Defective SLC35A1 does not exchange CMP-Neu5Ac for CMP

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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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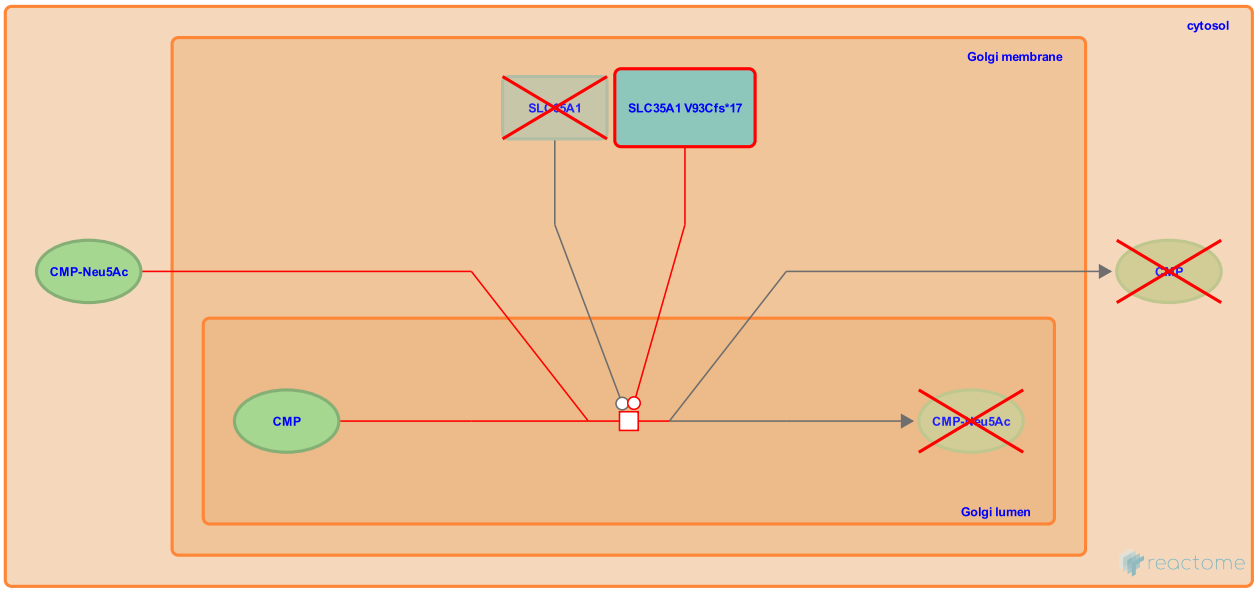
Stable identifier: R-HSA-5651942

Type: transition

Compartments: Golgi lumen, Golgi membrane, cytosol

Diseases: congenital disorder of glycosylation type II



The human gene SLC35A1 encodes the CMP-sialic acid transporter which mediates the antiport of CMP-sialic acid (CMP-Neu5Ac) into the Golgi lumen in exchange for CMP (Ishida et al. 1996). Defects in SLC35A1 are the cause of congenital disorder of glycosylation type 2F (CDG2F; MIM:603585), characterised by under-glycosylated serum proteins. CDGs are a family of severe inherited diseases caused by a defect in protein N-glycosylation. These multisystem disorders present with a wide spectrum of phenotypes such as disorders of nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders and immunodeficiency. A loss-of-function mutation causing CDG2F is V93Cfs*17 (Martinez-Duncker et al. 2005).

Literature references

Martinez-Duncker, I., Dupré, T., Piller, V., Piller, F., Candelier, JJ., Trichet, C. et al. (2005). Genetic complementation reveals a novel human congenital disorder of glycosylation of type II, due to inactivation of the Golgi CMP-sialic acid transporter. *Blood, 105*, 2671-6. ↗

Editions

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