POMGNT1 transfers GlcNAc from UDP-GlcNAc to Man-O-Ser-DAG1

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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. ↗

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. ↗

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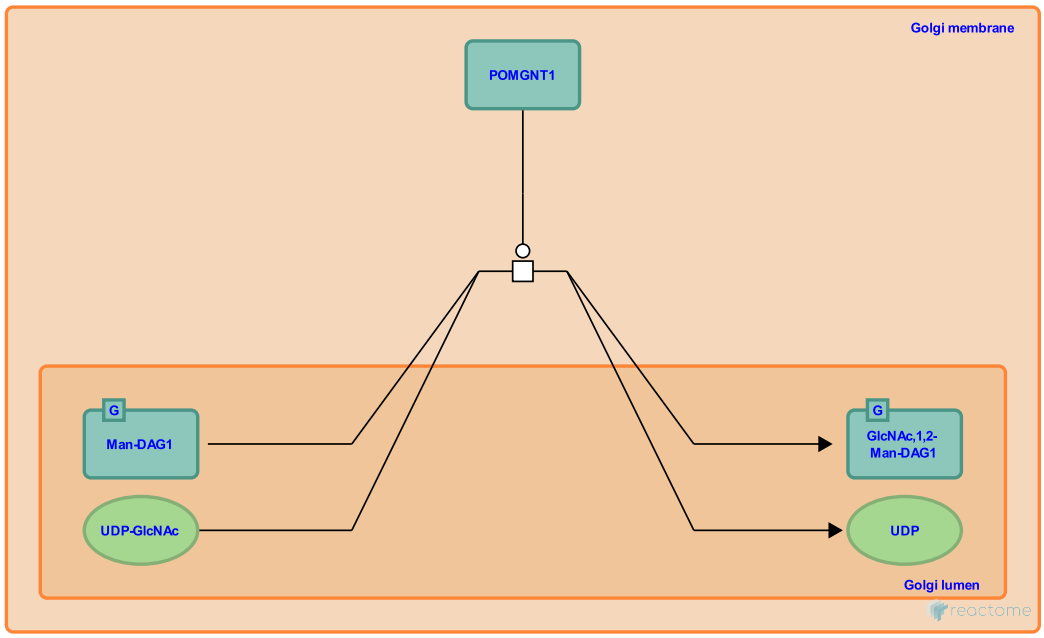
Stable identifier: R-HSA-5617037

Type: transition

Compartments: Golgi lumen, Golgi membrane



Golgi membrane resident protein O-linked-mannose beta-1,2-N-acetylglucosaminyltransferase 1 (POMGNT1; MIM:606822) mediates the transfer of N-acetylglucosaminyl (GlcNAc) residues to mannosylated proteins in the Golgi lumen. It can transfer GlcNAc to mannose-O-serine-dystroglycan (Man-DAG1) with a beta-1,2 linkage. Defects in POMGNT1 result in disrupted glycosylation of DAG1 and can cause congenital muscular dystrophy-dystroglycanopathies of varying severity (Yoshida et al. 2001).

Literature references

Inazu, T., Kano, H., Kobayashi, K., Voit, T., Endo, T., Manya, H. et al. (2001). Muscular dystrophy and neuronal migration disorder caused by mutations in a glycosyltransferase, POMGnT1. *Dev. Cell, 1*, 717-24. ↗

Editions

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