

Disease

Wilson disease^{1, 2}

Gene

ATP7B

Inheritance	AR
Current Treatments	zinc, D-penicillamine, trientine
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum ceruloplasmin and copper, urinary copper ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC028
Disease Frequency (per 100,000)	8.665 ^{4, 5}
Genes Included in Disease Frequency	ATP7B
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Wilson+Disease&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1512/>
- ² <https://pubmed.ncbi.nlm.nih.gov/34904029/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/31179302>
- ⁴ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3634195/>
- ⁵ <https://www.ncbi.nlm.nih.gov/pubmed/26207595>