Defective ALG3 does not add mannose to the N-glycan precursor

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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

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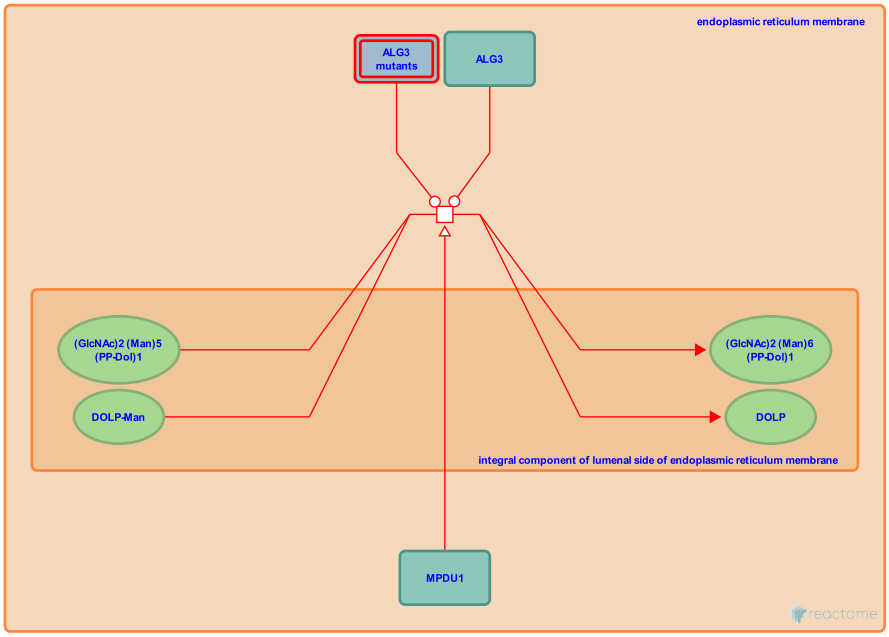
Stable identifier: R-HSA-4720473

Type: transition

Compartments: endoplasmic reticulum membrane, integral component of lumenal side of endoplasmic reticulum membrane

Diseases: congenital disorder of glycosylation type I



Dol-P-Man:Man(5)GlcNAc(2)-PP-Dol alpha-1,3-mannosyltransferase (ALG3) adds the sixth mannose (although the first to be derived from dolichyl-phosphate-mannose, DOLPman) to the lipid-linked oligosaccharide intermediate GlcNAc(2) Man(5) (PPDol)1 (Korner et al. 1999). Defects in ALG3 are associated with congenital disorder of glycosylation 1d (ALG3-CDG, CDG1d; MIM:601110), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins. CDG type 1 diseases result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. (Sun et al. 2005). Point mutations that cause ALG3-CDG are G118D, R171Q, W71R and M157K (Korner et al. 1999, Sun et al. 2005, Kranz et al. 2007).

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Editions

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