

Disease

SLC3A1 associated cystinuria^{1, 2, 3}

Gene

SLC3A1

Inheritance	AR and AD
Current Treatments	increased water consumption, urine alkalinization to a pH >7 by oral citrate, penicillamine and tiopronin
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	urinary cystine excretion ²
Disease Frequency (per 100,000)	14.29 ⁵
Genes Included in Disease Frequency	SLC3A1, SLC7A9
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=Cystinuria&viewType=Table

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/38240263/>
- ² <https://www.ncbi.nlm.nih.gov/books/NBK470527/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34812923/>
- ⁴ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10000469/>
- ⁵ <https://pubmed.ncbi.nlm.nih.gov/37893120/>