ALDH3A2 gene

aldehyde dehydrogenase 3 family member A2

Normal Function

The *ALDH3A2* gene is a member of the aldehyde dehydrogenase (ALDH) gene family. Genes in this family provide instructions for producing enzymes that alter molecules called aldehydes. The *ALDH3A2* gene provides instructions for making an enzyme called fatty aldehyde dehydrogenase (FALDH). This enzyme is involved in the breakdown of fats, specifically the breakdown of molecules called fatty aldehydes to fatty acids. This conversion of molecules is part of a multistep process called fatty acid oxidation in which fats are broken down and converted into energy.

The FALDH enzyme is found in most tissues, but its activity (expression) is highest in the liver. Within cells, the FALDH enzyme is located in the endoplasmic reticulum, a structure involved in protein processing and transport.

Health Conditions Related to Genetic Changes

Sjögren-Larsson syndrome

At least 80 mutations in the *ALDH3A2* gene have been found to cause Sjögren-Larsson syndrome, a condition characterized by dry, scaly skin (ichthyosis); neurological abnormalities; and eye problems. Many of these mutations change single protein building blocks (amino acids) in the FALDH enzyme. The gene mutations that cause Sjögren-Larsson syndrome lead to the production of a FALDH enzyme that is unable to break down fatty aldehyde molecules. As a result, fats that are not broken down can build up in cells. In all affected tissues, excess fat accumulation interferes with the normal formation of protective membranes or materials that are necessary for the body to function normally. These abnormalities underlie the characteristic signs and symptoms of Sjögren-Larsson syndrome.

Other Names for This Gene

- AL3A2_HUMAN
- aldehyde dehydrogenase 10
- aldehyde dehydrogenase 3 family, member A2
- aldehyde dehydrogenase family 3 member A2

- ALDH10
- FALDH
- fatty aldehyde dehydrogenase
- microsomal aldehyde dehydrogenase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALDH3A2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=224[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ALDH3A2%5BTIAB%5D%29+OR+%28FALDH%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ALDEHYDE DEHYDROGENASE, FAMILY 3, SUBFAMILY A, MEMBER 2; ALDH3A2 (https://omim.org/entry/609523)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/224)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH3A2[gene])

References

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- Rizzo WB, S'Aulis D, Jennings MA, Crumrine DA, Williams ML, Elias PM. Ichthyosis in Sjogren-Larsson syndrome reflects defective barrier function due toabnormal lamellar body structure and secretion. Arch Dermatol Res. 2010Aug;302(6):443-51. doi: 10.1007/s00403-009-1022-y. Epub 2010 Jan 5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20049467) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2892059/)
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biochemicalpathogenesis of fatty aldehyde dehydrogenase deficiency. Mol Genet Metab. 2007Ja
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Genomic Location

The *ALDH3A2* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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