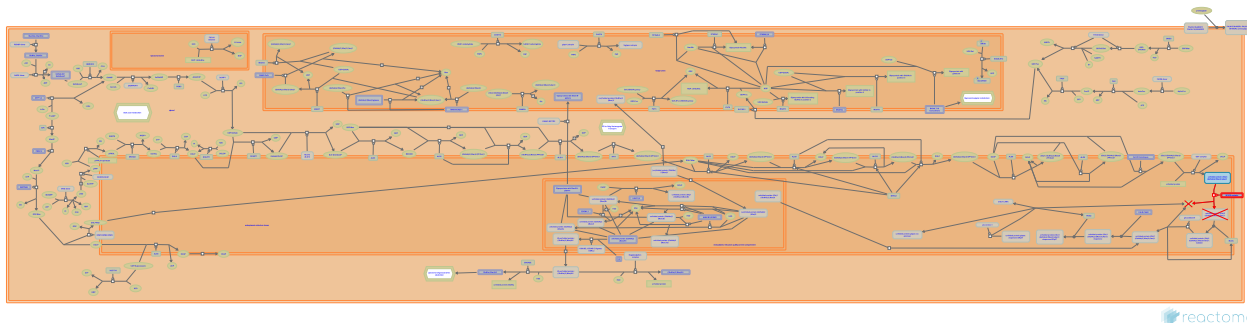


# Defective MOGS causes CDG-2b



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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](https://reactome.org/textbook).

29/04/2024

## 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

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- 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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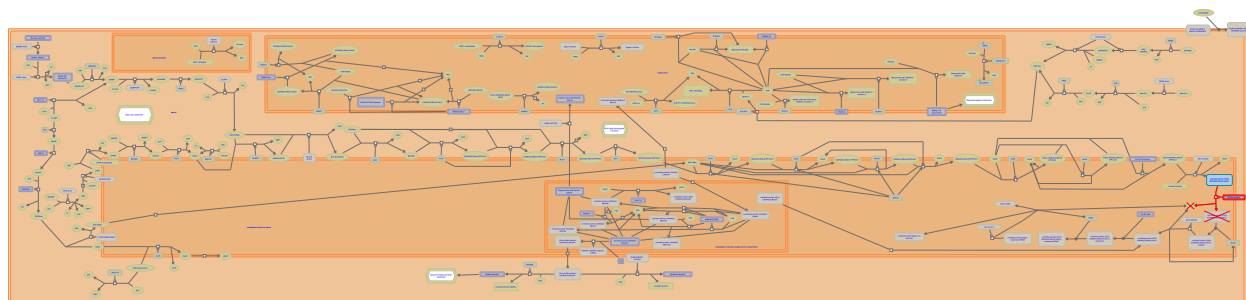
Reactome database release: 88

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

## Defective MOGS causes CDG-2b [↗](#)

**Stable identifier:** R-HSA-4793954

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



After the lipid-linked oligosaccharide (LLO) precursor is attached to the protein, the outer alpha-1,2-linked glucose is removed by by mannosyl-oligosaccharide glucosidase (MOGS). This is a mandatory step for protein folding control and glycan extension. Defects in MOGS are associated with congenital disorder of glycosylation type IIb (CDGIIb), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins (De Praeter et al. 2000, Voelker et al. 2002). Type II CDGs refer to defects in the trimming and processing of protein-bound glycans.

### Literature references

Espeel, MF., Chan, NW., Nuytinck, LK., Martin, JJ., Gerwig, GJ., Kamerling, JP. et al. (2000). A novel disorder caused by defective biosynthesis of N-linked oligosaccharides due to glucosidase I deficiency. *Am J Hum Genet*, 66, 1744-56. [↗](#)

Hardt, B., Kalz-Füller, B., Bause, E., De Praeter, CM., Breuer, W., Van Coster, RN. et al. (2002). Processing of N-linked carbohydrate chains in a patient with glucosidase I deficiency (CDG type IIb). *Glycobiology*, 12, 473-83. [↗](#)

### Editions

2013-07-29	Authored	Jassal, B.
2013-10-29	Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

