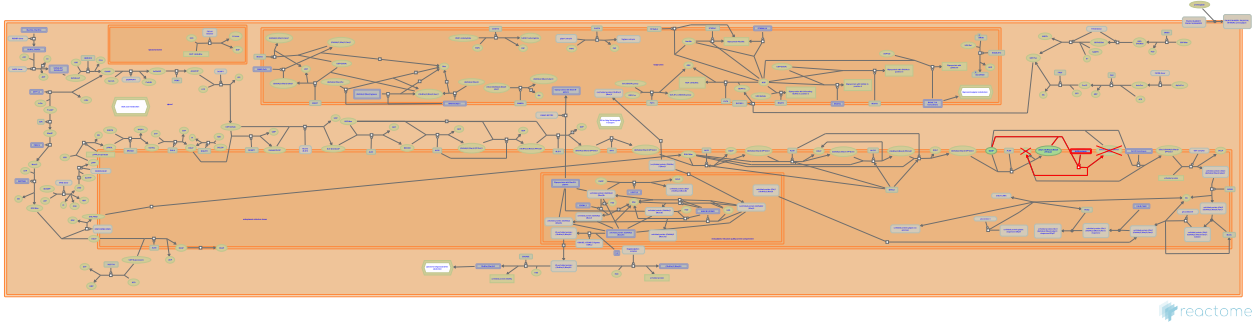


# Defective ALG8 causes CDG-1h



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This is just an excerpt of a full-length report for this pathway. To access the complete report, please download it at the [Reactome Textbook](#).

01/05/2024

## 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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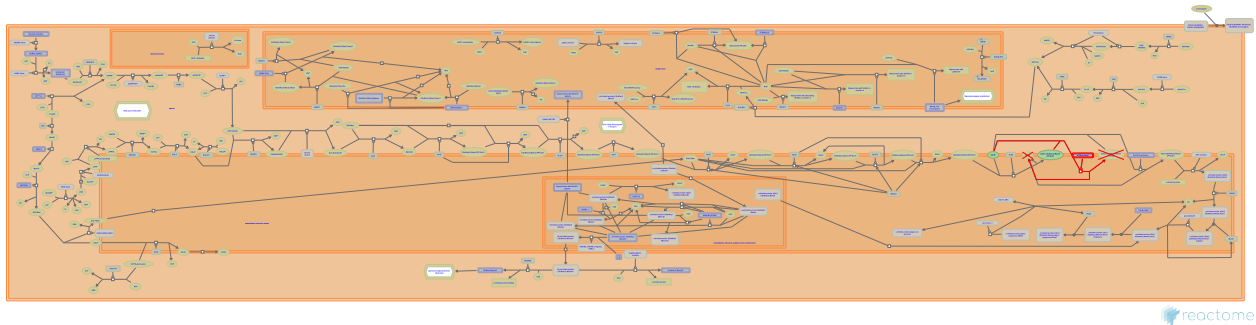
Reactome database release: 88

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

## Defective ALG8 causes CDG-1h [↗](#)

**Stable identifier:** R-HSA-4724325

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



The probable dolichyl pyrophosphate Glc1Man9GlcNAc2 alpha-1,3-glycosyltransferase (ALG8) (Stanchi et al. 2001, Chantret et al. 2003) normally adds the second glucose moiety to the lipid-linked oligosaccharide precursor (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins. Defects in ALG8 can cause congenital disorder of glycosylation 1h (ALG8-CDG, CDG-1h; MIM:608104), a multisystem disorder characterised by under-glycosylated serum glycoproteins (Chantret et al. 2003, Schollen et al. 2004). ALG8 deficiency is accompanied by an accumulation of the N-glycan precursor (Glc)1 (GlcNAc)2 (Man)9 (PP-Dol)1. 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.

