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 MATI and MATII. MATI is made up of four identical protein subunits, which is known as a homotetramer. MATII is made up of two of the same protein subunits, which is known as a homodimer. Both forms of the enzyme are found in the liver.

Both the MATI and MATII 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 processes 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

More than 70 variants (also called mutations) in the *MAT1A* gene have been found in individuals with hypermethioninemia, a condition characterized by an excess of methionine in the body. Most of these variants substitute one amino acid for another amino acid in the methionine adenosyltransferase enzyme, causing it to process methionine less efficiently. Other variants introduce a premature stop signal in the instructions for making the enzyme. As a result, a shortened, nonfunctional enzyme is produced. These alterations reduce the activity of the methionine adenosyltransferase enzyme. A reduction in the enzyme's function results in a buildup of methionine in the body and less efficient AdoMet production, which in severe cases, can cause neurological problems.

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

Tests Listed in the Genetic Testing Registry

- Tests of MAT1A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4143[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; MAT1A (https://omim.org/entry/610550)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4143)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MAT1A[gene])

References

- Biochemistry (fifth edition, 2002): Methionine Metabolism (https://www.ncbi.nlm.nih.gov/books/NBK22453/?rendertype=figure&id=A3252)
- Chamberlin ME, Ubagai T, Mudd SH, Thomas J, Pao VY, Nguyen TK, Levy HL, GreeneC, Freehauf C, Chou JY. Methionine adenosyltransferase I/III deficiency: novelmutations and clinical variations. Am J Hum Genet. 2000 Feb;66(2):347-55. doi: 10.1086/302752. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10677294)

or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1288087/)

- Chien YH, Chiang SC, Huang A, Hwu WL. Spectrum of hypermethioninemia inneonatal screening. Early Hum Dev. 2005 Jun;81(6):529-33. doi:10.1016/j.earlhumdev.2004.11.005. Epub 2004 Dec 19. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15935930)
- Chou JY. Molecular genetics of hepatic methionine adenosyltransferasedeficiency. Pharmacol Ther. 2000 Jan;85(1):1-9. doi:10.1016/s0163-7258(99)00047-9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/10674710)
- Smythies JR, Gottfries CG, Regland B. Disturbances of one-carbon metabolism inneuropsychiatric disorders: a review. Biol Psychiatry. 1997 Jan 15;41(2):230-3.doi:10.1016/S0006-3223(96)00068-6. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9018395)

Genomic Location

The *MAT1A* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

Last updated August 6, 2021