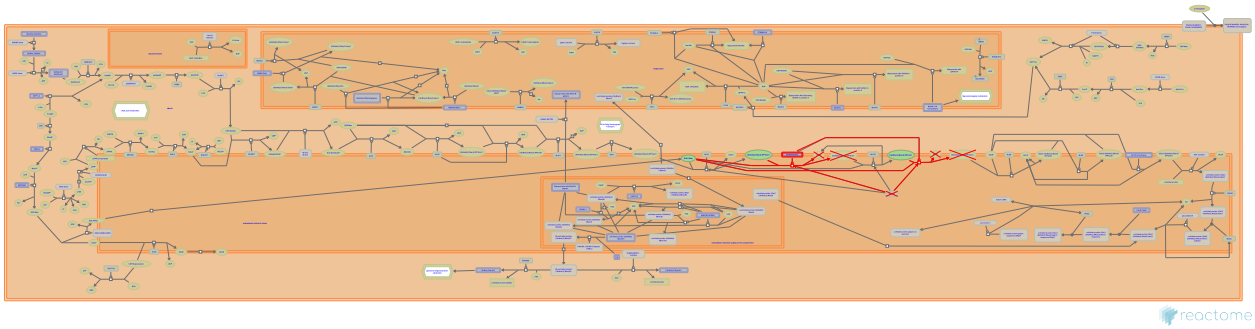
Defective ALG9 causes CDG-1l



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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. ↗

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

Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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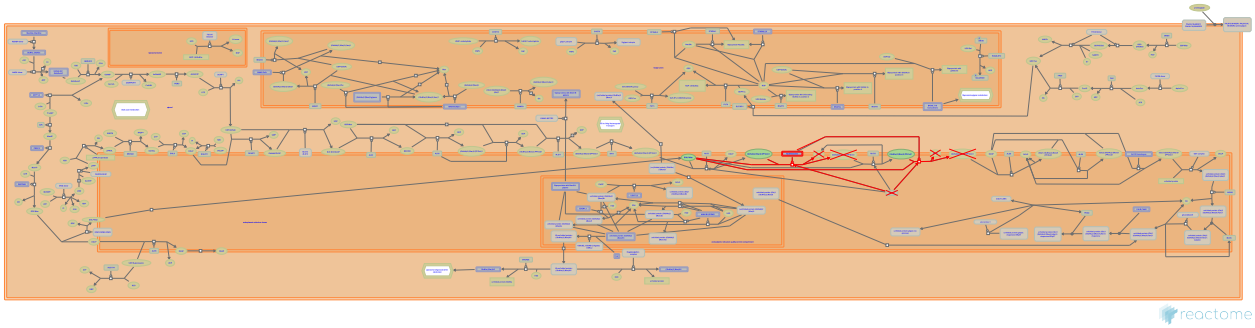
Defective ALG9 causes CDG-1l ↗

Stable identifier: R-HSA-4720454

Diseases: congenital disorder of glycosylation type I



Alpha-1,2-mannosyltransferase ALG9 (ALG9) normally catalyses the transfer of mannose to the lipid-linked oligosaccharide (LLO) precursor. It adds the 7th and 9th mannose moieties to LLO. Defects in ALG9 are associated with congenital disorder of glycosylation 1l (ALG9-CDG, CDG1l; MIM:608776), 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 (Frank et al. 2004, Weinstein et al. 2005). The LLO profile showed accumulation of (GlcNAc)2 (Man)6 (PP-Dol)1 and (GlcNAc)2 (Man)8 (PP-Dol)1 fragments, suggesting a defect in ALG9 and correlating with the normal function of ALG9 in adding the 7th and 9th mannose moieties (Frank et al. 2004).

Literature references

Hennet, T., Eyaid, W., Frank, CG., Grubenmann, CE., Berger, EG., Aebi, M. (2004). Identification and functional analysis of a defect in the human ALG9 gene: definition of congenital disorder of glycosylation type IL. *Am J Hum Genet, 75*, 146-50. ↗

Hennet, T., Clarke, JT., Frank, CG., Seargeant, L., Matthijs, G., Neupert, C. et al. (2005). CDG-IL: an infant with a novel mutation in the ALG9 gene and additional phenotypic features. *Am J Med Genet A, 136*, 194-7. ↗

