



## 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

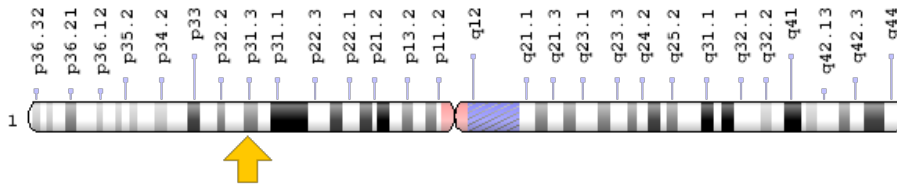
#### ALG6-congenital disorder of glycosylation

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.

## Chromosomal Location

Cytogenetic Location: 1p31.3, which is the short (p) arm of chromosome 1 at position 31.3

Molecular Location: base pairs 63,367,590 to 63,438,562 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## 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-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
- dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase
- dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase precursor
- Man(9)GlcNAc(2)-PP-Dol alpha-1,3-glucosyltransferase

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Carbohydrates Can Be Attached to Proteins to Form Glycoproteins  
<https://www.ncbi.nlm.nih.gov/books/NBK22521/>
- Sanford Burnham Medical Research Institute: Dr. Hudson Freeze Lab  
<https://www.sbpdiscovery.org/team/hudson-freeze-phd>

### Clinical Information from GeneReviews

- Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1332>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?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>

### Catalog of Genes and Diseases from OMIM

- ALG6, S. CEREVISIAE, HOMOLOG OF  
<http://omim.org/entry/604566>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ALG6.html](http://atlasgeneticsoncology.org/Genes/GC_ALG6.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ALG6%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:23157](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:23157)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:29929>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/29929>
- UniProt  
<https://www.uniprot.org/uniprot/Q9Y672>

### **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/ALG6>

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