MTRR gene

5-methyltetrahydrofolate-homocysteine methyltransferase reductase

Normal Function

The *MTRR* gene provides instructions for making an enzyme called methionine synthase reductase. This enzyme is required for the proper function of another enzyme called methionine synthase. Methionine synthase helps process amino acids, which are the building blocks of proteins. Specifically, it converts the amino acid homocysteine to another amino acid called methionine. After a period of being turned on (active), methionine synthase turns off (becomes inactive). Methionine synthase reductase reactivates methionine synthase so the enzyme can continue to produce methionine.

Health Conditions Related to Genetic Changes

Homocystinuria

At least 20 mutations in the *MTRR* gene have been identified in people with homocystinuria. Some of these mutations change single amino acids in methionine synthase reductase. Other mutations lead to an abnormally small, nonfunctional version of the enzyme. All these mutations prevent the enzyme from functioning normally. Without methionine synthase reductase, methionine synthase cannot convert homocysteine to methionine. As a result, homocysteine builds up in the bloodstream, and the amount of methionine is reduced. Some of the excess homocysteine is excreted in urine. Researchers have not determined how altered levels of homocysteine and methionine lead to the health problems associated with homocystinuria.

Other disorders

A specific version (variant) of the *MTRR* gene may be associated with an increased risk of various health problems before birth. The variant (written as A66G) replaces a building block of DNA (nucleotide) called adenine with the nucleotide guanine at a specific location in the *MTRR* gene. This variant is associated with birth defects that occur during the development of the brain and spinal cord (neural tube defects). This variant may also increase the risk of having a child with Down syndrome, a condition characterized by intellectual disability and associated health problems. Researchers have not determined why there may be a connection between the A66G variant of the *MTRR* gene and the risk of neural tube defects or Down syndrome. Many factors play a part in determining the risk of these disorders.

Other Names for This Gene

- cblE
- methionine synthase reductase
- MSR
- MTRR_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MTRR (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4552[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MTRR%5BTIAB%5D%29+OR+%285-methyltetrahydrofolate-homocysteine+methyltransferase+reductase%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

- DOWN SYNDROME (https://omim.org/entry/190685)
- NEURAL TUBE DEFECTS, SUSCEPTIBILITY TO; NTD (https://omim.org/entry/182940)
- METHIONINE SYNTHASE REDUCTASE; MTRR (https://omim.org/entry/602568)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4552)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MTRR[gene])

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Genomic Location

The *MTRR* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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