

Disease

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

Gene

TMEM165

Inheritance	AR
Current Treatments	D-galactose
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum transferrin isoelectric focusing ²
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/28323990>

² <https://omim.org/entry/614727>