

GNPTG gene

N-acetylglucosamine-1-phosphate transferase subunit gamma

Normal Function

The *GNPTG* gene provides instructions for making one part, the gamma subunit, of an enzyme called GlcNAc-1-phosphotransferase. This enzyme is made up of two alpha (α), two beta (β), and two gamma (γ) subunits. The alpha and beta subunits are produced from a different gene, called *GNPTAB*. GlcNAc-1-phosphotransferase helps prepare certain newly made enzymes for transport to lysosomes. Lysosomes are compartments within the cell that use digestive enzymes called hydrolases to break down large molecules into smaller ones that can be reused by cells.

GlcNAc-1-phosphotransferase is involved in the first step of making a molecule called mannose-6-phosphate (M6P). M6P acts as a tag that indicates a hydrolase should be transported to the lysosome. Specifically, GlcNAc-1-phosphotransferase transfers a molecule called GlcNAc-1-phosphate to a newly produced hydrolase. In the next step, a molecule is removed to reveal an M6P attached to the hydrolase. Once a hydrolase has an M6P tag, it can be transported to a lysosome.

Health Conditions Related to Genetic Changes

Mucopolidosis III gamma

At least 8 mutations in the *GNPTG* gene have been found to cause mucopolidosis III gamma. These mutations result in reduced the activity of GlcNAc-1-phosphotransferase, which disrupts tagging of hydrolases with M6P. Digestive enzymes that do not receive the M6P tag end up outside the cell, where they have increased activity. The shortage of these digestive enzymes within lysosomes causes large molecules to accumulate there. Conditions that cause molecules to build up inside lysosomes, including mucopolidosis III gamma, are called lysosomal storage disorders. The signs and symptoms of mucopolidosis III gamma are most likely due to the shortage of hydrolases inside lysosomes and the effects these enzymes have outside the cell.

Other Names for This Gene

- C16orf27
- c316G12.3

- CAB56184
- GlcNAc-phosphotransferase gamma-subunit
- GNPTAG
- GNPTG_HUMAN
- LP2537
- N-acetylglucosamine-1-phosphate transferase gamma subunit
- N-acetylglucosamine-1-phosphate transferase, gamma subunit
- N-acetylglucosamine-1-phosphotransferase, gamma subunit
- RJD9

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GNPTG ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=84572\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=84572[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28GNPTG%5BTIAB%5D%29+OR+%28GNPTAG%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

- N-ACETYLGLUCOSAMINE-1-PHOSPHOTRANSFERASE, GAMMA SUBUNIT; GNPTG (<https://omim.org/entry/607838>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/84572>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GNPTG\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GNPTG[gene]))

References

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Genomic Location

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

Last updated August 1, 2009