



AMT gene

aminomethyltransferase

Normal Function

The *AMT* gene provides instructions for making an enzyme called aminomethyltransferase. This enzyme is one of four components (subunits) that make up a large complex called glycine cleavage enzyme. Within cells, this complex is active in specialized energy-producing centers called mitochondria.

As its name suggests, glycine cleavage enzyme processes a molecule called glycine by cutting (cleaving) it into smaller pieces. Glycine is an amino acid, which is a building block of proteins. This molecule also acts as a neurotransmitter, which is a chemical messenger that transmits signals in the brain. The breakdown of excess glycine is necessary for the normal development and function of nerve cells in the brain and spinal cord.

Health Conditions Related to Genetic Changes

Glycine encephalopathy

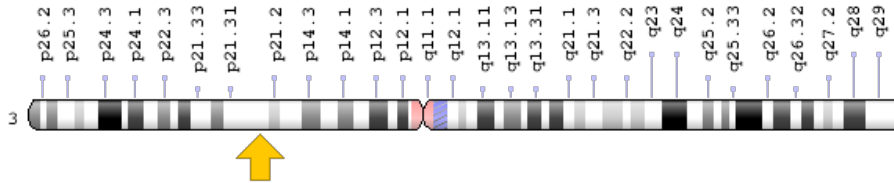
Mutations in the *AMT* gene are responsible for 10 percent to 15 percent of all cases of glycine encephalopathy. More than a dozen mutations have been identified in affected individuals. Most of these genetic changes alter single amino acids in aminomethyltransferase. Other mutations delete genetic material from the *AMT* gene or disrupt how genetic information from the gene is spliced together to make a blueprint for producing aminomethyltransferase.

AMT mutations alter the structure and function of aminomethyltransferase. When an altered version of this enzyme is incorporated into the glycine cleavage enzyme complex, it prevents the complex from breaking down glycine properly. As a result, excess glycine can build up to toxic levels in the body's organs and tissues. Damage caused by harmful amounts of this molecule in the brain and spinal cord is responsible for the intellectual disability, seizures, and breathing difficulties characteristic of glycine encephalopathy.

Chromosomal Location

Cytogenetic Location: 3p21.31, which is the short (p) arm of chromosome 3 at position 21.31

Molecular Location: base pairs 49,416,778 to 49,422,678 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GCE
- GCST
- GCST_HUMAN
- GCVT
- glycine cleavage system protein T
- Glycine Decarboxylase Complex T-Protein
- NKH

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Nonketotic hyperglycinemia is caused by deficiencies in the glycine-cleavage system
<https://www.ncbi.nlm.nih.gov/books/NBK27969/>
- Nomenclature Committee of the International Union of Biochemistry and Molecular Biology: Glycine Cleavage System
<https://www.qmul.ac.uk/sbcs/iubmb/enzyme/reaction/AminoAcid/GlyCleave.html>

Clinical Information from GeneReviews

- Nonketotic Hyperglycinemia
<https://www.ncbi.nlm.nih.gov/books/NBK1357>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28aminomethyltransferase%5BTIAB%5D%29+OR+%28%28glycine+cleavage%5BTIAB%5D%29+AND+%28T+protein+OR+protein+T%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

- AMINOMETHYLTRANSFERASE
<http://omim.org/entry/238310>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=AMT%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:473
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:275>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/275>
- UniProt
<https://www.uniprot.org/uniprot/P48728>

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