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.
**Chromosomal Location**

**Cytogenetic Location:** 13q32.3, which is the long (q) arm of chromosome 13 at position 32.3

**Molecular Location:** base pairs 100,089,015 to 100,530,437 on chromosome 13 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

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

**Additional Information & Resources**

**Educational Resources**

- Basic Neurochemistry (sixth edition, 1999): Propionyl-CoA carboxylase deficiency blocks the biotin- and ATP-dependent conversion of propionyl-CoA to methylmalonyl-CoA
  https://www.ncbi.nlm.nih.gov/books/NBK27933/#A3116

**Clinical Information from GeneReviews**

- Propionic Acidemia
  https://www.ncbi.nlm.nih.gov/books/NBK92946
Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- PROPIONYL-CoA CARBOXYLASE, ALPHA SUBUNIT
  http://omim.org/entry/232000

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PCCA.html
- ClinVar
- HGNC Gene Symbol Report
- Kraus Lab at the University of Colorado Health Sciences Center
  http://www.ucdenver.edu/academics/colleges/medicalschool/programs/kraus/Pages/home.aspx
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5095
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P05165

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15464417
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/27227689


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