PDHB gene

pyruvate dehydrogenase E1 beta subunit

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

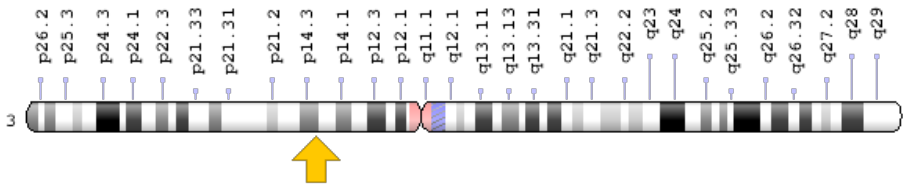
Mutations in the *PDHB* gene are a very rare cause of pyruvate dehydrogenase deficiency. This condition is characterized by a potentially life-threatening buildup of a chemical called lactic acid in the body (lactic acidosis), delayed development, and neurological problems. These mutations change single 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

Chromosomal Location

Cytogenetic Location: 3p14.3, which is the short (p) arm of chromosome 3 at position 14.3

Molecular Location: base pairs 58,427,630 to 58,433,852 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Biochemistry (fifth edition, 2002): The Formation of Acetyl Coenzyme A from Pyruvate
  https://www.ncbi.nlm.nih.gov/books/NBK22427/#A2376
- Biochemistry (fifth edition, 2002): The Pyruvate Dehydrogenase Complex Is Regulated Allosterically and by Reversible Phosphorylation
  https://www.ncbi.nlm.nih.gov/books/NBK22347/#A2410

- Molecular Biology of the Cell (fourth edition, 2002): Sugars and Fats Are Both Degraded to Acetyl CoA in Mitochondria
  https://www.ncbi.nlm.nih.gov/books/NBK26882/#A300
- Molecular Cell Biology (fourth edition, 2000): Mitochondrial Oxidation of Pyruvate Begins with the Formation of Acetyl CoA
  https://www.ncbi.nlm.nih.gov/books/NBK21624/#A4352

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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
  http://omim.org/entry/256000
- PYRUVATE DEHYDROGENASE, BETA POLYPEPTIDE
  http://omim.org/entry/179060

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PDHB.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PDHB%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:8808
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5162
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5162
- UniProt
  https://www.uniprot.org/uniprot/P11177

Sources for This Summary

- Biochemistry (fifth edition, 2002): The Formation of Acetyl Coenzyme A from Pyruvate
  https://www.ncbi.nlm.nih.gov/books/NBK22427/#A2376
- Brown RM, Head RA, Boubriak II, Leonard JV, Thomas NH, Brown GK. Mutations in the gene for the E1beta subunit: a novel cause of pyruvate dehydrogenase deficiency. Hum Genet. 2004 Jul; 115(2):123-7. Epub 2004 May 11.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15138885

- Okajima K, Korotchkina LG, Prasad C, Rupar T, Phillips JA 3rd, Ficicioglu C, Hertecant J, Patel MS, Kerr DS. Mutations of the E1beta subunit gene (PDHB) in four families with pyruvate dehydrogenase deficiency. Mol Genet Metab. 2008 Apr;93(4):371-80. doi: 10.1016/j.ymgme.2007.10.135. Epub 2008 Mar 4.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18164639
- OMIM: PYRUVATE DEHYDROGENASE, BETA POLYPEPTIDE
  http://omim.org/entry/179060
- Patel MS, Korotchkina LG, Sidhu S. Interaction of E1 and E3 components with the core proteins of the human pyruvate dehydrogenase complex. J Mol Catal B Enzym. 2009 Nov 1;61(1-2):2-6.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20160912
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