

## B3GLCT gene

beta 3-glucosyltransferase

### Normal Function

The *B3GLCT* gene (formerly known as *B3GALT*) provides instructions for making an enzyme called beta 3-glucosyltransferase (B3Glc-T), which is involved in the complex process of adding sugar molecules to proteins (glycosylation). Glycosylation modifies proteins so they can perform a wider variety of functions. The B3Glc-T enzyme is involved in a two-step glycosylation pathway that results in the formation of a sugar structure, made up of the sugars fucose and glucose, on a specific location of several different proteins. The B3Glc-T enzyme is responsible for the second step, which adds a glucose molecule to the fucose molecule already attached to the protein. The *B3GLCT* gene is normally turned on (active) in most cells of the body, which suggests that the B3Glc-T enzyme plays an important role across many cell types.

### Health Conditions Related to Genetic Changes

#### Peters plus syndrome

At least 10 mutations that cause Peters plus syndrome have been identified in the *B3GLCT* gene. Peters plus syndrome is characterized by eye abnormalities, short stature, intellectual disability, and distinctive facial features. The most common *B3GLCT* gene mutation replaces the DNA building block (nucleotide) guanine with the nucleotide adenine near an area of the gene called exon 8 (written as 660+1G>A). This mutation disrupts how genetic information is pieced together to produce the B3Glc-T enzyme. The resulting enzyme is abnormally short and nonfunctional. It is unclear how the loss of functional B3Glc-T enzyme leads to the signs and symptoms of Peters plus syndrome, but impaired glycosylation likely disrupts the function of many proteins, which may contribute to the variety of features.

### Other Names for This Gene

- B3GALT
- B3Glc-T
- B3GLT\_HUMAN
- B3GTL
- beta 1,3-galactosyltransferase-like

- beta-3-glycosyltransferase-like
- beta3Glc-T

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of B3GLCT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=145173\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=145173[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28B3GALTL%5BTIAB%5D%29+OR+%28beta1,3-galactosyltransferase%5BTIAB%5D%29+OR+%28B3GLCT%5BTIAB%5D%29+OR+%28beta+3-galactosyltransferase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- BETA-3-GLUCOSYLTRANSFERASE; B3GLCT (<https://omim.org/entry/610308>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/145173>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=B3GLCT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=B3GLCT[gene]))

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## Genomic Location

The *B3GLCT* gene is found on chromosome 13 (<https://medlineplus.gov/genetics/chromosome/13/>).

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