

Disease

Glycogen storage disease II¹

Gene

GAA

Inheritance	AR
Current Treatments	avalglucosidase alfa and alglucosidase alfa enzyme replacement therapy
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	Leukocyte enzyme activity ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC090
Disease Frequency (per 100,000)	6.85 ³
Genes Included in Disease Frequency	GAA
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=pompe&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1261/>
- ² https://pediatrics.aappublications.org/content/140/Supplement_1/S24
- ³ <https://pubmed.ncbi.nlm.nih.gov/33717985/>