ALDH7A1 gene

aldehyde dehydrogenase 7 family member A1

Normal Function

The *ALDH7A1* gene is a member of the aldehyde dehydrogenase (ALDH) gene family. These genes provide instructions for producing enzymes that alter molecules called aldehydes. The *ALDH7A1* gene provides instructions for making an enzyme called α-aminoadipic semialdehyde (α-AASA) dehydrogenase, also known as antiquitin. Within the cell, antiquitin is found in the internal fluid of the cell (cytosol) and in the nucleus. This enzyme is involved in the breakdown of the protein building block (amino acid) lysine in the brain. In one step in the breakdown of lysine to other molecules, antiquitin facilitates the conversion of α-aminoadipic semialdehyde to α-aminoadipate. The breakdown of lysine in the brain is necessary for energy production and to produce other needed molecules.

Health Conditions Related to Genetic Changes

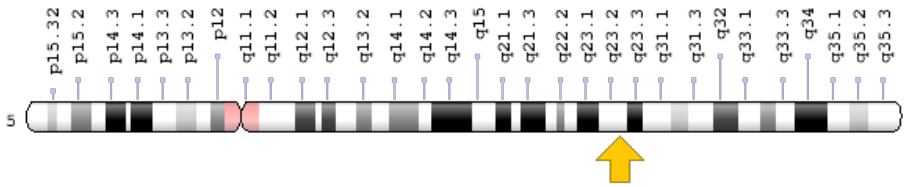
Pyridoxine-dependent epilepsy

A variety of mutations in the *ALDH7A1* gene have been found to cause pyridoxine-dependent epilepsy. Most of these mutations are specific to single families. One mutation occurs in multiple people with this condition; it replaces the amino acid glutamine with the amino acid glycine at position 399 in the antiquitin protein (written as Glu399Gln or E399Q). All mutations that cause pyridoxine-dependent epilepsy produce a nonfunctional antiquitin protein. A shortage (deficiency) of antiquitin leads to the buildup of α-aminoadipic semialdehyde, resulting in a disruption in the activity of pyridoxine, a form of vitamin B6 derived from food. Pyridoxine plays a role many processes in the body, such as the breakdown of amino acids and chemicals in the brain called neurotransmitters. It is unclear how a lack of pyridoxine causes the seizures characteristic of this condition.

Chromosomal Location

Cytogenetic Location: 5q23.2, which is the long (q) arm of chromosome 5 at position 23.2

Molecular Location: base pairs 126,541,841 to 126,595,219 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AL7A1_HUMAN
- aldehyde dehydrogenase 7 family, member A1
- aldehyde dehydrogenase 7A1
- antiquitin
- antiquitin 1
- ATQ1
- EPD
- PDE

Additional Information & Resources

Clinical Information from GeneReviews

- Pyridoxine-Dependent Epilepsy
  https://www.ncbi.nlm.nih.gov/books/NBK1486

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ALDH7A1%5BTIAB%5D%29+OR+%28antiquitin%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

- ALDEHYDE DEHYDROGENASE 7 FAMILY, MEMBER A1
  http://omim.org/entry/107323

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALDH7A1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH7A1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:877
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:501
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/501
- UniProt
  https://www.uniprot.org/uniprot/P49419

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