PCCB gene

propionyl-CoA carboxylase subunit beta

Normal Function

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

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 the B vitamin 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

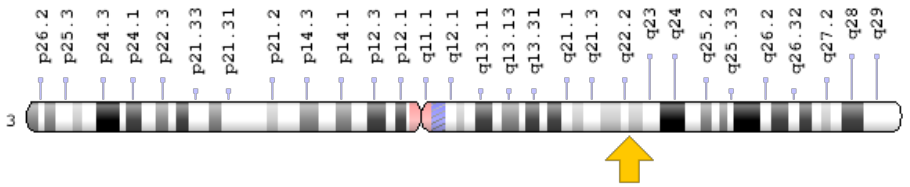
Propionic acidemia

More than 100 mutations in the *PCCB* 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 *PCCB* gene. *PCCB* mutations prevent the production of functional propionyl-CoA carboxylase or reduce the enzyme's activity. The altered or missing enzyme prevents certain parts of proteins and lipids from being broken down properly. As a result, propionyl-CoA and other potentially toxic 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: 3q22.3, which is the long (q) arm of chromosome 3 at position 22.3

Molecular Location: base pairs 136,250,325 to 136,330,171 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- PCCase beta subunit
- PCCB_HUMAN
- propanoyl-CoA:carbon dioxide ligase beta subunit
- propionyl-CoA carboxylase beta subunit
- propionyl CoA carboxylase, beta polypeptide
- propionyl-CoA carboxylase, beta subunit

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%28PCCB%5BTIAB%5D%29+OR+%28propionyl+Coenzyme+A+carboxylase%5BTIAB%5D%29%29+OR+%28propanoyl-CoA%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- PROPIONYL-CoA CARBOXYLASE, BETA SUBUNIT
  http://omim.org/entry/232050

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PCCB%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:8654
- 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:5096
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5096
- UniProt
  https://www.uniprot.org/uniprot/P05166

Sources for This Summary

- Desviat LR, Pérez B, Pérez-Cerdá C, Rodríguez-Pombo P, Clavero S, Ugarte M. Propionic acidemia: mutation update and functional and structural effects of the variant alleles. Mol Genet Metab. 2004 Sep-Oct;83(1-2):28-37. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15464417
- Gupta D, Bijarnia-Mahay S, Kohli S, Saxena R, Puri RD, Shigematsu Y, Yamaguchi S, Sakamoto O, Gupta N, Kabra M, Thakur S, Deb R, Verma IC. Seventeen Novel Mutations in PCCA and PCCB Genes in Indian Propionic Acidemia Patients, and Their Outcomes. Genet Test Mol Biomarkers. 2016 Jul;20(7):373-82. doi: 10.1089/gtmb.2016.0017. Epub 2016 May 26.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/27227689

- Kraus JP, Spector E, Venezia S, Estes P, Chiang PW, Creadon-Swindell G, Müllerleile S, de Silva L, Barth M, Walter M, Walter K, Meissner T, Lindner M, Ensenauer R, Santer R, Bodamer OA, Baumgartner MR, Brunner-Krainz M, Karall D, Haase C, Knerr I, Marquardt T, Hennermann JB, Steinfeld R, Beblo S, Koch HG, Konstantopoulou V, Scholl-Bürgi S, van Teeffelen-Heithoff A, Suormala T, Ugarte M, Sperl W, Superti-Furga A, Schwab KO, Grünert SC, Sass JO. Mutation analysis in 54 propionic acidemia patients. J Inherit Metab Dis. 2012 Jan;35(1):51-63. doi: 10.1007/s10545-011-9399-0. Epub 2011 Oct 27.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22033733
- Pérez-Cerdá C, Clavero S, Pérez B, Rodríguez-Pombo P, Desviat LR, Ugarte M. Functional analysis of PCCB mutations causing propionic acidemia based on expression studies in deficient human skin fibroblasts. Biochim Biophys Acta. 2003 May 20;1638(1):43-9.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12757933

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