Defective ALG9 does not add the seventh 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


Reactome database release: 83

This document contains 1 reaction (see Table of Contents)

https://reactome.org
Defective ALG9 does not add the seventh mannose to the N-glycan precursor

Stable identifier: R-HSA-4720478

Type: transition

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

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 type I (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). Point mutations that can cause ALG9-CDG are E523K and Y286C (Frank et al. 2004, Weinstein et al. 2005).

Literature references


Editions

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