ALG12 gene

ALG12 alpha-1,6-mannosyltransferase

Normal Function

The *ALG12* 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. 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 *ALG12* 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

ALG12-congenital disorder of glycosylation

At least 13 mutations in the *ALG12* gene have been found to cause *ALG12*-congenital disorder of glycosylation (*ALG12*-CDG). This condition typically leads to delayed growth and development, weak muscle tone (hypotonia), and other signs and symptoms. Mutations in the *ALG12* gene result in the production of an abnormal enzyme with little activity. Without a properly functioning enzyme, mannose cannot be added to the chain 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. As a result, glycosylation is reduced. The wide variety of signs and symptoms in *ALG12*-CDG are likely due to impaired glycosylation of proteins and lipids that are needed for normal function in many organs and tissues, including the brain.

Other Names for This Gene

- asparagine-linked glycosylation 12 homolog (S. cerevisiae, alpha-1,6-mannosyltransferase)
- asparagine-linked glycosylation 12 homolog (yeast, alpha-1,6-mannosyltransferase)
- asparagine-linked glycosylation 12, alpha-1,6-mannosyltransferase homolog

- asparagine-linked glycosylation protein 12 homolog
- CDG1G
- dol-P-Man dependent alpha-1,6-mannosyltransferase
- dol-P-Man:Man(7)GlcNAc(2)-PP-Dol alpha-1,6-mannosyltransferase
- dolichyl-P-Man:Man(7)GlcNAc(2)-PP-dolichol alpha-1,6-mannosyltransferase
- dolichyl-P-Man:Man(7)GlcNAc(2)-PP-dolichyl-alpha-1,6-mannosyltransferase
- dolichyl-P-mannose:Man-7-GlcNAc-2-PP-dolichyl-alpha-6-mannosyltransferase
- ECM39
- hALG12
- mannosyltransferase ALG12 homolog
- membrane protein SB87
- PP14673

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALG12 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79087[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ALG12%5BTIAB%5D%29+OR+%28%28dol-P-Man%5BTIAB%5D%29+OR+%28hALG12%5BTIAB%5D%29+OR+%28dolichyl-P-Man%5BTIAB%5D%29+OR+%28dolichyl-P-mannose%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%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

- ALG12 ALPHA-1,6-MANNOSYLTRANSFERASE; ALG12 (https://omim.org/entry/607144)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/79087)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALG12[gene])

References

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- Grubenmann CE, Frank CG, Kjaergaard S, Berger EG, Aebi M, Hennet T. ALG12mannosyltransferase defect in congenital disorder of glycosylation type lg. HumMol Genet. 2002 Sep 15;11(19):2331-9. doi: 10.1093/hmg/11.19.2331. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12217961)
- Thiel C, Schwarz M, Hasilik M, Grieben U, Hanefeld F, Lehle L, von Figura K, Korner C. Deficiency of dolichyl-P-Man:Man7GlcNAc2-PP-dolichylmannosyltransferase causes congenital disorder of glycosylation type Ig. BiochemJ. 2002 Oct 1;367(Pt 1):195-201. doi: 10.1042/BJ20020794. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12093361) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1222867/)

Genomic Location

The *ALG12* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromosome/22/).

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