Editions

2013-10-21	Authored, Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

Defective ALG9 does not add the seventh mannose to the N-glycan precursor ↗

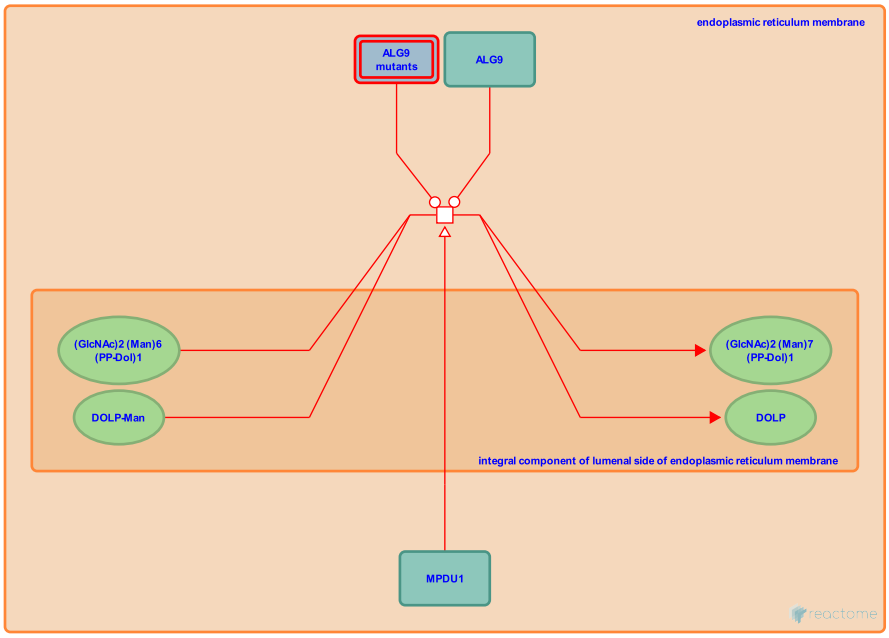
Location: Defective ALG9 causes CDG-1l

Stable identifier: R-HSA-4720478

Type: transition

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

Diseases: congenital disorder of glycosylation type I



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Literature references

Hennet, T., Eyaid, W., Frank, CG., Grubenmann, CE., Berger, EG., Aebi, M. (2004). Identification and functional analysis of a defect in the human ALG9 gene: definition of congenital disorder of glycosylation type IL. *Am J Hum Genet, 75*, 146-50. ↗

Hennet, T., Clarke, JT., Frank, CG., Seargeant, L., Matthijs, G., Neupert, C. et al. (2005). CDG-IL: an infant with a novel mutation in the ALG9 gene and additional phenotypic features. *Am J Med Genet A, 136*, 194-7. ↗

Editions

2013-10-21	Authored, Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

Defective ALG9 does not add the last mannose to the N-glycan precursor ↗

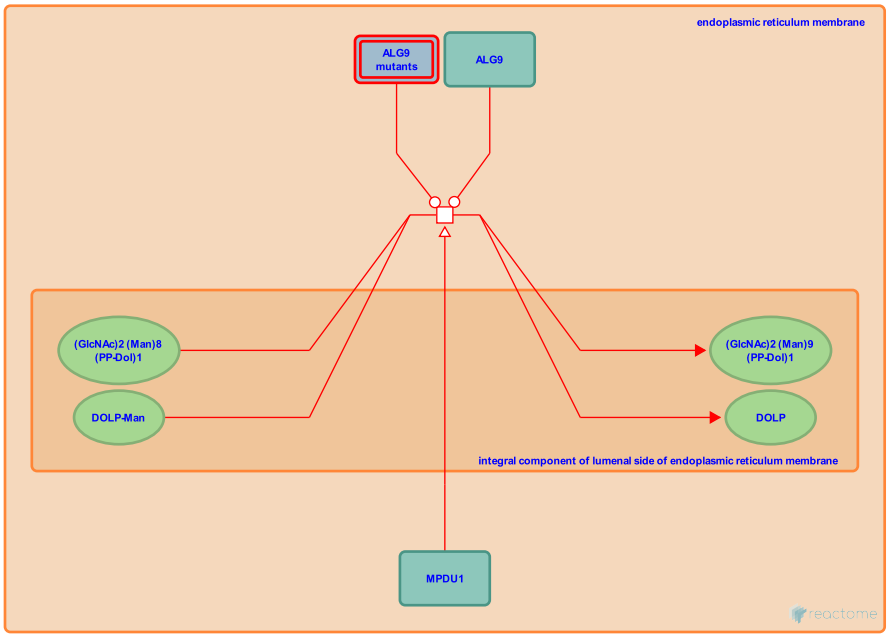
Location: Defective ALG9 causes CDG-1l

Stable identifier: R-HSA-9035514

Type: transition

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

Diseases: congenital disorder of glycosylation type I



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Editions

2013-10-21	Authored, Edited	Jassal, B.
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