

Disease

Vitamin B6-dependent epilepsy^{1, 2}

Gene

PLPBP

Inheritance	AR
Current Treatments	vitamin B6 (pyridoxine), lysine restrict and suppl arginine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Non-Molecular Test	
Disease Frequency (per 100,000)	2.565 ³
Genes Included in Disease Frequency	ALDH7A1, PLPBP, PNPO
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=plbp&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/30668673>

² <https://pubmed.ncbi.nlm.nih.gov/33425341/>

³ <https://www.ncbi.nlm.nih.gov/pubmed/28779752>