

Disease

Mild non-BH4-deficient hyperphenylalaninemia^{1, 2, 3}

Gene

DNAJC12

Inheritance	AR
Current Treatments	5-hydroxytryptamine, levodopa, and tetrahydrobiopterin
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	CSF neurotransmitter metabolites and pterins, plasma amino acids ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Hyperphenylalaninemia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/617384>
- ² <https://doi.org/10.1002/mgg3.1303>
- ³ <https://pubmed.ncbi.nlm.nih.gov/36833190/>
- ⁴ <https://ojrd.biomedcentral.com/articles/10.1186/s13023-020-01379-8>