ALG1 gene

ALG1 chitobiosyldiphosphodolichol beta-mannosyltransferase

Normal Function

The *ALG1* gene provides instructions for making an enzyme that is involved in a process called glycosylation. During this process, complex chains of sugar molecules (oligosaccharides) are attached to proteins and fats (lipids). Glycosylation modifies proteins so they can fully perform their functions and modifies lipids so they can help cells interact with each other. Oligosaccharides are made up of many sugar molecules that are attached to one another in a stepwise process, forming a complex chain. The enzyme produced from the *ALG1* gene transfers a simple sugar called mannose to growing oligosaccharides at a particular step in the formation of the chain. Once the correct number of sugar molecules are linked together, the oligosaccharide is attached to a protein or lipid.

Health Conditions Related to Genetic Changes

ALG1-congenital disorder of glycosylation

At least 15 mutations in the *ALG1* gene have been found to cause *ALG1*-congenital disorder of glycosylation (*ALG1*-CDG). This condition typically leads to intellectual disability, delayed development, weak muscle tone (hypotonia), and other signs and symptoms that affect many body systems. Mutations in the *ALG1* gene result in the production of an abnormal enzyme with little activity. The poorly functioning enzyme cannot add mannose to sugar chains efficiently, and the resulting oligosaccharides are often incomplete. Although the short oligosaccharides can be transferred to proteins and lipids, the process is not as efficient as with the full-length oligosaccharide. The wide variety of signs and symptoms in *ALG1*-CDG are likely due to impaired glycosylation of proteins and lipids that are needed for normal function of many organs and tissues.

Other Names for This Gene

- asparagine-linked glycosylation 1 homolog (yeast, beta-1,4-mannosyltransferase)
- asparagine-linked glycosylation 1, beta-1,4-mannosyltransferase homolog
- asparagine-linked glycosylation protein 1 homolog
- beta-1,4 mannosyltransferase

- beta-1,4-mannosyltransferase
- chitobiosyldiphosphodolichol beta-mannosyltransferase
- GDP-Man:GlcNAc2-PP-dolichol mannosyltransferase
- GDP-mannose-dolichol diphosphochitobiose mannosyltransferase
- hMat-1
- HMAT1
- HMT-1
- HMT1
- mannosyltransferase-1
- Mat-1
- MT-1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALG1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=56052[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ALG1%5BTIAB%5D%29+OR+%28%28GlcNAc2-PP-dolichol+mannosyltransferase%5BTIAB%5D%29+OR+%28beta-1,4+mannosyltransferase%5BTIAB%5D%29+OR+%28beta-1,4-mannosyltransferase%5BTIAB%5D%29+OR+%28mannosyltransferase-1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ALG1 CHITOBIOSYLDIPHOSPHODOLICHOL BETA-MANNOSYLTRANSFERASE; ALG1 (https://omim.org/entry/605907)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/56052)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALG1[gene])

References

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Genomic Location

The *ALG1* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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