

## PDP1 gene

pyruvate dehydrogenase phosphatase catalytic subunit 1

### Normal Function

The *PDP1* gene provides instructions for making a protein called pyruvate dehydrogenase phosphatase 1, which is part of a large group of proteins called the pyruvate dehydrogenase complex. The pyruvate dehydrogenase phosphatase 1 protein turns on (activates) the complex by removing a phosphate group (a cluster of oxygen and phosphorus atoms) from the 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 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

At least one mutation in the *PDP1* gene has been identified in individuals with pyruvate dehydrogenase deficiency; mutation of the *PDP1* gene is a very rare cause of this condition. Pyruvate dehydrogenase deficiency is characterized by a potentially life-threatening buildup of a chemical called lactic acid in the body (lactic acidosis), delayed development, and neurological problems.

The identified mutation removes one protein building block (amino acid) of the pyruvate dehydrogenase phosphatase 1 protein, which is thought to change its shape. The abnormal protein cannot remove the phosphate group from the pyruvate dehydrogenase complex, which reduces the activity of the complex. With decreased activity 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.

## Other Names for This Gene

- PDH
- PDP
- PDP 1
- PDP1\_HUMAN
- PDPC
- PDPC 1
- PPM2C
- protein phosphatase 2C, magnesium-dependent, catalytic subunit
- pyruvate dehydrogenase (Lipoamide) phosphatase-phosphatase
- pyruvate dehydrogenase [acetyl-transferring]-phosphatase 1, mitochondrial
- pyruvate dehydrogenase phosphatase catalytic subunit 1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of PDP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54704\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54704[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PDP1%5BTIAB%5D%29+OR+%28PDP+1%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+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- PYRUVATE DEHYDROGENASE PHOSPHATASE, CATALYTIC SUBUNIT 1; PDP1 (<https://omim.org/entry/605993>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/54704>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PDP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PDP1[gene]))

## References

- Cameron JM, Maj M, Levandovskiy V, Barnett CP, Blaser S, Mackay N, Raiman J, Feigenbaum A, Schulze A, Robinson BH. Pyruvate dehydrogenase phosphatase 1 (

PDP1) null mutation produces a lethal infantile phenotype. Hum Genet. 2009 Apr;125(3):319-26. doi: 10.1007/s00439-009-0629-6. Epub 2009 Jan 30. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19184109>)

- Ganetzky R, McCormick EM, Falk MJ. Primary Pyruvate Dehydrogenase Complex Deficiency Overview. 2021 Jun 17. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews(R) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from <http://www.ncbi.nlm.nih.gov/books/NBK571223/> Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/34138529>)
- Maj MC, MacKay N, Levandovskiy V, Addis J, Baumgartner ER, Baumgartner MR, Robinson BH, Cameron JM. Pyruvate dehydrogenase phosphatase deficiency: identification of the first mutation in two brothers and restoration of activity by protein complementation. J Clin Endocrinol Metab. 2005 Jul;90(7):4101-7. doi:10.1210/jc.2005-0123. Epub 2005 Apr 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15855260>)
- Roche TE, Hiromasa Y, Turkan A, Gong X, Peng T, Yan X, Kasten SA, Bao H, Dong J. Essential roles of lipoyl domains in the activated function and control of pyruvate dehydrogenase kinases and phosphatase isoform 1. Eur J Biochem. 2003 Mar;270(6):1050-6. doi: 10.1046/j.1432-1033.2003.03468.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12631265>)

## Genomic Location

The *PDP1* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome/8/>).

**Last updated July 1, 2012**