

Disease

Dopa-responsive dystonia due to GTP cyclohydrolase 1 deficiency^{1, 2}

Gene

GCH1

Inheritance	AR and AD
Current Treatments	Dominant form: levodopa combined with a decarboxylase inhibitor. Recessive form: levodopa combined with a decarboxylase inhibitor, 5-hydroxytryptophan, and BH4
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	CSF neurotransmitter metabolites and pterins ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC128
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/books/NBK1508/>
- ² <https://omim.org/entry/128230>
- 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>