

## Disease

# Congenital disorder of glycosylation, type IIc<sup>1, 2, 3</sup>

## Gene

### SLC35C1

|                                                 |                                                                                                                                                                                                                                                             |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                          |
| <b>Current Treatments</b>                       | fucose                                                                                                                                                                                                                                                      |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                              |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                                         |
| <b>Non-Molecular Test</b>                       | ABO blood group testing for H antigen <sup>2</sup>                                                                                                                                                                                                          |
| <b>Disease Frequency (per 100,000)</b>          | 1 <sup>4</sup>                                                                                                                                                                                                                                              |
| <b>Genes Included in Disease Frequency</b>      | ITGB2, FERMT3, SLC35C1                                                                                                                                                                                                                                      |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=Congenital+Disorders+of+Glycosylation&amp;term=&amp;cntry=&amp;state=&amp;">https://clinicaltrials.gov/ct2/results?cond=Congenital+Disorders+of+Glycosylation&amp;term=&amp;cntry=&amp;state=&amp;</a> |

## References

- <sup>1</sup> <https://www.omim.org/entry/266265>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/35338746/>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/32313197/>
- <sup>4</sup> <https://www.ncbi.nlm.nih.gov/pubmed/30412664>