

Disease

Congenital disorder of glycosylation, type Ia^{1, 2}

Gene

PMM2

Inheritance	AR
Current Treatments	epalrestat
Evidence for Treatment	guideline ¹
Non-Molecular Testing Availability	yes
Non-Molecular Test	phosphomannomutase activity in leukocytes ¹
Disease Frequency (per 100,000)	5 ¹
Genes Included in Disease Frequency	PMM2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Congenital+Disorders+of+Glycosylation&term=&cntry=&state=&

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/30740725>

² <https://www.ncbi.nlm.nih.gov/pubmed/31636082>