

Disease

Neuronal ceroid lipofuscinosis 2^{1, 2, 3}

Gene

TPP1

Inheritance	AR
Current Treatments	Cerliponase alfa enzyme replacement therapy (ERT)
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	leukocyte tripeptidyl peptidase enzyme activity ⁵
Disease Frequency (per 100,000)	0.465 ⁶
Genes Included in Disease Frequency	TPP1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Neuronal+Ceroid-Lipofuscinoses&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/204500>
- ² <https://pubmed.ncbi.nlm.nih.gov/33882967/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32280231/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/32684372/>
- ⁵ <https://pubmed.ncbi.nlm.nih.gov/29599076/>
- ⁶ <https://www.ncbi.nlm.nih.gov.ezproxy3.lhl.uab.edu/books/NBK544815/>