

Disease

Glycogen storage disease VI^{1, 2}

Gene

PYGL

Inheritance	AR
Current Treatments	high-protein diet with cornstarch supplementation
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	glycogen phosphorylase activity in frozen liver biopsy tissue, leukocytes, and erythrocytes ⁴
Disease Frequency (per 100,000)	1.36 ¹
Genes Included in Disease Frequency	PYGL
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Glycogen+Storage+Disease+VI&term=&cntry=&state=&city=&rank=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK5941/#gsd6.Summary>

² <https://pubmed.ncbi.nlm.nih.gov/35725468/>

³ <https://www.ncbi.nlm.nih.gov/pubmed/30659246>

⁴ <https://pubmed.ncbi.nlm.nih.gov/30659246/>