

Disease

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

Gene

SLC39A8

Inheritance	AR
Current Treatments	manganese, galactose, uridine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	Mn level ³
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/28749473>

² <https://www.ncbi.nlm.nih.gov/pubmed/26637979>

³ <https://pubmed.ncbi.nlm.nih.gov/31089831/>