GARS1 gene

glycyl-tRNA synthetase 1

Normal Function

The *GARS1* gene provides instructions for making an enzyme called glycine--tRNA ligase. This enzyme is found in all cell types and plays an important role in the production of proteins. During protein production, building blocks (amino acids) are connected together in a specific order, creating a chain of amino acids. Glycine--tRNA ligase plays a role in adding the amino acid glycine at the proper place in a protein's chain of amino acids.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Distal hereditary motor neuropathy, type V

Several *GARS1* gene mutations have been identified in individuals with distal hereditary motor neuropathy, type V. This disorder affects nerve cells in the spinal cord, resulting in muscle weakness and affecting movement of the hands and feet. The *GARS1* gene mutations that cause distal hereditary motor neuropathy, type V change single amino acids used to make glycine--tRNA ligase. It is unclear how *GARS1* gene mutations lead to this disorder. The mutations probably reduce the activity of glycine--tRNA ligase. A reduction in glycine--tRNA ligase activity may impair transmission of nerve impulses.

Other Names for This Gene

- CMT2D
- DSMAV
- GARS
- glycine tRNA ligase
- GlyRS
- SMAD1
- SYG_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GARS1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2617[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GARS%5BTIAB%5D%29+OR+%28glycyl-tRNA+synthetase%5BTIAB%5D%29+NOT+%28chromosome+21%5BTIAB%5D%29%29+OR+%28%28CMT2D%5BTIAB%5D%29+OR+%28glycyl+tRNA+synthetase%29+NOT+%2812q24%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLYCYL-tRNA SYNTHETASE 1; GARS1 (https://omim.org/entry/600287)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2617)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GARS1[gene])

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Genomic Location

The *GARS1* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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