

GATM gene

glycine amidinotransferase

Normal Function

The *GATM* gene provides instructions for making the enzyme arginine:glycine amidinotransferase. 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, arginine:glycine amidinotransferase controls the first step of the process. In this step, a compound called guanidinoacetic acid is produced by transferring a cluster of nitrogen and hydrogen atoms called a guanidino group from arginine to glycine. Guanidinoacetic acid is converted to creatine in the second step of the process. Creatine is needed for the body to store and use energy properly.

Health Conditions Related to Genetic Changes

Arginine:glycine amidinotransferase deficiency

At least two mutations in the *GATM* gene cause arginine:glycine amidinotransferase deficiency, a disorder that involves delayed development, intellectual disability, and in some cases muscle weakness. These mutations result in the production of an abnormally shortened arginine:glycine amidinotransferase enzyme or disrupt how genetic information is pieced together to make a blueprint for producing the enzyme.

GATM gene mutations interfere with the ability of the arginine:glycine amidinotransferase enzyme to participate in creatine synthesis, resulting in a shortage of creatine. The effects of arginine:glycine amidinotransferase deficiency are most severe in organs and tissues that require large amounts of energy, especially the brain.

Other Names for This Gene

- AGAT
- AT
- GATM_HUMAN
- glycine amidinotransferase (L-arginine:glycine amidinotransferase)
- glycine amidinotransferase, mitochondrial
- glycine amidinotransferase, mitochondrial precursor

- L-arginine:glycine amidinotransferase
- transamidinase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GATM ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2628\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2628[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GATM%5BTIAB%5D%29+OR+%28glycine+amidinotransferase%5BTIAB%5D%29%29+OR+%28%28AGAT%5BTIAB%5D%29+OR+%28transamidinase%5BTIAB%5D%29+OR+%28glycine+amidinotransferase%5BTIAB%5D%29%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

- L-ARGININE:GLYCINE AMIDINOTRANSFERASE; GATM (<https://omim.org/entry/602360>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2628>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GATM\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GATM[gene]))

References

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Genomic Location

The *GATM* gene is found on chromosome 15 (<https://medlineplus.gov/genetics/chromosome/15/>).

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