CA5A gene

carbonic anhydrase 5A

Normal Function

The *CA5A* gene provides instructions for making an enzyme called carbonic anhydrase VA. This enzyme helps convert carbon dioxide to a substance called bicarbonate. Bicarbonate is necessary to maintain the proper acid-base balance in the body, which is necessary for most biological reactions to proceed properly.

The carbonic anhydrase VA enzyme is particularly important in the liver, where it provides bicarbonate needed by four enzymes in the energy-producing centers of cells (mitochondria): carbomoyl phosphate synthetase-1, pyruvate carboxylase, propionyl-CoA carboxylase, and 3-methylcrotonyl-CoA carboxylase. These enzymes help control the amount of certain other substances in the body. Carbomoyl phosphate synthetase-1 is involved in the urea cycle, which processes excess nitrogen and prevents it from accumulating as ammonia, a substance that is toxic to the brain. Pyruvate carboxylase is involved in the production of the simple sugar glucose (gluconeogenesis) in the liver. Propionyl-CoA carboxylase and 3-methylcrotonyl-CoA carboxylase help break down certain protein building blocks (amino acids).

Health Conditions Related to Genetic Changes

Carbonic anhydrase VA deficiency

At least three *CA5A* gene mutations have been identified in people with carbonic anhydrase VA deficiency. This inherited disorder is characterized by potentially life-threatening episodes of poor feeding, vomiting, weight loss, tiredness (lethargy), rapid breathing (tachypnea), seizures, or coma. The risk of these episodes is thought to decline after childhood.

Mutations in the *CA5A* gene result in absent or impaired carbonic anhydrase VA enzyme function, leading to reduced bicarbonate production. Insufficient bicarbonate results in impaired control of acid-base balance and reduces the activity of the four affected mitochondrial enzymes, leading to various biochemical abnormalities that are associated with carbonic anhydrase VA deficiency and that cause the episodes that occur in this disorder. Studies suggest that a related enzyme produced from the *CA5B* gene may increasingly compensate for the lack of carbonic anhydrase VA as affected individuals mature, which may result in a reduced risk of disease episodes after

childhood.

Other Names for This Gene

- CA-VA
- CA5
- CA5AD
- carbonate dehydratase VA
- carbonic anhydrase 5A, mitochondrial precursor
- carbonic anhydrase V, mitochondrial
- carbonic anhydrase VA, mitochondrial
- carbonic dehydratase
- CAV
- CAVA
- GS1-21A4.1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CA5A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=763[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CA5A%5BTIAB%5D%29+OR+%28carbonic+anhydrase+5A%5BTIAB%5D%29%29+OR+%28%28CA-VA%5BTIAB%5D%29+OR+%28carbonate+dehydratase+VA%5BTIAB%5D%29+OR+%28carbonic+anhydrase+5A,+mitochondrial+precursor%5BTIAB%5D%29+OR+%28carbonic+anhydrase+V,+mitochondrial%5BTIAB%5D%29+OR+%28carbonic+anhydrase+VA,+mitochondrial%5BTIAB%5D%29+OR+%28carbonic+dehydratase%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)

Catalog of Genes and Diseases from OMIM

- CARBONIC ANHYDRASE VA; CA5A (https://omim.org/entry/114761)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/763)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CA5A[gene])

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Genomic Location

The *CA5A* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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