PROC gene

protein C, inactivator of coagulation factors Va and VIIIa

Normal Function

The *PROC* gene provides instructions for making a protein called protein C that is important for controlling blood clotting. Protein C blocks the activity of two proteins that promote the formation of blood clots, called factor Va and factor VIIIa. Protein C is also involved in controlling inflammation. Inflammation is a normal body response to infection, irritation, or other injury.

Protein C is made in the liver and then released into the bloodstream. The protein remains turned off (inactive) until it attaches to a protein called thrombin, which converts it to activated protein C (APC). APC cuts (cleaves) the factor Va protein at specific sites, which partially or completely inactivates factor Va. (The inactive form is called factor V.) APC then works with factor V to inactivate factor VIIIa.

Health Conditions Related to Genetic Changes

Protein C deficiency

At least 270 mutations in the *PROC* gene have been found to cause protein C deficiency. Most of these mutations change single protein building blocks (amino acids) in protein C, which disrupts its ability to control blood clotting. Protein C deficiency can be divided into type I and type II based on the mutation in the *PROC* gene.

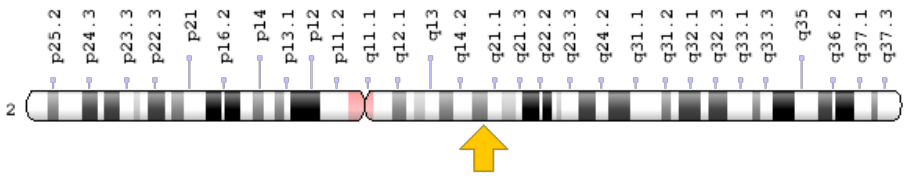
Protein C deficiency type I is caused by *PROC* gene mutations that result in reduced levels of protein C. Affected individuals do not have enough protein C to control blood clotting, which causes the increased risk for abnormal blood clots in protein C deficiency.

Mutations that cause protein C deficiency type II result in the production of an altered protein C with reduced activity. Individuals with this form of the condition have normal levels of protein C, but the protein is not able to interact with other molecules involved in blood clotting. If protein C cannot control blood clotting, abnormal blood clots may form.

Chromosomal Location

Cytogenetic Location: 2q14.3, which is the long (q) arm of chromosome 2 at position 14.3

Molecular Location: base pairs 127,418,143 to 127,429,246 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- PC
- PROC1
- PROC_HUMAN
- protein C
- protein C (inactivator of coagulation factors Va and VIIIa)

Additional Information & Resources

Educational Resources

- Madam Curie Bioscience Database: Protein C Deficiencies https://www.ncbi.nlm.nih.gov/books/NBK6069/#A2907

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PROC%5BTI%5D%29+OR+%28protein+C%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PROTEIN C http://omim.org/entry/612283

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PROC.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PROC%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:9451
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5624
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5624
- UniProt
  https://www.uniprot.org/uniprot/P04070

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