

Disease

Pyridoxine-dependent epilepsy^{1, 2}

Gene

ALDH7A1

Inheritance	AR
Current Treatments	vitamin B6 (pyridoxine), lysine restrict and suppl arginine
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	Urine organic acids ⁴
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1002
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=ALDH7A1&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1486/>
- ² <https://pubmed.ncbi.nlm.nih.gov/27212567/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/33200442/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/32339489/>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/28779752>