

Disease

Pyridoxamine 5-prime-phosphate oxidase deficiency^{1, 2, 3}

Gene

PNPO

Inheritance	AR
Current Treatments	vitamin B6 (pyridoxine), lysine restrict and suppl arginine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	PNPO enzyme activity in dried blood spots ⁴
Disease Frequency (per 100,000)	2.565 ⁵
Genes Included in Disease Frequency	ALDH7A1, PLPBP, PNPO
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=pnpo&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK581452/>
- ² <https://pubmed.ncbi.nlm.nih.gov/34769443/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32888189/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/28782931/>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/28779752>