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

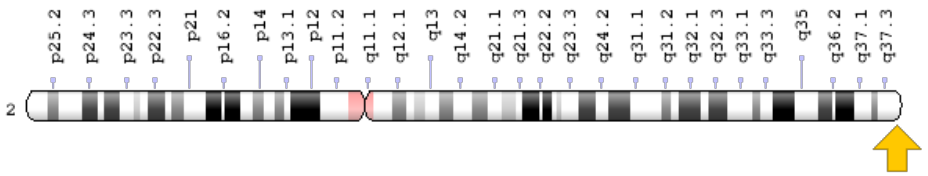
More than 175 mutations in the *AGXT* gene have been found to cause primary hyperoxaluria type 1. This condition is caused by the overproduction of a substance called oxalate. Excess amounts of this substance lead to kidney and bladder stones, which can begin anytime from childhood to early adulthood 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 240,868,745 to 240,879,119 on chromosome 2 (Homo sapiens Annotation Release 109, GRCh38.p12) (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
  https://www.ncbi.nlm.nih.gov/clinvar?term=AGXT%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:341
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:189
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/189
- UniProt
  https://www.uniprot.org/uniprot/P21549

Sources for This Summary

- Cochat P, Rumsby G. Primary hyperoxaluria. N Engl J Med. 2013 Aug 15;369(7):649-58. doi: 10.1056/NEJMra1301564. Review. Erratum in: N Engl J Med. 2013 Nov 28;369(22):2168.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23944302
- Coulter-Mackie MB, White CT, Lange D, Chew BH. Primary Hyperoxaluria Type 1. 2002 Jun 19 [updated 2014 Jul 17]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from http://www.ncbi.nlm.nih.gov/books/NBK1283/
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301460

- Hopp K, Cogal AG, Bergstralh EJ, Seide BM, Olson JB, Meek AM, Lieske JC, Milliner DS, Harris PC; Rare Kidney Stone Consortium. Phenotype-Genotype Correlations and Estimated Carrier Frequencies of Primary Hyperoxaluria. J Am Soc Nephrol. 2015 Oct;26(10):2559-70. doi: 10.1681/ASN.2014070698. Epub 2015 Feb 2.
  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/
- Williams EL, Acquaviva C, Amoroso A, Chevalier F, Coulter-Mackie M, Monico CG, Giachino D, Owen T, Robbiano A, Salido E, Waterham H, Rumsby G. Primary hyperoxaluria type 1: update and additional mutation analysis of the AGXT gene. Hum Mutat. 2009 Jun;30(6):910-7. doi: 10.1002/humu.21021. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19479957

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/AGXT

Reviewed: December 2015
Published: May 14, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services