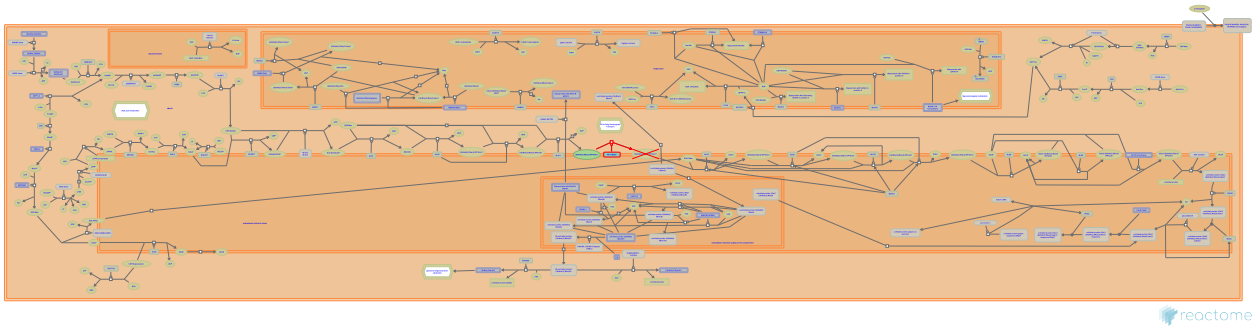
Defective RFT1 causes CDG-1n



Belaya, K., Jassal, B.

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

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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

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. ↗

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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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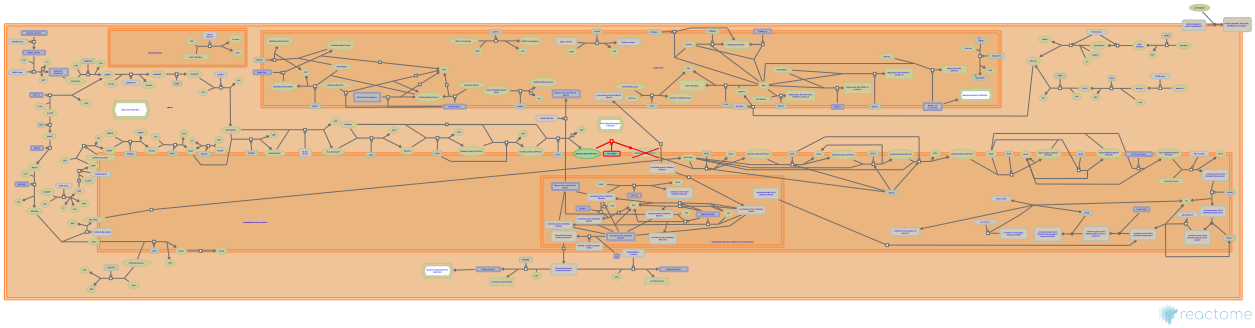
Defective RFT1 causes CDG-1n ↗

Stable identifier: R-HSA-4570571

Diseases: congenital disorder of glycosylation type I



The N-glycan precursor is flipped across the ER membrane, moving it from the cytosolic side to the ER lumenal side. The exact mechanism of this translocation is not well understood but protein RFT1 homolog (RFT1) is known to be involved (Helenius et al. 2002). Defects in RFT1 are associated with congenital disorder of glycosylation 1n (RFT1-CDG, CDG-1n). The disease is a multi-system disorder characterised by under-glycosylated serum glycoproteins. Early-onset developmental retardation, dysmorphic features, hypotonia, coagulation disorders and immunodeficiency are reported features of this disorder (Haeuptle et al. 2008).

Literature references

Hennet, T., Haeuptle, MA., Kastaniotis, AJ., Neupert, C., Pujol, FM., Winchester, B. et al. (2008). Human RFT1 deficiency leads to a disorder of N-linked glycosylation. *Am J Hum Genet, 82*, 600-6. ↗

Walter, P., Valvano, MA., Helenius, J., Marolda, CL., Aebi, M., Ng, DT. (2002). Translocation of lipid-linked oligosaccharides across the ER membrane requires Rft1 protein. *Nature, 415*, 447-50. ↗

Editions

2013-09-23	Authored, Edited	Jassal, B.
2014-10-31	Reviewed	Belaya, K.

Defective RFT1 does not flip the N-glycan precursor ↗

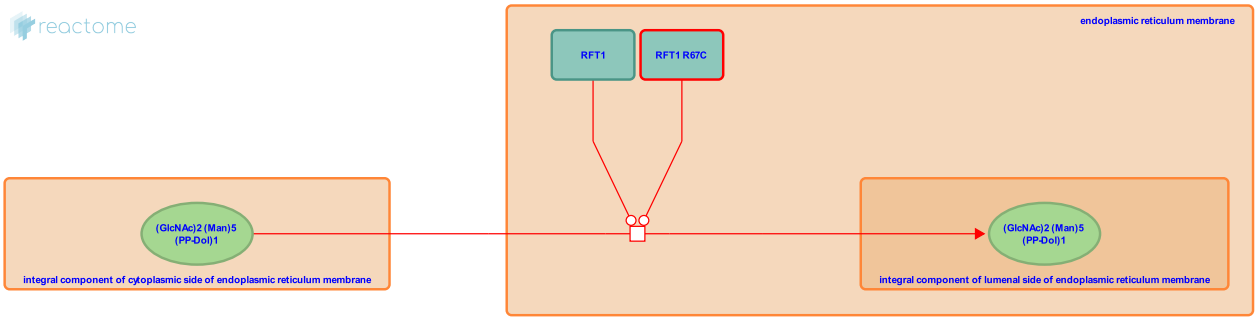
Location: Defective RFT1 causes CDG-1n

Stable identifier: R-HSA-4570573

Type: transition

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

Diseases: congenital disorder of glycosylation type I



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Literature references

Hennet, T., Haeuptle, MA., Kastaniotis, AJ., Neupert, C., Pujol, FM., Winchester, B. et al. (2008). Human RFT1 deficiency leads to a disorder of N-linked glycosylation. *Am J Hum Genet, 82*, 600-6. ↗

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