

Disease

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

Gene

MPI

Inheritance	AR
Current Treatments	mannose
Evidence for Treatment	guideline ³
Non-Molecular Testing Availability	yes
Non-Molecular Test	phosphomannose isomerase activity in leukocytes 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/19101627/>

² <https://omim.org/entry/602579>

³ <https://pubmed.ncbi.nlm.nih.gov/32266963/>