

PC gene

pyruvate carboxylase

Normal Function

The *PC* gene provides instructions for making an enzyme called pyruvate carboxylase. This enzyme is active in mitochondria, which are the energy-producing centers within cells.

Pyruvate carboxylase is responsible for a chemical reaction that converts a molecule called pyruvate to another molecule called oxaloacetate. This reaction is essential for several different cellular functions. In the kidneys and liver, it is the first step in a process called gluconeogenesis. Gluconeogenesis generates glucose, a simple sugar that is the body's main energy source. This chemical reaction also occurs in the pancreas, where it helps regulate the secretion of a hormone called insulin. Insulin controls the amount of glucose in the blood that is passed into cells for conversion to energy.

In fat-storing (adipose) tissue, pyruvate carboxylase is involved in the formation of certain fats (lipogenesis). This enzyme also plays an important role in the nervous system, where it replenishes the building blocks needed to make brain chemicals called neurotransmitters. Additionally, pyruvate carboxylase is necessary for the formation of myelin, which is the fatty covering that insulates and protects certain nerve cells.

Health Conditions Related to Genetic Changes

Pyruvate carboxylase deficiency

More than 30 mutations in the *PC* gene have been identified in people with pyruvate carboxylase deficiency. This condition causes lactic acid and other potentially toxic compounds to accumulate in the blood. High levels of these substances can damage the body's organs and tissues, particularly in the nervous system.

Most *PC* gene mutations change a single protein building block (amino acid) in pyruvate carboxylase, which reduces the amount of this enzyme in cells or disrupts its ability to effectively convert pyruvate to oxaloacetate. Other genetic changes lead to the production of an abnormally short version of the enzyme that is completely nonfunctional.

If pyruvate carboxylase is missing or altered, it cannot carry out its role in generating

glucose. Any disruption in gluconeogenesis impairs the body's ability to make energy in mitochondria. Additionally, a loss of pyruvate carboxylase allows lactic acid and ammonia, among other compounds, to build up and damage organs and tissues. Researchers suggest that the loss of pyruvate carboxylase function in the nervous system, particularly the role of the enzyme in myelin formation and neurotransmitter production, also contributes to the neurologic features of pyruvate carboxylase deficiency.

Other Names for This Gene

- PCB
- PYC_HUMAN
- Pyruvic carboxylase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PC ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5091\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5091[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28pyruvate+carboxylase%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- PYRUVATE CARBOXYLASE; PC (<https://omim.org/entry/608786>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5091>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PC\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PC[gene]))

References

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Genomic Location

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

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