

Disease

Phosphoserine phosphatase deficiency^{1, 2}

Gene

PSPH

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 ³
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Phosphoserine+phosphatase+deficiency&term=&cntry=&state=&

References

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

² <https://pubmed.ncbi.nlm.nih.gov/26589312/>

³ <https://pubmed.ncbi.nlm.nih.gov/27161889/>