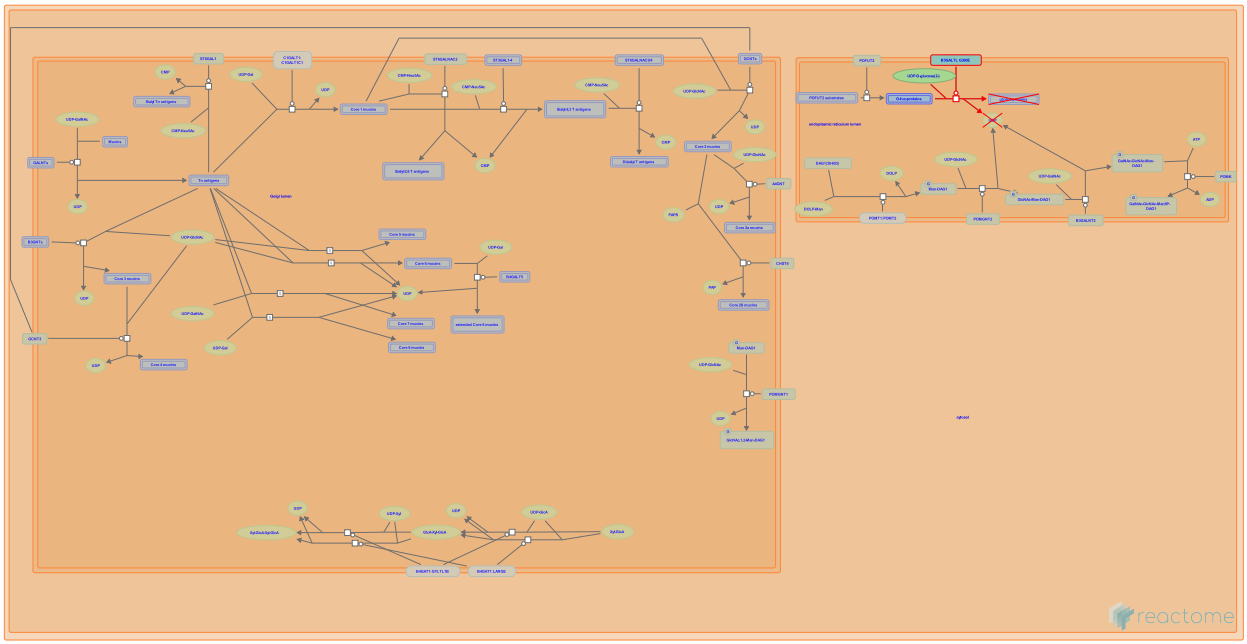
Defective B3GALTL causes PpS



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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.

04/05/2024

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.

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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. ↗

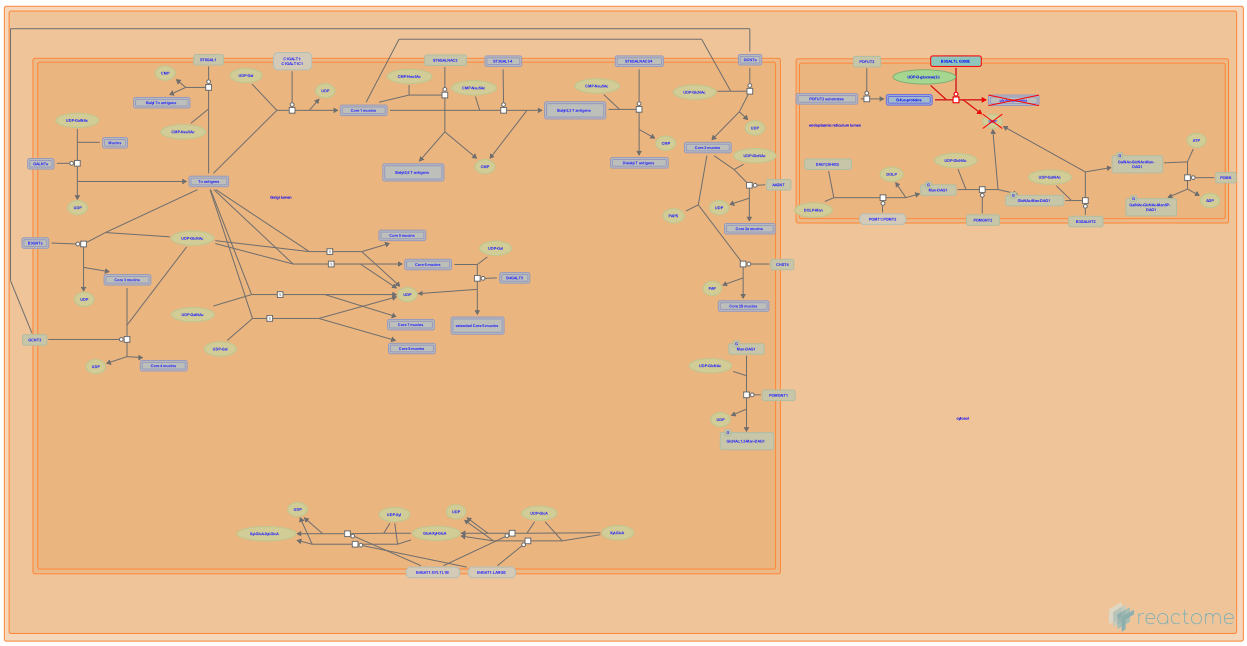
Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

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. ↗

Defective B3GALTL causes PpS ↗

Stable identifier: R-HSA-5083635

Diseases: eye disease, orofacial cleft



Human beta-1,3-glucosyltransferase like protein (B3GALTL, HGNC Approved Gene Symbol: B3GLCT; MIM:610308; CAZy family GT31), localised on the ER membrane, glucosylates O-fucosylated proteins. The resultant glc-beta-1,3-fuc disaccharide modification on thrombospondin type 1 repeat (TSR1) domain-containing proteins is thought to assist in the secretion of many of these proteins from the ER lumen, and mediate an ER quality-control mechanism of folded TSRs (Vasudevan et al. 2015). Defects in B3GALTL can cause Peters plus syndrome (PpS; MIM:261540), an autosomal recessive disorder characterised by anterior eye chamber defects, short stature, delay in growth and mental developmental and cleft lip and/or palate (Heinonen & Maki 2009).

