

Disease

Menkes disease^{1, 2}

Gene

ATP7A

Inheritance	XL
Current Treatments	subcutaneous injections of copper histidine or copper chloride
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum ceruloplasmin and copper, plasma catechols ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC013
Disease Frequency (per 100,000)	1 ⁴
Genes Included in Disease Frequency	ATP7A
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Menkes+Disease&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1413/#menkes.Summary>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/28657131>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/30594472>
- ⁴ https://www.ncbi.nlm.nih.gov/books/NBK1413/#menkes.Genetically_Related_Allelic_Disor