Defective ALG14 causes ALG14-CMS

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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.

24/12/2022
**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.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

**Literature references**


Reactome database release: 83

This document contains 1 pathway and 1 reaction (see Table of Contents)

https://reactome.org
Defective ALG14 causes ALG14-CMS

Stable identifier: R-HSA-5633231

Diseases: congenital myasthenic syndrome

UDP-N-acetylglucosamine transferase subunit ALG14 homolog (ALG14) forms a complex with ALG13 protein and is required for the addition of the second N-acetylglucosamine (GlcNAc) to the lipid linked oligosaccharide (LLO) intermediate (GlcNAcDOLDP) (Gao et al. 2005). Defects in ALG14 can cause congenital myasthenic syndrome (ALG14-CMS), which is due to a defect in neuromuscular signal transmission (Cossins et al. 2013). The most commonly affected muscles include proximal limb muscles. Mutations causing ALG14-CMS include p.P65L and p.R104* (Cossins et al. 2013).

Literature references


Editions

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Defective ALG14 does not transfer GlcNAc from UDP-GlcNAc to GlcNAcDOLP

**Location:** Defective ALG14 causes ALG14-CMS

**Stable identifier:** R-HSA-5633241

**Type:** transition

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

**Diseases:** congenital myasthenic syndrome

UDP-N-acetylglucosamine transferase subunit ALG14 homolog (ALG14) forms a complex with ALG13 protein and is required for the addition of the second N-acetylglucosamine (GlcNAc) to the lipid linked oligosaccharide (LLO) intermediate (GlcNAcDOLDP) (Gao et al. 2005). Defects in ALG14 can cause congenital myasthenic syndrome (ALG14-CMS), which is due to a defect in neuromuscular signal transmission (Cossins et al. 2013). The most commonly affected muscles include proximal limb muscles. Mutations causing ALG14-CMS include p.P65L and p.R104* (Cossins et al. 2013).

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