Literature references

Vasudevan, D., Haltiwanger, RS., Johar, SS., Takeuchi, H., Majerus, E. (2015). Peters plus syndrome mutations disrupt a noncanonical ER quality-control mechanism. *Curr. Biol., 25*, 286-95. ↗

Maki, M., Heinonen, TY. (2009). Peters'-plus syndrome is a congenital disorder of glycosylation caused by a defect in the beta1,3-glucosyltransferase that modifies thrombospondin type 1 repeats. *Ann. Med., 41*, 2-10. ↗

Editions

2013-11-07	Authored, Edited	Jassal, B.
2015-12-18	Reviewed	Joshi, HJ., Hansen, L.

Defective B3GALTL does not transfer glucose to O-fucosyl-proteins ↗

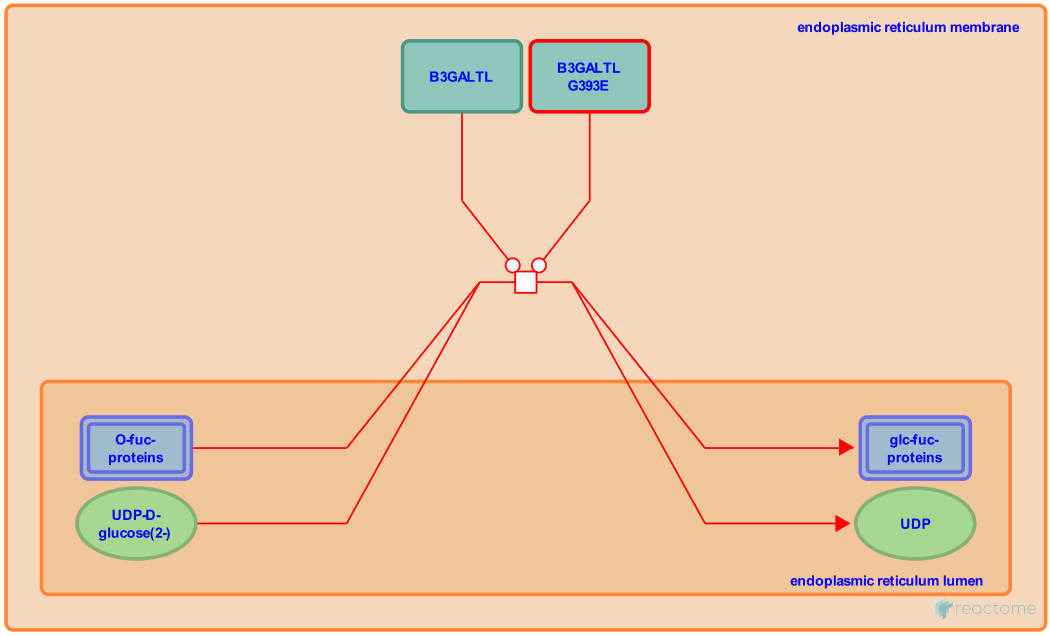
Location: Defective B3GALTL causes PpS

Stable identifier: R-HSA-6785565

Type: transition

Compartments: endoplasmic reticulum membrane, endoplasmic reticulum lumen

Diseases: eye disease, orofacial cleft



Human beta-1,3-glucosyltransferase-like protein (B3GALTL, HGNC Approved Gene Symbol: B3GLCT; MIM:610308; CAZy family GT31), localised on the ER membrane, glucosylates O-fucosylated proteins. The resultant glc-beta-1,3-fuc disaccharide modification on thrombospondin type 1 repeat (TSR1) domain-containing proteins is thought to assist in the secretion of many of these proteins from the ER lumen, and mediate an ER quality-control mechanism of folded TSRs (Vasudevan et al. 2015). Defects in B3GALTL can cause Peters plus syndrome (PpS; MIM:261540), an autosomal recessive disorder characterised by anterior eye chamber defects, short stature, delay in growth and mental developmental and cleft lip and/or palate (Heinonen & Maki 2009). More than 10 mutations in B3GALTL causing PsP are known (Weh et al. 2014) including the missense mutation G393E (Dassie Ajdid et al. 2009).

Literature references

Vigouroux, A., Poidvin, A., Burglen, L., Calvas, P., Malecaze, F., Doummar, D. et al. (2009). Novel B3GALTL mutation in Peters-plus Syndrome. *Clin. Genet., 76*, 490-2. ↗

Bick, D., Dills, SK., Murray, JC., Rhead, WJ., Reis, LM., Semina, EV. et al. (2014). Novel B3GALTL mutations in classic Peters plus syndrome and lack of mutations in a large cohort of patients with similar phenotypes. *Clin. Genet., 86*, 142-8. ↗

Vasudevan, D., Haltiwanger, RS., Johar, SS., Takeuchi, H., Majerus, E. (2015). Peters plus syndrome mutations disrupt a noncanonical ER quality-control mechanism. *Curr. Biol., 25*, 286-95. ↗

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Editions

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