### Literature references

- Codogno, P., Chantret, I., Vuillaumier-Barrot, S., Oriol, R., Moore, SE., Danos, O. et al. (2003). A deficiency in dolichyl-P-glucose:Glc1Man9GlcNAc2-PP-dolichyl alpha3-glycosyltransferase defines a new subtype of congenital disorders of glycosylation. *J. Biol. Chem.*, 278, 9962-71. [↗](#)
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### Editions

2013-10-23	Authored, Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

## Defective ALG8 does not add glucose to the N-glycan precursor ↗

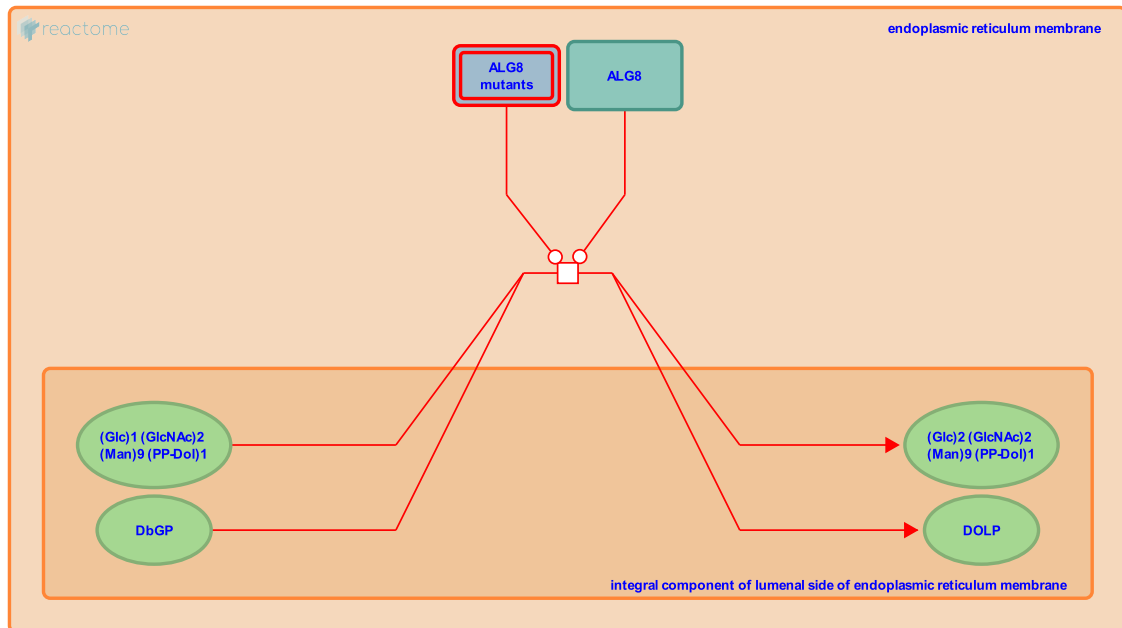
**Location:** [Defective ALG8 causes CDG-1h](#)

**Stable identifier:** R-HSA-4724330

**Type:** transition

**Compartments:** endoplasmic reticulum membrane, integral component of luminal side of endoplasmic reticulum membrane

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



The probable dolichyl pyrophosphate Glc1Man9GlcNAc2 alpha-1,3-glucosyltransferase (ALG8) (Chantret et al. 2003) normally adds the second glucose moiety to the lipid-linked oligosaccharide precursor (LLO aka N-glycan precursor) which is required for subsequent N-glycosylation of proteins. Defects in ALG8 can cause congenital disorder of glycosylation 1h (ALG8-CDG, CDG-1h; MIM:608104), a multisystem disorder characterised by underglycosylated serum glycoproteins (Chantret et al. 2003, Schollen et al. 2004). ALG8 deficiency is accompanied by an accumulation of the N-glycan precursor (Glc)1 (GlcNAc)2 (Man)9 (PP-Dol)1. Mutations that can cause ALG8-CDG include T47P, G275D, V133Sfs\*3 and T138Kfs\*19 (Chantret et al. 2003, Schollen et al. 2004).

### Literature references

Codogno, P., Chantret, I., Vuillaumier-Barrot, S., Oriol, R., Moore, SE., Danos, O. et al. (2003). A deficiency in dolichyl-P-glucose:Glc1Man9GlcNAc2-PP-dolichyl alpha3-glucosyltransferase defines a new subtype of congenital disorders of glycosylation. *J. Biol. Chem.*, 278, 9962-71. ↗

Reyntjens, R., Henet, T., Wevers, RA., Smeitink, J., Frank, CG., Matthijs, G. et al. (2004). Clinical and molecular features of three patients with congenital disorders of glycosylation type 1h (CDG-1h) (ALG8 deficiency). *J Med Genet*, 41, 550-6. ↗

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