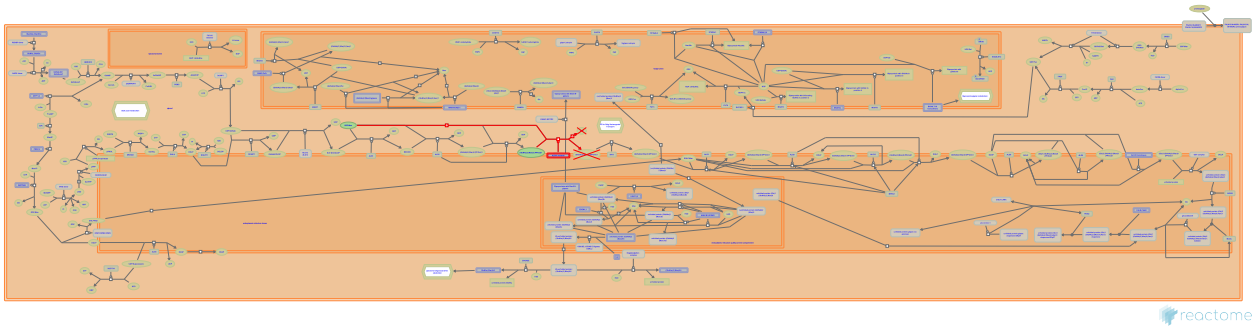
Defective ALG11 causes CDG-1p



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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.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

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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. ↗

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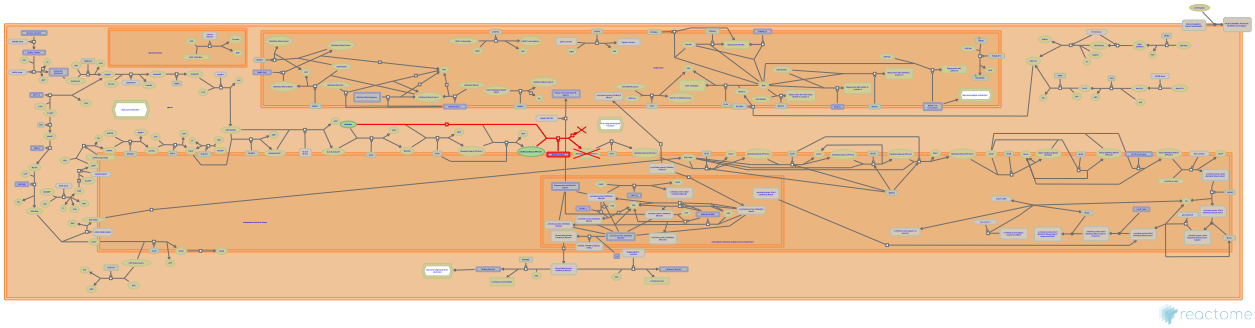
Defective ALG11 causes CDG-1p ↗

Stable identifier: R-HSA-4551295

Diseases: congenital disorder of glycosylation type I



GDP-Man:Man(3)GlcNAc(2)-PP-Dol alpha-1,2-mannosyltransferase (ALG11) transfers the fourth and fifth mannoses (Man) to the N-glycan precursor in an alpha-1,2 orientation. These additions are the last two on the cytosolic side of the ER membrane before the N-glycan is flipped to the luminal side of the membrane. Recently discovered defects in ALG11 have been linked to congential disorder of glycosylation, type 1p (ALG11-CDG, CGD1p) (Rind et al. 2010, Thiel et al. 2012). The disease is a multi-system disorder characterised by under-glycosylated serum glycoproteins. Early-onset developmental retardation, dysmorphic features, hypotonia, coagulation disorders and immunodeficiency are reported features of this disorder (Rind et al. 2010, Thiel et al. 2012).

