AHCY gene

adenosylhomocysteinase

Normal Function

The *AHCY* gene provides instructions for producing the enzyme S-adenosylhomocysteine hydrolase. This enzyme is involved in a multistep process that breaks down the protein building block (amino acid) methionine. Specifically, S-adenosylhomocysteine hydrolase controls the step that converts the compound S-adenosylhomocysteine to the compounds adenosine and homocysteine. This reaction also plays an important role in regulating the addition of methyl groups, consisting of one carbon atom and three hydrogen atoms, to other compounds (methylation). Methylation 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

More than 10 variants (also known as mutations) in the *AHCY* gene have been described in people with hypermethioninemia. Most of these variants substitute one amino acid for another amino acid in the S-adenosylhomocysteine hydrolase enzyme, causing it to process methionine less efficiently. Other variants introduce a premature stop signal in the instructions for making the S-adenosylhomocysteine hydrolase enzyme. As a result, a shortened, nonfunctional enzyme is produced. These changes reduce the activity of the S-adenosylhomocysteine hydrolase enzyme, impairing the breakdown of methionine. As a result, methionine builds up in the body, which is known as hypermethioninemia. In some cases, excess methionine can cause intellectual disability or other neurological problems in affected individuals.

Other Names for This Gene

- SAHH
- SAHH_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AHCY (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=191[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AHCY%5BTIAB%5D%29+OR+%28S-adenosylhomocysteine+hydrolase%5BTIAB%5D%29%29+OR+%28%28SAHH%5BTIAB%5D%29+OR+%28adenosylhomocysteinase%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- S-ADENOSYLHOMOCYSTEINE HYDROLASE; AHCY (https://omim.org/entry/180960)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/191)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AHCY[gene])

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Genomic Location

The *AHCY* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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