

Disease

Aceruloplasminemia¹

Gene

CP

Inheritance	AR
Current Treatments	desferrioxamine, fresh-frozen plasma
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	Serum ceruloplasmin and copper levels ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC157
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Aceruloplasminemia&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1493/>