

GAMT gene

guanidinoacetate N-methyltransferase

Normal Function

The *GAMT* gene provides instructions for making the enzyme guanidinoacetate methyltransferase, which is active (expressed) mainly in the liver. This enzyme participates in the two-step production (synthesis) of the compound creatine from the protein building blocks (amino acids) glycine, arginine, and methionine. Specifically, guanidinoacetate methyltransferase controls the second step of this process. In this step, creatine is produced from another compound called guanidinoacetate. Creatine is needed for the body to store and use energy properly. It is involved in providing energy for muscle contraction, and is also important in nervous system functioning.

In addition to its role in creatine synthesis, the guanidinoacetate methyltransferase enzyme is thought to help activate a process called fatty acid oxidation. This process provides an energy source for cells during times of stress when their normal fuel, the simple sugar glucose, is scarce.

Health Conditions Related to Genetic Changes

Guanidinoacetate methyltransferase deficiency

At least 49 mutations in the *GAMT* gene cause guanidinoacetate methyltransferase deficiency, a disorder that involves intellectual disability and seizures. Most affected individuals of Portuguese ancestry have a particular mutation in which the amino acid tryptophan is replaced by the amino acid serine at position 20 in the enzyme (written as Trp20Ser or W20S).

GAMT gene mutations impair the ability of the guanidinoacetate methyltransferase enzyme to participate in creatine synthesis, resulting in a shortage of creatine. The effects of guanidinoacetate methyltransferase deficiency are most severe in organs and tissues that require large amounts of energy, especially the brain.

Other Names for This Gene

- GAMT_HUMAN
- PIG2
- TP53I2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GAMT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2593\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2593[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GAMT%5BTIAB%5D%29+OR+%28guanidinoacetate+N-methyltransferase%5BTIAB%5D%29%29+OR+%28PIG2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bm%5D+AND+%22last+3600+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- GUANIDINOACETATE METHYLTRANSFERASE; GAMT (<https://omim.org/entry/601240>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2593>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GAMT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GAMT[gene]))

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Genomic Location

The *GAMT* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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