## Defective MOGS does not cleave glucose from an N-glycosylated protein ↗

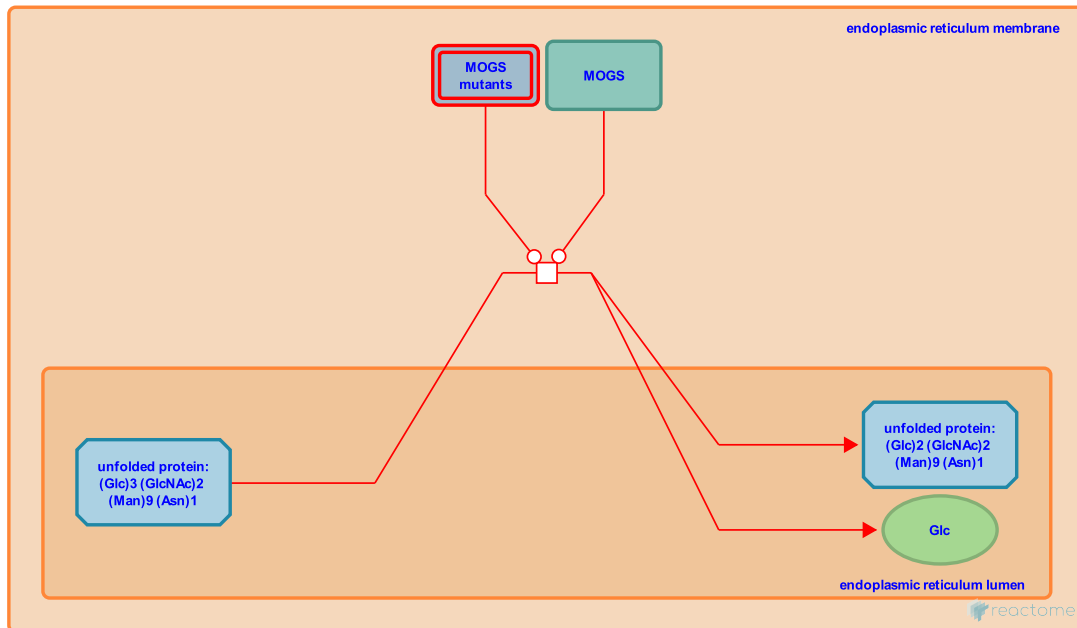
**Location:** [Defective MOGS causes CDG-2b](#)

**Stable identifier:** R-HSA-4793947

**Type:** transition

**Compartments:** endoplasmic reticulum membrane, endoplasmic reticulum lumen

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



After the lipid-linked oligosaccharide (LLO) precursor is attached to the protein, the outer alpha-1,2-linked glucose is removed by by mannosyl-oligosaccharide glucosidase (MOGS). This is a mandatory step for protein folding control and glycan extension. Defects in MOGS are associated with congenital disorder of glycosylation type IIb (MOGS-CDG, CDGIIB; MIM:606056), a multisystem disorder caused by a defect in glycoprotein biosynthesis and characterised by under-glycosylated serum glycoproteins. Mutations causing MOGS-CDG are R486T and F652L. Kinetic studies using cultured fibroblasts showed that the by mannosyl-oligosaccharide glucosidase activity in the patient's cells was < 1% of control activity (De Praeter et al. 2000, Voelker et al. 2002).

### Literature references

Espeel, MF., Chan, NW., Nuytinck, LK., Martin, JJ., Gerwig, GJ., Kamerling, JP. et al. (2000). A novel disorder caused by defective biosynthesis of N-linked oligosaccharides due to glucosidase I deficiency. *Am J Hum Genet*, 66, 1744-56. ↗

Hardt, B., Kalz-Füller, B., Bause, E., De Praeter, CM., Breuer, W., Van Coster, RN. et al. (2002). Processing of N-linked carbohydrate chains in a patient with glucosidase I deficiency (CDG type IIb). *Glycobiology*, 12, 473-83. ↗

### Editions

2013-07-29	Authored	Jassal, B.
2013-10-29	Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

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