

Disease

Dopa-responsive dystonia due to sepiapterin reductase deficiency^{1, 2}

Gene

SPR

Inheritance	AR
Current Treatments	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 ²
Disease Frequency (per 100,000)	0.725 ^{4, 5}
Genes Included in Disease Frequency	TH, GCH1, SPR
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Dopa+Responsive+Dystonia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/28189489>
- ² <https://omim.org/entry/612716>
- ³ <https://ojrd.biomedcentral.com/articles/10.1186/s13023-020-01379-8>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/19427147>
- ⁵ <https://www.ncbi.nlm.nih.gov/books/NBK1508/#drd.Summary>