deficiency<sup>1, 2</sup>

Gene

## QDPR

|                                                 |                                                                                                                                                                                                                                                           |
|-------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | Tetrahydrobiopterin, levodopa combined with a decarboxylase inhibitor, 5-hydroxytryptophan, low Phe diet                                                                                                                                                  |
| <b>Evidence for Treatment</b>                   | guideline <sup>3</sup>                                                                                                                                                                                                                                    |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                       |
| <b>Non-Molecular Test</b>                       | CSF neurotransmitter metabolites and pterins, plasma amino acids <sup>2</sup>                                                                                                                                                                             |
| <b>Disease Frequency (per 100,000)</b>          | 0.31 <sup>4</sup>                                                                                                                                                                                                                                         |
| <b>Genes Included in Disease Frequency</b>      | QDPR                                                                                                                                                                                                                                                      |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Hyperphenylalaninemia&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Hyperphenylalaninemia&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/23138986>
- <sup>2</sup> <https://omim.org/entry/261630>
- <sup>3</sup> <https://ojrd.biomedcentral.com/articles/10.1186/s13023-020-01379-8>
- <sup>4</sup> <https://www.ncbi.nlm.nih.gov/pubmed/25758715>