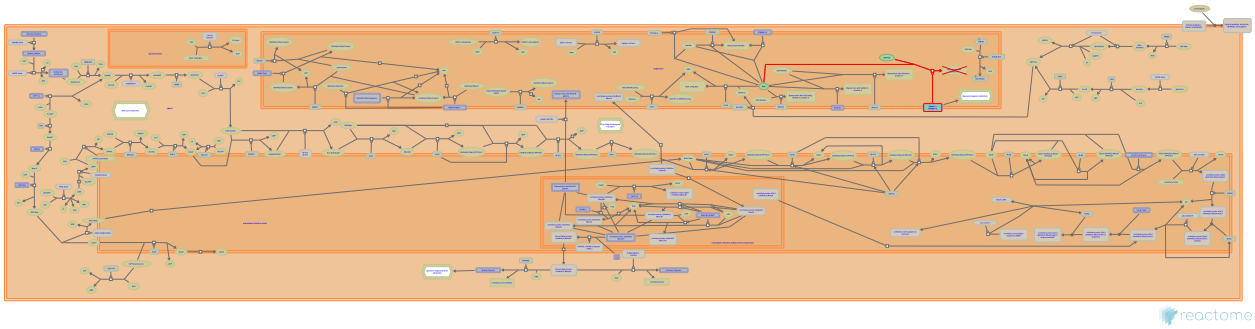
Defective B4GALT1 causes CDG-2d



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Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

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Literature references

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

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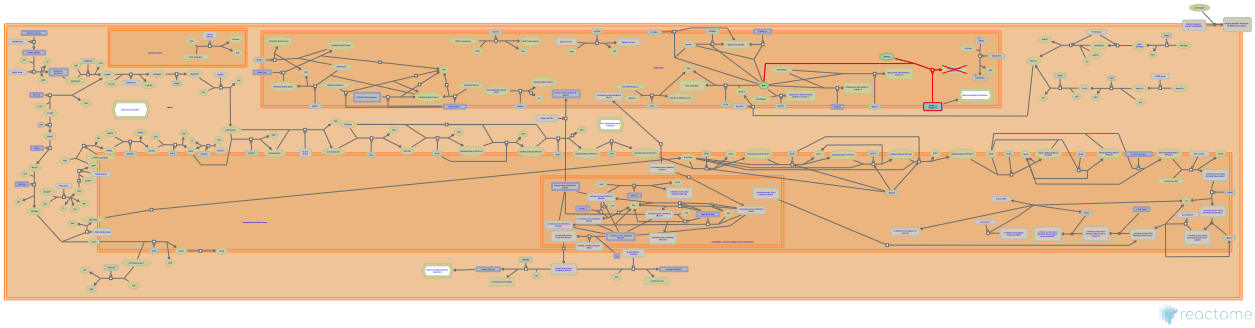
Defective B4GALT1 causes CDG-2d ↗

Stable identifier: R-HSA-4793953

Diseases: congenital disorder of glycosylation type II



Congenital disorders of glycosylation (CDG, previously called carbohydrate-deficient glycoprotein syndromes, CDGSs), are a group of hereditary multisystem disorders. They are characterized biochemically by hypoglycosylation of glycoproteins, diagnosed by isoelectric focusing (IEF) of serum transferrin. There are two types of CDG, types I and II. Type I CDG has defects in the assembly of lipid-linked oligosaccharides or their transfer onto nascent glycoproteins, whereas type II CDG comprises defects of trimming, elongation, and processing of protein-bound glycans. Clinical symptoms are dominated by severe psychomotor and mental retardation, as well as blood coagulation abnormalities (Jaeken 2013). B4GALT1-CDG (CDG type IId) is a multisystem disease, characterized by dysmorphic features, hydrocephalus, hypotonia and blood clotting abnormalities (Hansske et al. 2002).

