

Disease

Alpha-1-antitrypsin deficiency^{1, 2}

Gene

SERPINA1

Inheritance	AR
Current Treatments	liver transplantation, infusion of purified human AAT
Evidence for Treatment	guideline ^{3, 4}
Non-Molecular Testing Availability	yes
Non-Molecular Test	AAT serum concentration with PI typing ⁵
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC093
Disease Frequency (per 100,000)	20 ^{1, 6}
Genes Included in Disease Frequency	SERPINA1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SERPINA1&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1519/#alpha1-a.Summary>
- ² <https://pubmed.ncbi.nlm.nih.gov/35748699/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/29191952>
- ⁴ <https://www.ncbi.nlm.nih.gov/pubmed/31564856>
- ⁵ <https://www.ncbi.nlm.nih.gov/books/NBK1519/>
- ⁶ <https://www.nice.org.uk/media/default/About/what-we-do/NICE-guidance/NICE-technology-appraisals/Proposed-appraisals-no-wave/ID8565%20Emphysema%20-%20human%20alpha1-proteinase%20inhibitor%20-%20draft%20scope.pdf>