

## ALDH4A1 gene

aldehyde dehydrogenase 4 family member A1

### Normal Function

The *ALDH4A1* gene provides instructions for producing the enzyme pyrroline-5-carboxylate dehydrogenase, which is found in tissues throughout the body. Within the cells of these tissues, this enzyme functions in energy-producing structures called mitochondria.

Pyrroline-5-carboxylate dehydrogenase starts the second step in the process that breaks down the protein building block (amino acid) proline. This step converts pyrroline-5-carboxylate, which is produced in the first step, to the amino acid glutamate.

The conversion of proline to glutamate (and the conversion of glutamate to proline, which is controlled by different enzymes) is important for maintaining a supply of amino acids needed for protein production, and for energy transfer within the cell.

### Health Conditions Related to Genetic Changes

#### Hyperprolinemia

At least seven variants (also known as mutations) in the *ALDH4A1* gene have been found to cause hyperprolinemia type II. Hyperprolinemia is an excess of proline in the blood. Type II is generally the most severe form of the disorder and is characterized by neurological problems such as seizures or intellectual disability.

*ALDH4A1* gene variants reduce or eliminate the function of the pyrroline-5-carboxylate dehydrogenase enzyme. A lack of pyrroline-5-carboxylate dehydrogenase function leads to decreased breakdown of proline and elevated levels of proline and intermediate breakdown product pyrroline-5-carboxylate, causing the signs and symptoms of hyperprolinemia type II.

### Other Names for This Gene

- AL4H1\_HUMAN
- aldehyde dehydrogenase 4 family, member A1
- aldehyde dehydrogenase 4A1
- ALDH4

- mitochondrial delta-1-pyrroline 5-carboxylate dehydrogenase
- P5C dehydrogenase
- P5CD
- P5CDh
- P5CDhL
- P5CDhS

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of ALDH4A1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8659\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8659[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ALDH4A1%5BTIAB%5D%29+OR+%28%28P5CD%5BTIAB%5D%29+OR+%28ALDH4%5BTIAB%5D%29+OR+%28P5CDh%5BTIAB%5D%29+OR+%28P5CDhS%5BTIAB%5D%29+OR+%28P5C+dehydrogenase%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+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- ALDEHYDE DEHYDROGENASE, FAMILY 4, SUBFAMILY A, MEMBER 1; ALDH4A1 (<https://omim.org/entry/606811>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8659>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH4A1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH4A1[gene]))

## **References**

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## Genomic Location

The *ALDH4A1* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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