

PDHX gene

pyruvate dehydrogenase complex component X

Normal Function

The *PDHX* gene provides instructions for making a protein called E3 binding protein, which is part of a large group of proteins known as the pyruvate dehydrogenase complex. This complex is made up of several enzymes, including one called E3, and other proteins. E3 binding protein attaches E3 to the complex and provides the correct structure for the complex to perform its function.

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 enzyme converts a molecule called pyruvate, which is formed from the breakdown of carbohydrates, into another molecule called acetyl-CoA. This conversion 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 *PDHX* gene cause pyruvate dehydrogenase deficiency in a small number of people. 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. *PDHX* gene mutations associated with pyruvate dehydrogenase deficiency result in the complete absence of E3 binding protein. Loss of this protein impairs the binding of the E3 enzyme to the pyruvate dehydrogenase complex, which leads to a reduction of the complex's 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

- dihydrolipoamide dehydrogenase-binding protein of pyruvate dehydrogenase complex
- DLDBP
- E3BP
- lipoyl-containing pyruvate dehydrogenase complex component X
- ODPX_HUMAN
- OPDX
- proX
- pyruvate dehydrogenase complex, component X
- pyruvate dehydrogenase complex, E3-binding protein subunit
- pyruvate dehydrogenase complex, lipoyl-containing component X
- pyruvate dehydrogenase protein X component, mitochondrial

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PDHX ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8050\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8050[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PDHX%5BTIAB%5D%29+OR+%28E3BP%5BTIAB%5D%29+OR+%28DLDBP%5BTIAB%5D%29%29+AND+%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

- PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X; PDHX (<https://omim.org/entry/608769>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8050>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PDHX\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PDHX[gene]))

References

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- Smolle M, Prior AE, Brown AE, Cooper A, Byron O, Lindsay JG. A new level of architectural complexity in the human pyruvate dehydrogenase complex. J Biol Chem. 2006 Jul 14;281(28):19772-80. doi: 10.1074/jbc.M601140200. Epub 2006 May 5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16679318>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3954457/>)

Genomic Location

The *PDHX* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

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