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

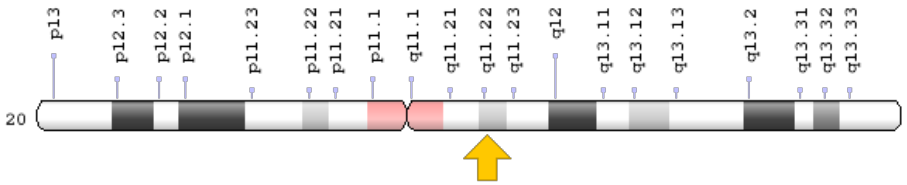
Hypermethioninemia

Three mutations in the *AHCY* gene have been described in people with hypermethioninemia. In a Croatian family, one mutation substitutes the amino acid cysteine for the amino acid tyrosine at protein position 143 (written as Tyr143Cys or Y143C). Another mutation replaces the amino acid tryptophan with a premature stop signal at position 112 (written as Trp112X or W112X), resulting in an enzyme that is abnormally short. A U.S. patient was found to have, in addition to the Y143C mutation, a mutation that substitutes the amino acid valine for the amino acid alanine at position 89 (written as Ala89Val or A89V). These mutations reduce the activity of the S-adenosylhomocysteine hydrolase enzyme, resulting in the signs and symptoms of hypermethioninemia.

Chromosomal Location

Cytogenetic Location: 20q11.22, which is the long (q) arm of chromosome 20 at position 11.22

Molecular Location: base pairs 34,235,012 to 34,311,976 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- SAHH
- SAHH_HUMAN

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%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
  http://omim.org/entry/180960

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_AHCY.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=AHCY%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:343
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:191
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/191
- UniProt
  https://www.uniprot.org/uniprot/P23526

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