Literature references

Wilichowski, E., Schmeiser, V., Hocks, J., Apeshiotis, N., Thiel, C., Lehle, L. et al. (2010). A severe human metabolic disease caused by deficiency of the endoplasmatic mannosyltransferase hALG11 leads to congenital disorder of glycosylation-Ip. *Hum. Mol. Genet., 19*, 1413-24. ↗

Thiel, C., Thiels, C., Conway, RL., Hoffmann, GF., Apeshiotis, N., Popovici, D. et al. (2012). Improved diagnostics lead to identification of three new patients with congenital disorder of glycosylation-Ip. *Hum. Mutat., 33*, 485-7. ↗

Editions

2013-09-13	Authored, Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

Defective ALG11 does not transfer Man to the N-glycan precursor ↗

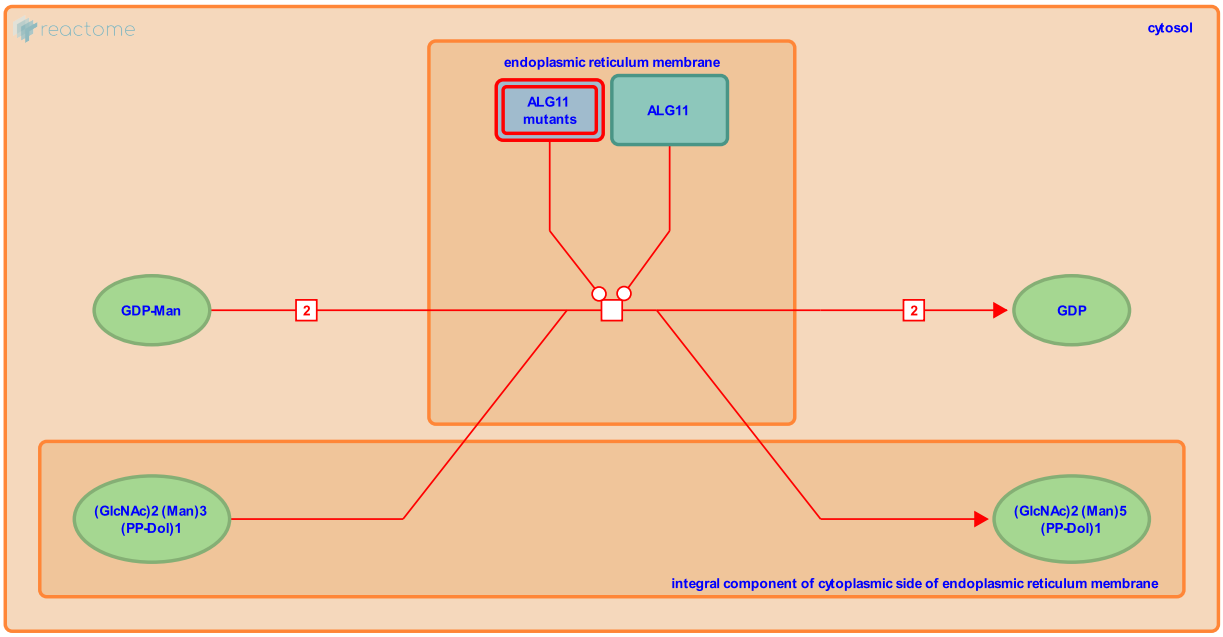
Location: Defective ALG11 causes CDG-1p

Stable identifier: R-HSA-4551297

Type: transition

Compartments: endoplasmic reticulum membrane, integral component of cytoplasmic side of endoplasmic reticulum membrane, cytosol

Diseases: congenital disorder of glycosylation type I



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Literature references

Wilichowski, E., Schmeiser, V., Hocks, J., Apeshiotis, N., Thiel, C., Lehle, L. et al. (2010). A severe human metabolic disease caused by deficiency of the endoplasmatic mannosyltransferase hALG11 leads to congenital disorder of glycosylation-Ip. *Hum. Mol. Genet., 19*, 1413-24. ↗

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Editions

2013-09-13	Authored, Edited	Jassal, B.
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