

ALG6 gene

ALG6 alpha-1,3-glucosyltransferase

Normal Function

The *ALG6* gene provides instructions for making an enzyme that is involved in a process called glycosylation. Glycosylation is a process by which sugar molecules (oligosaccharides) are attached to proteins and fats. Oligosaccharides are made up of many sugar molecules that are attached to one another in a stepwise process forming a complex chain. Glycosylation modifies proteins so they can perform a wider variety of functions. The enzyme produced from the *ALG6* gene transfers a simple sugar called glucose to the growing oligosaccharide. Once the correct number of sugar molecules are linked together, the oligosaccharide is attached to a protein or fat.

Health Conditions Related to Genetic Changes

ALG6-congenital disorder of glycosylation

At least 20 mutations in the *ALG6* gene have been found to cause *ALG6*-congenital disorder of glycosylation (*ALG6*-CDG, also known as congenital disorder of glycosylation type Ic). This condition typically leads to developmental delay, vision problems, seizures, and other signs and symptoms. Mutations in the *ALG6* gene result in the production of an abnormal enzyme with reduced or no activity. A common mutation replaces the protein building block (amino acid) alanine with the amino acid valine at position 333 in the enzyme. This mutation, written as Ala333Val or A333V, results in an enzyme with reduced activity. Without a properly functioning enzyme, glycosylation cannot proceed normally, and oligosaccharides are incomplete. As a result, glycosylation is reduced or absent. The wide variety of signs and symptoms in *ALG6*-CDG are likely due to impaired glycosylation of proteins and fats that are needed for normal function in many organs and tissues, including the brain, eyes, and hormone-producing (endocrine) system.

Other Names for This Gene

- asparagine-linked glycosylation 6 homolog (*S. cerevisiae*, alpha-1,3-glucosyltransferase)
- asparagine-linked glycosylation 6 homolog (yeast, alpha-1,3-glucosyltransferase)
- asparagine-linked glycosylation 6, alpha-1,3-glucosyltransferase homolog

- asparagine-linked glycosylation protein 6 homolog
- dol-P-Glc:Man(9)GlcNAc(2)-PP-Dol alpha-1,3-glucosyltransferase
- dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase
- dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase precursor
- dolichyl-P-Glc:Man(9)GlcNAc(2)-PP-dolichol alpha- 1->3-glucosyltransferase
- dolichyl-P-Glc:Man9GlcNAc2-PP-dolichyl glucosyltransferase
- dolichyl-P-Glc:Man9GlcNAc2-PP-dolichylglucosyltransferase
- Man(9)GlcNAc(2)-PP-Dol alpha-1,3-glucosyltransferase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALG6 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=29929\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=29929[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ALG6%5BTIAB%5D%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%29>)

Catalog of Genes and Diseases from OMIM

- ALG6 ALPHA-1,3-GLUCOSYLTRANSFERASE; ALG6 (<https://omim.org/entry/604566>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/29929>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ALG6\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ALG6[gene]))

References

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- Westphal V, Xiao M, Kwok PY, Freeze HH. Identification of a frequent variant in ALG6, the cause of Congenital Disorder of Glycosylation-Ic. Hum Mutat. 2003 Nov; 22(5):420-1. doi: 10.1002/humu.9195. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14517965>)

Genomic Location

The *ALG6* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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