

# Defective ALG1 does not transfer the first Man 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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Reactome database release: 88

This document contains 1 reaction ([see Table of Contents](#))

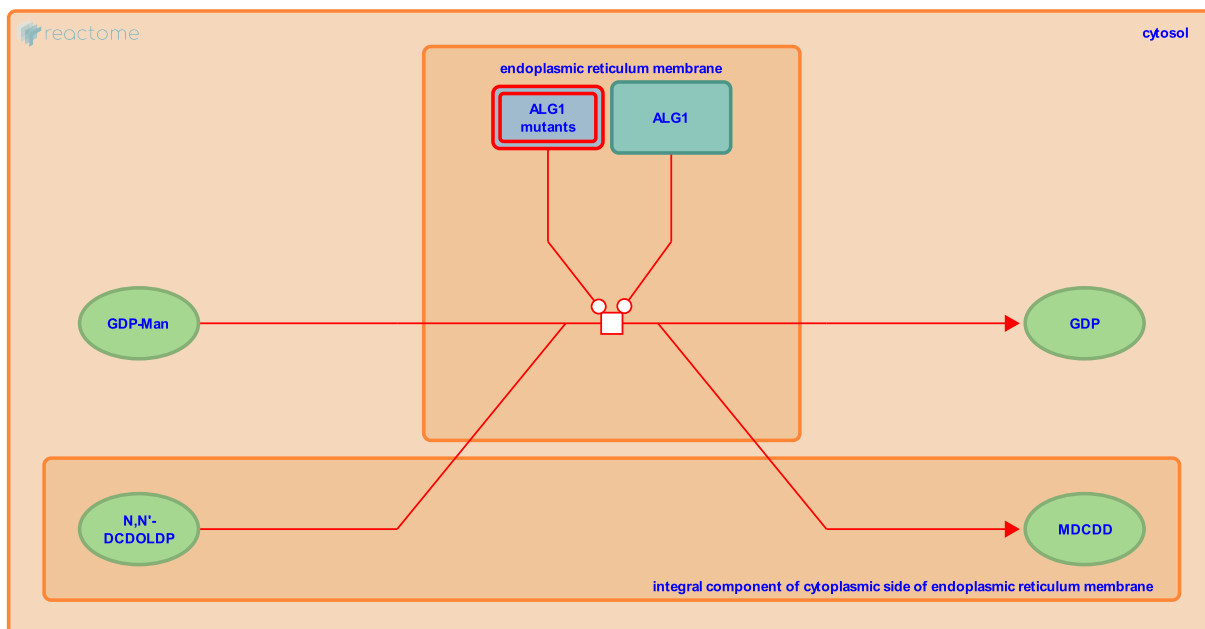
## Defective ALG1 does not transfer the first Man to the N-glycan precursor [↗](#)

**Stable identifier:** R-HSA-4549382

**Type:** transition

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

**Diseases:** congenital disorder of glycosylation type I



Chitobiosyldiphosphodolichol beta-mannosyltransferase (ALG1) normally transfers a mannose moiety to the lipid-linked oligosaccharide (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins. Defects in ALG1 can cause congenital disorder of glycosylation 1k (ALG1-CDG, previously known as CDG1k; MIM:608540), a multisystem disorder 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. Compared to other CDGs, ALG1-CDG has a very severe phenotype, which can result in an early death. Mutations in ALG1 causing ALG1-CDG include S258L, G342P, S150R, M377V, G145D, C396\* and R276W (Schwarz et al. 2004, Kranz et al. 2004, Grubenmann et al. 2004, Dupre et al. 2010).

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### Editions

2013-09-12

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2014-10-31

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