

Disease

Carbamoyl phosphate synthetase I deficiency¹

Gene

CPS1

Inheritance	AR
Current Treatments	protein restriction, citrulline, sodium benzoate, phenylbutyrate, liver transplatation, N-carbamylglutamate
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	ammonia, plasma amino acid analysis (glutamine and citrulline), urine orotate ³
Disease Frequency (per 100,000)	0.08 ⁴
Genes Included in Disease Frequency	CPS1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Carbamoyl&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/28281899>
- ² http://www.bimdg.org.uk/store/guidelines/ER-UCD1-v4_256112_09092016.pdf
- ³ <https://pubmed.ncbi.nlm.nih.gov/22173106/>
- ⁴ <https://www.ncbi.nlm.nih.gov/books/NBK1217/#ucd-overview.Summary>