

Disease

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

Gene

PGM1

Inheritance	AR
Current Treatments	D-galactose
Evidence for Treatment	guideline
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum liquid chromatography / mass spectrometry (LC/MS) ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Congenital+Disorders+of+Glycosylation&term=&cntry=&state=&

References

¹ <https://pubmed.ncbi.nlm.nih.gov/28617415/>

² <https://pubmed.ncbi.nlm.nih.gov/32681750/>