

Disease

Fabry disease^{1, 2}

Gene

GLA

Inheritance	XL
Current Treatments	agalsidase alfa enzyme replacement
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum globotriaosylsphingosine ⁴
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC047
Disease Frequency (per 100,000)	17.425 ⁵
Genes Included in Disease Frequency	GLA
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Fabry+Disease&term=&cntry=&state=&city=&dist=

References

¹ <https://ojrd.biomedcentral.com/articles/10.1186/s13023-015-0338-2>

² <https://www.ncbi.nlm.nih.gov/books/NBK1292/>

³ <https://www.ncbi.nlm.nih.gov/pubmed/30017653>

⁴ <https://pubmed.ncbi.nlm.nih.gov/32963035/>

⁵ <https://www.mdpi.com/2409-515X/4/3/24/htm>