PDHB gene

pyruvate dehydrogenase E1 subunit beta

Normal Function

The *PDHB* gene provides instructions for making a protein called E1 beta. E1 beta is a piece (a subunit) of a larger protein: two E1 beta proteins combine with two copies of another protein, called E1 alpha (produced from the *PDHA1* gene), to form the E1 enzyme. This enzyme, also known as pyruvate dehydrogenase, is a component of a group of proteins called the pyruvate dehydrogenase complex.

The pyruvate dehydrogenase complex plays an important role in the pathways that convert the energy from food into a form that cells can use. This complex converts a molecule called pyruvate, which is formed from the breakdown of carbohydrates, into another molecule called acetyl-CoA. The E1 enzyme performs one part of this chemical reaction. The conversion of pyruvate is essential to begin the series of chemical reactions that produces adenosine triphosphate (ATP), the cell's main energy source.

Health Conditions Related to Genetic Changes

Pyruvate dehydrogenase deficiency

Mutations in the *PDHB* gene are a very rare cause of pyruvate dehydrogenase deficiency. This condition is characterized by a potentially life-threatening buildup of a chemical called lactic acid in the body (lactic acidosis), delayed development, and neurological problems. These mutations change single protein building blocks (amino acids) in the E1 beta protein, resulting in an abnormal E1 beta protein that cannot function properly. The abnormal protein may not be able to interact with E1 alpha to form the E1 enzyme or with other factors needed for the E1 enzyme to perform its chemical reaction. A decrease of functional E1 beta leads to a reduction of pyruvate dehydrogenase complex activity. With decreased function of this complex, pyruvate builds up and is converted, in another chemical reaction, to lactic acid, causing lactic acidosis. In addition, the production of cellular energy is diminished. The brain, which is especially dependent on this form of energy, is severely affected, resulting in the neurological problems associated with pyruvate dehydrogenase deficiency.

Leigh syndrome

MedlinePlus Genetics provides information about Leigh syndrome

Other Names for This Gene

- ODPB_HUMAN
- PDHBD
- PDHE1-B
- PDHE1B
- PHE1B
- pyruvate dehydrogenase (lipoamide) beta
- pyruvate dehydrogenase E1 component subunit beta, mitochondrial
- pyruvate dehydrogenase, E1 beta polypeptide

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PDHB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5162[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PDHB%5BTIAB%5D%29+OR+%28E1+beta%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)

Catalog of Genes and Diseases from OMIM

- LEIGH SYNDROME, NUCLEAR; NULS (https://omim.org/entry/256000)
- PYRUVATE DEHYDROGENASE E1, SUBUNIT BETA; PDHB (https://omim.org/entry/179060)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5162)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PDHB[gene])

References

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- Patel MS, Korotchkina LG, Sidhu S. Interaction of E1 and E3 components withthe core proteins of the human pyruvate dehydrogenase complex. J Mol Catal BEnzym. 2009 Nov 1;61(1-2):2-6. doi: 10.1016/j.molcatb.2009.05.001. Citation on PubMed (ht tps://pubmed.ncbi.nlm.nih.gov/20160912) or Free article on PubMed Central (https:// www.ncbi.nlm.nih.gov/pmc/articles/PMC2770179/)

Genomic Location

The *PDHB* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromoso me/3/).

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