PRODH gene

proline dehydrogenase 1

Normal Function

The *PRODH* gene provides instructions for producing the enzyme proline dehydrogenase (also known as proline oxidase), which is found primarily in the brain, lung, and muscle. Within cells of these organs, this enzyme functions in energy-producing structures called mitochondria.

Proline dehydrogenase plays a role in the process of breaking down the protein building block (amino acid) proline. Specifically, the enzyme starts the reaction that converts proline to pyrroline-5-carboxylate. A subsequent step converts this intermediate product 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

More than 20 variants (also known as mutations) in the *PRODH* gene have been found to cause hyperprolinemia type I. Hyperprolinemia is an excess of proline in the blood. Type I is generally characterized by a lack of symptoms, although some severely affected individual have neurological problems such as seizures or intellectual disability. *PRODH* gene variants change single amino acids in the enzyme and reduce the activity of the proline dehydrogenase enzyme, causing a decrease in its ability to break down proline. A decrease in proline breakdown results in a buildup of proline in the body, and leads to hyperprolinemia.

Other disorders

Several studies have shown an association between variations in the *PRODH* gene and psychiatric disorders such as schizophrenia, while others have shown no significant association. Most of the variations in the *PRODH* gene result in the substitution of one amino acid for another in the proline dehydrogenase enzyme. The amino acid

substitution reduces the enzyme's activity, resulting in decreased breakdown of proline and a buildup of proline in the body. Researchers believe that hyperprolinemia (described above) may affect the action of certain chemicals that transmit signals between nerve cells (neurons) in the brain (neurotransmitters), resulting in an increased risk of psychiatric disorders.

Other Names for This Gene

- FLJ33744
- HSPOX2
- MGC148078
- MGC148079
- p53 induced protein
- PIG6
- PROD_HUMAN
- PRODH1
- PRODH2
- proline dehydrogenase (oxidase) 1
- proline dehydrogenase (proline oxidase)
- proline oxidase 2
- Proline oxidase, mitochondrial
- SCZD4
- TP53I6
- tumor protein p53 inducible protein 6

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PRODH (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5625[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PRODH%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PROLINE DEHYDROGENASE 1; PRODH (https://omim.org/entry/606810)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5625)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PRODH[gene])

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Genomic Location

The *PRODH* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromosome/22/).

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