

Disease

Transthyretin associated hereditary amyloidosis^{1, 2, 3}

Gene

TTR

Inheritance	AD
Current Treatments	Patisiran, diflunisal, tafamidis, NTLA-2001, eplontersen
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	radionuclide scan ⁵
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC145
Disease Frequency (per 100,000)	1 ^{6, 7}
Genes Included in Disease Frequency	TTR
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Hereditary+Amyloidosis%2C+Transthyretin-Related&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1194/>
- ² <https://pubmed.ncbi.nlm.nih.gov/38053464/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/37869146/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/32476490/>
- ⁵ <https://pubmed.ncbi.nlm.nih.gov/32725834/>
- ⁶ <https://www.nice.org.uk/guidance/gid-hst10014/documents/draft-scope-pre-referral>
- ⁷ <https://ghr.nlm.nih.gov/condition/transthyretin-amyloidosis#statistics>