Literature references

Jaeken, J. (2013). Congenital disorders of glycosylation. *Handb Clin Neurol, 113*, 1737-43. ↗

Hoffmann, GF., Lübke, T., Peters, V., Hansske, B., Körner, C., Heidemann, PH. et al. (2002). Deficiency of UDP-galactose:N-acetylglucosamine beta-1,4-galactosyltransferase I causes the congenital disorder of glycosylation type IId. *J Clin Invest, 109*, 725-33. ↗

Editions

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Defective B4GALT1 does not add Gal to N-glycan ↗

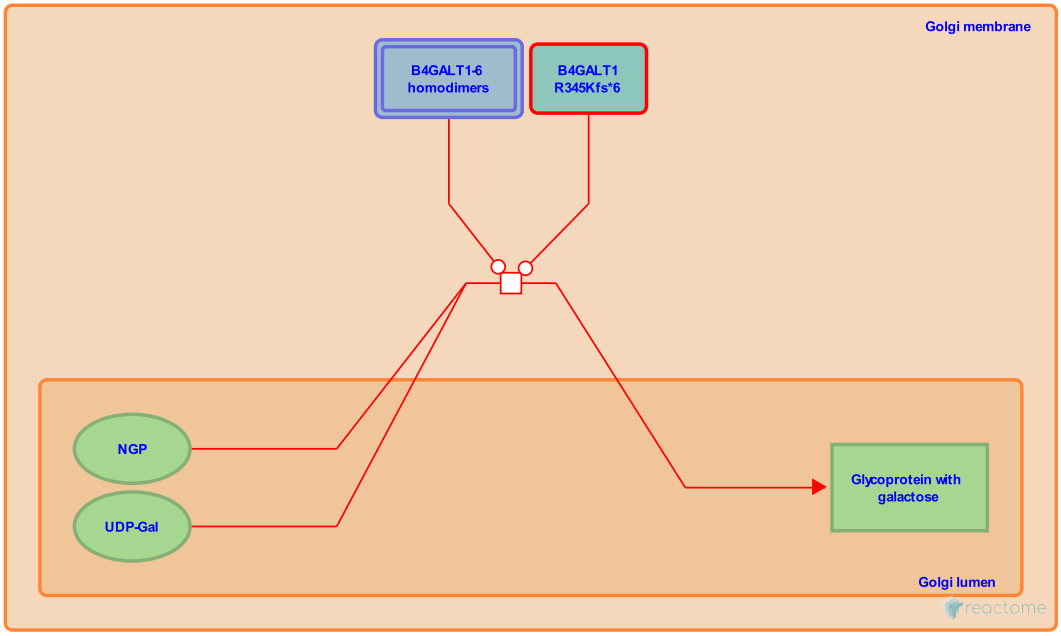
Location: Defective B4GALT1 causes CDG-2d

Stable identifier: R-HSA-4793956

Type: transition

Compartments: Golgi membrane, Golgi lumen

Diseases: congenital disorder of glycosylation type II



The family of beta 4-galactosyltransferases (B4GALTs) is composed by at least six known members that mediate the transfer of galactose to N-glycan structures and either to begin or elongate keratan chains. Defective B4GALT1 is associated with congenital disorder of glycosylation type IId (B4GALT1-CDG, CDG-2d; MIM:607091), in which clinical symptoms are dominated by dysmorphic features, psychomotor and mental retardation, hypotonia, as well as blood coagulation abnormalities (Hansske et al. 2002). The mutant R345Kfs*6 results in a truncated, inactive polypeptide. Analysis of oligosaccharides from serum transferrin from these patients reveals loss of sialic acid and galactose residues (Hansske et al. 2002).

Literature references

Hoffmann, GF., Lübke, T., Peters, V., Hansske, B., Körner, C., Heidemann, PH. et al. (2002). Deficiency of UDP-galactose:N-acetylglucosamine beta-1,4-galactosyltransferase I causes the congenital disorder of glycosylation type IId. *J Clin Invest, 109*, 725-33. ↗

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