

Disease

Citrullinemia¹

Gene

ASS1

Inheritance	AR
Current Treatments	protein restriction, sodium phenylbutyrate, Ravicti, L-carnitine, Liver transplantation
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma amino acid analysis ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1017
Disease Frequency (per 100,000)	1.295 ¹
Genes Included in Disease Frequency	ASS1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Citrullinemia&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1458/#ctIm.Summary>