

PCCA gene

propionyl-CoA carboxylase subunit alpha

Normal Function

The *PCCA* gene provides instructions for making part of an enzyme called propionyl-CoA carboxylase, specifically, the alpha subunit of this enzyme. Six alpha subunits come together with six beta subunits (produced from the *PCCB* gene) to form a functioning enzyme. The alpha subunit also includes a region for binding to the B vitamin biotin.

Propionyl-CoA carboxylase plays a role in the normal processing of proteins. It carries out a particular step in the breakdown of several protein building blocks (amino acids) called isoleucine, methionine, threonine, and valine. Propionyl-CoA carboxylase also helps break down certain types of lipids (fats) and cholesterol. First, several chemical reactions convert the amino acids, lipids, or cholesterol to a molecule called propionyl-CoA. Using biotin, propionyl-CoA carboxylase then converts propionyl-CoA to a molecule called methylmalonyl-CoA. Additional enzymes break down methylmalonyl-CoA into other molecules that are used for energy.

Health Conditions Related to Genetic Changes

Propionic acidemia

More than 120 mutations in the *PCCA* gene have been identified in people with propionic acidemia, a condition that causes severe health problems appearing shortly after birth. These mutations include changes in single DNA building blocks (nucleotides) and insertions or deletions of genetic material in the *PCCA* gene. *PCCA* mutations prevent the production of functional propionyl-CoA carboxylase or reduce the enzyme's activity. The altered or missing enzyme is unable to process certain parts of proteins and lipids properly. As a result, propionyl-CoA and other potentially harmful compounds can build up to toxic levels in the body. This buildup damages the brain and nervous system, causing the serious health problems associated with propionic acidemia.

Other Names for This Gene

- PCCA_HUMAN
- PCCase alpha subunit
- propionyl CoA carboxylase, alpha polypeptide

- propionyl Coenzyme A carboxylase, alpha polypeptide
- propionyl-CoA carboxylase alpha subunit
- propionyl-CoA:carbon dioxide ligase alpha subunit

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PCCA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5095\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5095[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PCCA%5BTIAB%5D%29+OR+%28propionyl+Coenzyme+A+carboxylase%5BTIAB%5D%29%29+OR+%28propionyl-CoA%5BTIAB%5D%29+NOT+%28cholangiocarcinoma%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

Catalog of Genes and Diseases from OMIM

- PROPIONYL-CoA CARBOXYLASE, ALPHA SUBUNIT; PCCA (<https://omim.org/entry/232000>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5095>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PCCA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PCCA[gene]))

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Genomic Location

The *PCCA* gene is found on chromosome 13 (<https://medlineplus.gov/genetics/chromosome/13/>).

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