ALDH5A1 gene

aldehyde dehydrogenase 5 family member A1

Normal Function

The *ALDH5A1* gene provides instructions for producing the succinic semialdehyde dehydrogenase enzyme. This enzyme is found in the energy-producing centers of cells (mitochondria). Succinic semialdehyde dehydrogenase is involved in the breakdown of a chemical that transmits signals in the brain (neurotransmitter) called gamma-amino butyric acid (GABA). The primary role of GABA is to prevent the brain from being overloaded with too many signals. Once GABA molecules have been released from nerve cells, they are broken down by succinic semialdehyde dehydrogenase and other enzymes.

Health Conditions Related to Genetic Changes

Succinic semialdehyde dehydrogenase deficiency

Variants (also called mutations) in the *ALDH5A1* gene have been found to cause succinic semialdehyde dehydrogenase deficiency. This is a disorder that can cause a variety of neurological problems. Most of the variants that cause this condition lead to the production of an enzyme with little or no activity. A lack of functional succinic semialdehyde dehydrogenase disrupts the conversion of succinic semialdehyde to succinic acid. Instead, succinic semialdehyde is converted back into GABA or a related molecule, gamma-hydroxybutyrate (GHB). It is unclear how increases in GHB and GABA cause developmental delays, seizures, and other features of succinic semialdehyde dehydrogenase deficiency.

Research has shown that people with *ALDH5A1* gene variants that result in a lack of functional succinic semialdehyde dehydrogenase tend to have more severe signs and symptoms than people who have gene variants that impair protein activity.

Other Names for This Gene

- aldehyde dehydrogenase 5 family, member A1
- aldehyde dehydrogenase 5 family, member A1 (succinate-semialdehyde dehydrogenase)
- aldehyde dehydrogenase 5A1

- mitochondrial succinate semialdehyde dehydrogenase
- NAD(+)-dependent succinic semialdehyde dehydrogenase
- SSADH
- SSDH
- SSDH_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ALDH5A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7915[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ALDH5A1%5BTIAB%5D%29+OR+%28%28SSDH%5BTIAB%5D%29+OR+%28SSADH%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ALDEHYDE DEHYDROGENASE 5 FAMILY, MEMBER A1; ALDH5A1 (https://omim.org/entry/610045)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7915)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH5A1[gene])

References

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Genomic Location

The *ALDH5A1* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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