

Disease

Gaucher disease, type I^{1, 2}

Gene

GBA

Inheritance	AR
Current Treatments	enzyme replacement therapy (ERT): imiglucerase (Cerezyme); velaglucerase alfa (VPRIV); and taliglucerase alfa (Elelyso) or substrate reduction therapy (SRT) miglustat. Ambroxol
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	glucocerebrosidase enzyme activity in leukocytes ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC104
Disease Frequency (per 100,000)	1.94 ⁴
Genes Included in Disease Frequency	GBA
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Gaucher+Disease&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1269/>
- ² <https://www.sciencedirect.com/science/article/pii/S2214426920301245>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/28274788>
- ⁴ <https://www.mdpi.com/2409-515X/4/3/24/htm>