

Disease

Cystinosis¹

Gene

CTNS

Inheritance	AR
Current Treatments	cysteamine, potassium phosphate, vitamin D, sodium bicarbonate, chlorophyllin to replace copper, carnitine, growth hormone, levothyroxine, insulin, testosterone, renal transplant
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	slit-lamp examination of the eye, leukocytes cystine concentration ³
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC007
Disease Frequency (per 100,000)	0.75 ⁴
Genes Included in Disease Frequency	CTNS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Cystinosis&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1400/#ctns.Summary>
- ² https://cystinosis.org/wp-content/uploads/2019/01/CRN_StandardsOfCare_FINAL-1.pdf
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK1400/>
- ⁴ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3992566/>