

Disease

Tyrosinemia, type I¹

Gene

FAH

Inheritance	AR
Current Treatments	nitisinone, Low-tyrosine diet
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	succinylacetone in the blood and urine, plasma amino acids ¹
Disease Frequency (per 100,000)	1 ³
Genes Included in Disease Frequency	FAH
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Tyrosinemia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1515/>
- ² <https://pubmed.ncbi.nlm.nih.gov/28771246/>
- ³ <https://ghr.nlm.nih.gov/condition/tyrosinemia#statistics>