

Disease

Phosphoglycerate dehydrogenase deficiency¹

Gene

PHGDH

Inheritance	AR
Current Treatments	serine, glycine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	CSF and plasma serine and glycine levels ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/show/NCT02635269?cond=Phosphoglycerate+dehydrogenase+deficiency&draw

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/29899766>

² <https://pubmed.ncbi.nlm.nih.gov/27161889/>