AGXT gene
alanine--glyoxylate and serine--pyruvate aminotransferase

Normal Function

The AGXT gene provides instructions for making an enzyme called serine-pyruvate aminotransferase. This enzyme is found in liver cells, specifically within cell structures called peroxisomes. These structures are important for several cellular activities, such as ridding the cell of toxic substances and helping to break down certain fats. In the peroxisome, serine-pyruvate aminotransferase converts a compound called glyoxylate to the protein building block (amino acid) glycine.

Health Conditions Related to Genetic Changes

Primary hyperoxaluria

More than 175 mutations in the AGXT gene have been found to cause primary hyperoxaluria type 1. This condition is caused by the overproduction of a substance called oxalate. Excess amounts of this substance lead to kidney and bladder stones, which can begin anytime from childhood to early adulthood with kidney disease developing at any age. Deposition of oxalate in multiple other tissues throughout the body (systemic oxalosis) can cause additional health problems.

Most of the AGXT gene mutations decrease or eliminate serine-pyruvate aminotransferase activity, which impairs the conversion of glyoxylate to glycine. Other mutations cause the enzyme to be misplaced in cells, transporting it to structures called mitochondria instead of to peroxisomes. While the enzyme in the mitochondria retains activity, it cannot access glyoxylate, which is in peroxisomes. All AGXT gene mutations result in the accumulation of glyoxylate, which is converted to oxalate instead of glycine. The oxalate is filtered through the kidneys and is either excreted in urine as a waste product or combines with calcium to form calcium oxalate, a hard compound that is the main component of kidney and bladder stones. Increased oxalate levels in the blood can lead to systemic oxalosis, particularly affecting bones and the walls of blood vessels in people with primary hyperoxaluria type 1.
Chromosomal Location

Cytogenetic Location: 2q37.3, which is the long (q) arm of chromosome 2 at position 37.3

Molecular Location: base pairs 240,868,824 to 240,880,500 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AGT
- AGT1
- AGXT1
- alanine glyoxylate aminotransferase
- alanine-glyoxylate aminotransferase
- alanine-glyoxylate aminotransferase (oxalosis I; hyperoxaluria I; glycolicaciduria; serine-pyruvate aminotransferase)
- alanine-glyoxylate transaminase
- L-alanine: glyoxylate aminotransferase 1
- pyruvate (glyoxylate) aminotransferase
- serine-pyruvate aminotransferase
- serine:pyruvate aminotransferase
- SPAT
- SPT

Additional Information & Resources

Clinical Information from GeneReviews

- Primary Hyperoxaluria Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1283
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28AGXT%5BTIAB%5D%29+OR+%28alanine-glyoxylate+aminotransferase%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ALANINE-GLYOXYLATE AMINOTRANSFERASE
  http://omim.org/entry/604285

Research Resources

- ClinVar

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:189

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P21549

Sources for This Summary


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/25644115
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4587693/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19479957

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