GALT gene

galactose-1-phosphate uridylyltransferase

Normal Function

The *GALT* gene provides instructions for making an enzyme called galactose-1-phosphate uridylyltransferase. This enzyme enables the body to process a simple sugar called galactose, which is present in small amounts in many foods. Galactose is primarily part of a larger sugar called lactose, which is found in all dairy products and many baby formulas.

Galactose-1-phosphate uridylyltransferase is responsible for one step in a chemical process that breaks down galactose into other molecules that can be used by the body. Specifically, this enzyme converts a modified form of galactose (galactose-1-phosphate) to glucose, which is another simple sugar. Glucose is the main energy source for most cells. This chemical reaction also produces another form of galactose (UDP-galactose) that is used to build galactose-containing proteins and fats. These modified proteins and fats play critical roles in chemical signaling, building cellular structures, transporting molecules, and producing energy.

Health Conditions Related to Genetic Changes

Galactosemia

More than 300 mutations in the *GALT* gene have been identified in people with the classic form of galactosemia, a condition that causes life-threatening signs and symptoms beginning shortly after birth. Most of these mutations severely reduce or eliminate the activity of galactose-1-phosphate uridylyltransferase. A shortage of this enzyme prevents cells from processing galactose obtained from the diet. As a result, galactose-1-phosphate and related compounds can build up to toxic levels in the body. The accumulation of these substances damages tissues and organs, leading to the serious medical problems associated with classic galactosemia.

Most changes in the *GALT* gene alter single protein building blocks (amino acids) in galactose-1-phosphate uridylyltransferase. The most common *GALT* mutation in white Europeans and North Americans replaces the amino acid glutamine with the amino acid arginine at position 188 in the enzyme (written as Gln188Arg or Q188R). Another mutation occurs almost exclusively in people of African descent. This genetic change substitutes the amino acid leucine for the amino acid serine at position 135 (written as Ser135Leu or S135L).

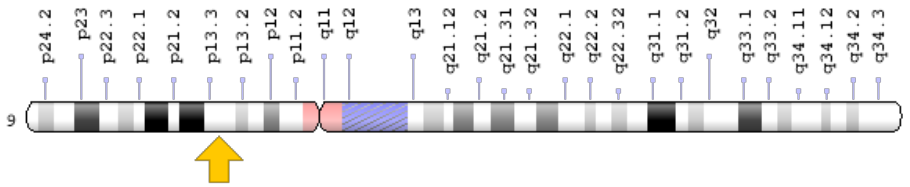
A particular *GALT* mutation called the Duarte variant results in a form of galactosemia with less serious complications than the classic type. This mutation replaces the amino acid asparagine with the amino acid aspartic acid at protein position 314

(written as Asn314Asp or N314D). The Duarte variant reduces but does not eliminate the activity of galactose-1-phosphate uridylyltransferase. The signs and symptoms associated with this variant tend to be milder because the enzyme retains 5 percent to 20 percent of its normal activity.

Chromosomal Location

Cytogenetic Location: 9p13.3, which is the short (p) arm of chromosome 9 at position 13.3

Molecular Location: base pairs 34,646,589 to 34,650,598 on chromosome 9 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Gal-1-P uridylyltransferase
- Galactosephosphate Uridylyltransferase
- GALT_HUMAN
- UDP Galactose Pyrophosphorylase
- UTP-Hexose-1-Phosphate Uridylyltransferase
- UTP:alpha-D-hexose-1-phosphate uridylyltransferase

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Figure: UDP-galactose Synthesis and Galactosemia
  https://www.ncbi.nlm.nih.gov/books/NBK1939/figure/ch42.f3/

Clinical Information from GeneReviews

- Classic Galactosemia and Clinical Variant Galactosemia
  https://www.ncbi.nlm.nih.gov/books/NBK1518
- Duarte Variant Galactosemia
  https://www.ncbi.nlm.nih.gov/books/NBK258640

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GALT%5BTI%5D%29+OR+%28galactose-1-phosphate+uridylyltransferase%5BTIAB%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

- GALACTOSE-1-PHOSPHATE URIDYLYLTRANSFERASE
  http://omim.org/entry/606999

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GALT.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GALT%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:4135
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2592
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2592
- UniProt
  https://www.uniprot.org/uniprot/P07902
- University of Utah GALT Mutation Database
  http://www.arup.utah.edu/database/GALT/GALT_welcome.php

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