

Disease

Primary hyperoxaluria type I^{1, 2}

Gene

AGXT

Inheritance	AR
Current Treatments	lumasiran, pyridoxine, drinking large volumes, alkalization of urine, pyrophosphate-containing solutions, liver-kidney transplant
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urinary oxalate ³
Disease Frequency (per 100,000)	1.7 ⁴
Genes Included in Disease Frequency	AGXT, GRHPR, HOGA1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=hyperoxaluria&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1283/>
- ² <https://pubmed.ncbi.nlm.nih.gov/33789010/>
- ³ <https://omim.org/entry/259900>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/25644115>