# Defective GALNT12 does not transfer Gal-NAc to mucins

Hansen, L., Jassal, B., Joshi, HJ.

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

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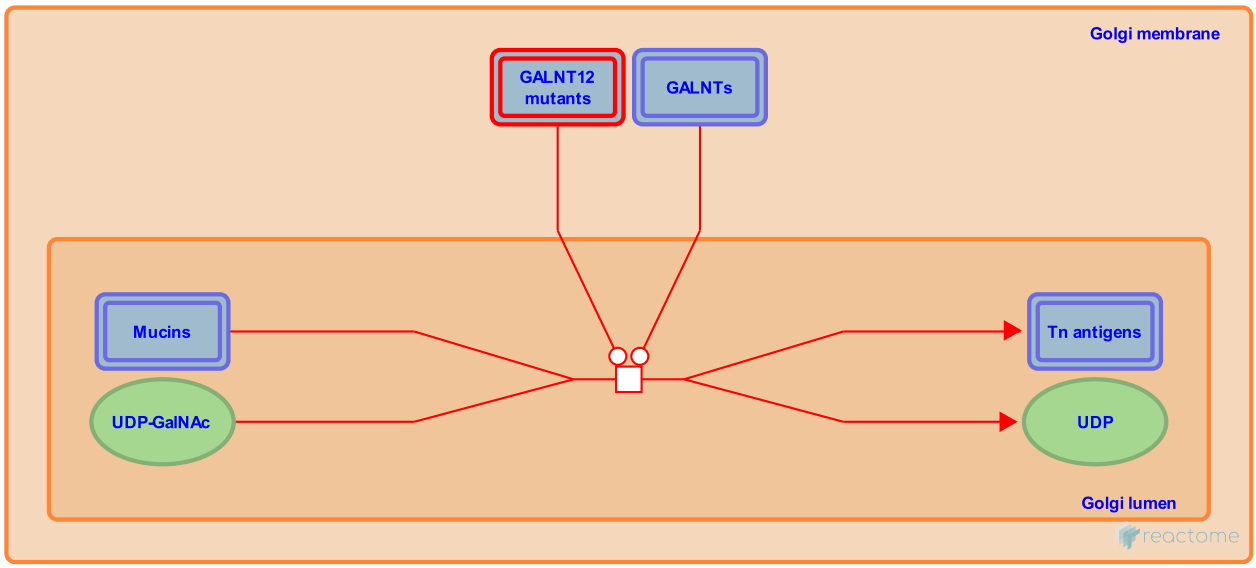
## Defective GALNT12 does not transfer GalNAc to mucins ↗

**Stable identifier:** R-HSA-5096532

**Type:** transition

**Compartments:** Golgi membrane, Golgi lumen

**Diseases:** colorectal cancer




The family of UDP-GalNAc:polypeptide N-acetylgalactosaminyltransferases (GalNAc-transferases, GALNTs) carry out the addition of N-acetylgalactosamine (GalNAc) on serine or threonine residues of proteins, especially mucins. This is the initial reaction in the formation of O-linked oligosaccharide biosynthesis (Guo et al. 2002). Defects in one of the GALNT family, GALNT12, can result in decreased glycosylation of mucins, mainly expressed in the digestive organs such as the stomach, small intestine and colon, and may play a role in colorectal cancer 1 (CRCS1; MIM:608812). CRCS1 is a complex disease characterised by malignant lesions arising from the inner walls of the colon and rectum. Mutations implicated in CRCS1 are M1I, T491M, Y395* (Guda et al. 2009), D303N and Y396C (Clarke et al. 2012).

## Literature references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## Editions

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