

Disease

Citrullinemia, type II^{1, 2}

Gene

SLC25A13

Inheritance	AR
Current Treatments	medium chain triglycerides (MCT) milk and lactose-free milk, lipid-soluble vitamins and ursodeoxycholic acid. Liver transplantation
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC118
Disease Frequency (per 100,000)	3.155 ^{1, 3}
Genes Included in Disease Frequency	SLC25A13
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Citrullinemia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1181/>
- ² <https://doi.org/10.1016/j.ymgme.2019.06.004>
- ³ <https://ghr.nlm.nih.gov/condition/citrullinemia#statistics>