

Disease

Hyperphenylalaninemia due to 6-pyruvoyl-tetrahydropterin synthase deficiency^{1, 2}

Gene

PTS

Inheritance	AR
Current Treatments	Tetrahydrobiopterin, levodopa combined with a decarboxylase inhibitor, 5-hydroxytryptophan
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	CSF neurotransmitter metabolites and pterins, plasma amino acids ⁴
Disease Frequency (per 100,000)	0.31 ⁵
Genes Included in Disease Frequency	PTS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=tetrahydrobiopterin&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/30926181>
- ² <https://omim.org/entry/605407>
- ³ <https://ojrd.biomedcentral.com/articles/10.1186/s13023-020-01379-8>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/30926181/>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/25758715>