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.

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

Tests Listed in the Genetic Testing Registry

- Tests of ALDH7A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=501[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; ALDH7A1 (https://omim.org/entry/107323)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/501)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH7A1[gene])

References

- Bok LA, Struys E, Willemsen MA, Been JV, Jakobs C. Pyridoxine-dependentseizures in Dutch patients: diagnosis by elevated urinary alpha-aminoadipicsemialdehyde levels. Arch Dis Child. 2007 Aug;92(8):687-9. doi:10.1136/adc.2006.103192. Epub 2006 Nov 6. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17088338) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2083882/)
- Fong WP, Cheng CH, Tang WK. Antiquitin, a relatively unexplored member in thesuperfamily of aldehyde dehydrogenases with diversified physiological functions. Cell Mol Life Sci. 2006 Dec;63(24):2881-5. doi: 10.1007/s00018-006-6089-4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17131062)
- Kanno J, Kure S, Narisawa A, Kamada F, Takayanagi M, Yamamoto K, Hoshino H, Goto T, Takahashi T, Haginoya K, Tsuchiya S, Baumeister FA, Hasegawa Y, Aoki Y,

Yamaguchi S, Matsubara Y. Allelic and non-allelic heterogeneities in pyridoxinedependent seizures revealed by ALDH7A1 mutational analysis. Mol Genet Metab. 2007Aug;91(4):384-9. doi: 10.1016/j.ymgme.2007.02.010. Epub 2007 Apr 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17433748)

- Mills PB, Struys E, Jakobs C, Plecko B, Baxter P, Baumgartner M, Willemsen MA, Omran H, Tacke U, Uhlenberg B, Weschke B, Clayton PT. Mutations in antiquitin inindividuals with pyridoxine-dependent seizures. Nat Med. 2006 Mar;12(3):307-9. doi: 10.1038/nm1366. Epub 2006 Feb 19. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16491085)
- Pearl PL, Taylor JL, Trzcinski S, Sokohl A. The pediatric neurotransmitterdisorders. J Child Neurol. 2007 May;22(5):606-16. doi: 10.1177/0883073807302619. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17690069)
- Plecko B, Paul K, Paschke E, Stoeckler-Ipsiroglu S, Struys E, Jakobs C,Hartmann H, Luecke T, di Capua M, Korenke C, Hikel C, Reutershahn E, FreilingerM, Baumeister F, Bosch F, Erwa W. Biochemical and molecular characterization of18 patients with pyridoxine-dependent epilepsy and mutations of the antiquitin(ALDH7A1) gene. Hum Mutat. 2007 Jan;28(1):19-26. doi: 10.1002/humu.20433. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17068770)

Genomic Location

The *ALDH7A1* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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