

# Defective ALG3 does not add mannose to the N-glycan precursor

Belaya, K., Jassal, B.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of [Creative Commons Attribution 4.0 International \(CC BY 4.0\) License](#). For more information see our [license](#).

30/04/2024

## 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.

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).

## Literature references

- 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. [↗](#)
- Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology*, 14, e1005968. [↗](#)

Reactome database release: 88

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

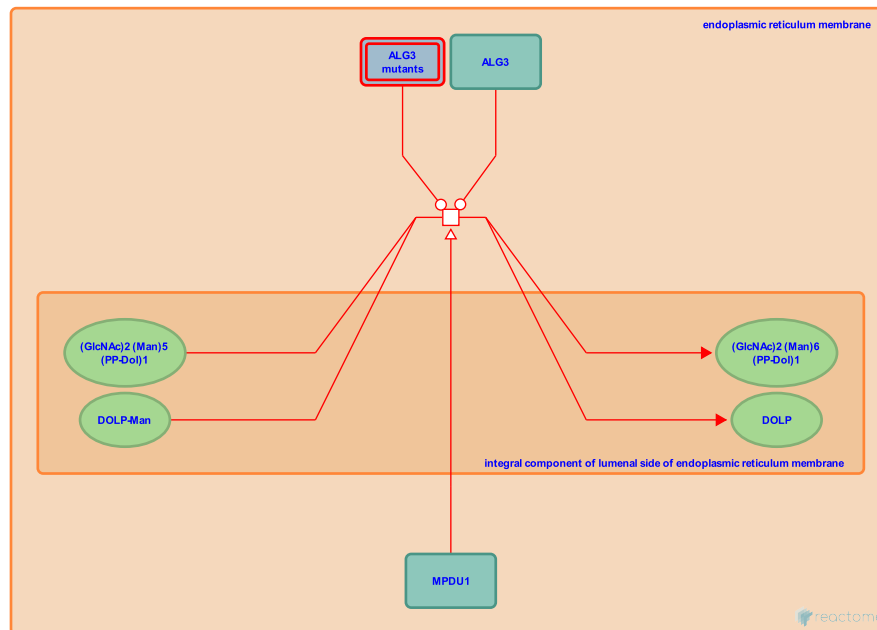
## Defective ALG3 does not add mannose to the N-glycan precursor [↗](#)

**Stable identifier:** R-HSA-4720473

**Type:** transition

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

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



Dol-P-Man:Man(5)GlcNAc(2)-PP-Dol alpha-1,3-mannosyltransferase (ALG3) adds the sixth mannose (although the first to be derived from dolichyl-phosphate-mannose, DOLPman) to the lipid-linked oligosaccharide intermediate GlcNAc(2) Man(5) (PPDol)1 (Korner et al. 1999). Defects in ALG3 are associated with congenital disorder of glycosylation 1d (ALG3-CDG, CDG1d; MIM:601110), 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. (Sun et al. 2005). Point mutations that cause ALG3-CDG are G118D, R171Q, W71R and M157K (Korner et al. 1999, Sun et al. 2005, Kranz et al. 2007).

### Literature references

- Körner, C., Lehle, L., von Figura, K., Marquardt, T., Stephani, U., Knauer, R. (1999). Carbohydrate deficient glycoprotein syndrome type IV: deficiency of dolichyl-P-Man:Man(5)GlcNAc(2)-PP-dolichyl mannosyltransferase. *EMBO J.*, 18, 6816-22. [↗](#)
- Krasnewich, D., Freeze, HH., Sun, L., Casey, JR., Eklund, EA., Kranz, C. (2007). CDG-Id in two siblings with partially different phenotypes. *Am. J. Med. Genet. A*, 143, 1414-20. [↗](#)
- Cohen, J., Chung, WK., Freeze, HH., Wang, C., Eklund, EA., Sun, L. (2005). Congenital disorder of glycosylation id presenting with hyperinsulinemic hypoglycemia and islet cell hyperplasia. *J Clin Endocrinol Metab*, 90, 4371-5. [↗](#)

### Editions

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