

Disease

Ornithine transcarbamylase deficiency¹

Gene

OTC

Inheritance	XL
Current Treatments	protein restriction, citrulline, sodium benzoate, phenylbutyrate, Ravicti, liver transplatation
Evidence for Treatment	guideline ^{2, 3}
Non-Molecular Testing Availability	yes
Non-Molecular Test	ammonia level, plasma amino acids, and urine orotate
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC112
Disease Frequency (per 100,000)	1.455 ⁴
Genes Included in Disease Frequency	OTC
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Ornithine+transcarbamylase+deficiency&term=&cntry=&state=&

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK154378/>
- ² <https://newenglandconsortium.org/for-professionals/acute-illness-protocols/urea-cycle-disorders/ornithine-transcarbamylase-deficiency-otc/>
- ³ http://www.bimdg.org.uk/store/guidelines/ER-UCD1-v4_256112_09092016.pdf
- ⁴ https://www.ncbi.nlm.nih.gov/books/NBK154378/#otc-def.Genetically_Related_Allelic_Diso