MAT1A gene

methionine adenosyltransferase 1A

Normal Function

The *MAT1A* gene provides instructions for producing the enzyme methionine adenosyltransferase. The enzyme is produced from the *MAT1A* gene in two forms, designated alpha and beta. The alpha form, called a homotetramer, is made up of four identical protein subunits. The beta form, called a homodimer, is made up of two of the same protein subunits. Both forms of the enzyme are found in the liver.

Both the alpha and beta forms of methionine adenosyltransferase help break down a protein building block (amino acid) called methionine. The enzyme starts the reaction that converts methionine to S-adenosylmethionine, also called AdoMet or SAMe. AdoMet is involved in transferring methyl groups, consisting of a carbon atom and three hydrogen atoms, to other compounds, a process called transmethylation. Transmethylation is important in many cellular processes. These include determining whether the instructions in a particular segment of DNA are carried out, regulating reactions involving proteins and lipids, and controlling the processing of chemicals that relay signals in the nervous system (neurotransmitters).

Health Conditions Related to Genetic Changes

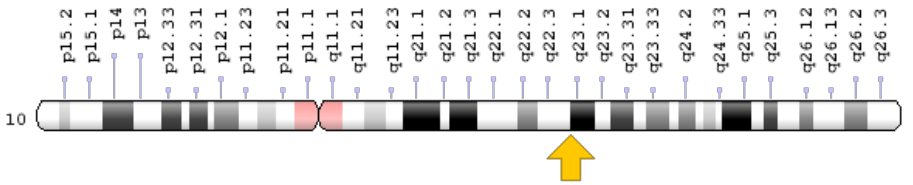
Hypermethioninemia

Approximately 15 mutations in the *MAT1A* gene have been found to reduce the activity of the methionine adenosyltransferase enzyme. Most of these mutations substitute one amino acid for another amino acid in the enzyme, causing it to process methionine less efficiently. Other mutations introduce a premature stop signal in the instructions for making the methionine adenosyltransferase enzyme. As a result, a shortened, nonfunctional enzyme is produced. A reduction in methionine adenosyltransferase function results in a buildup of methionine in the body and less efficient AdoMet production, and in severe cases can cause neurological problems.

Chromosomal Location

Cytogenetic Location: 10q22.3, which is the long (q) arm of chromosome 10 at position 22.3

Molecular Location: base pairs 80,271,820 to 80,289,658 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MAT
- MATA1
- methionine adenosyltransferase I, alpha
- METK1_HUMAN
- S-adenosylmethionine synthetase 1
- SAMS
- SAMS, liver-specific
- SAMS1

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Methionine Metabolism https://www.ncbi.nlm.nih.gov/books/NBK22453/?rendertype=figure&id=A3252

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MAT1A%5BALL%5D%29+OR+%28methionine+adenosyltransferase%5BALL%5D%29%29+OR+%28%28SAMS%5BTIAB%5D%29+OR+%28MATA1%5BTIAB%5D%29+OR+%28SAMS1%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+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- METHIONINE ADENOSYLTRANSFERASE I, ALPHA
  http://omim.org/entry/610550

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MAT1A.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MAT1A%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6903
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4143
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4143
- UniProt
  https://www.uniprot.org/uniprot/Q00266

Sources for This Summary

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