

## 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.

### Other Names for This Gene

- PC

- PROC1
- PROC\_HUMAN
- protein C
- protein C (inactivator of coagulation factors Va and VIIIa)

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of PROC ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5624\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5624[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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; PROC (<https://omim.org/entry/612283>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5624>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PROC\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PROC[gene]))

## **References**

- Brouwer JL, Lijfering WM, Ten Kate MK, Kluin-Nelemans HC, Veeger NJ, van der Meer J. High long-term absolute risk of recurrent venous thromboembolism inpatients with hereditary deficiencies of protein S, protein C or antithrombin. *Thromb Haemost.* 2009 Jan;101(1):93-9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19132194>)
- Ursi P, Marino F, Caprera A, Milanesi L, Faioni EM, Rovida E. ProCMD: a database and 3D web resource for protein C mutants. *BMC Bioinformatics.* 2007 Mar;8 Suppl 1(Suppl 1):S11. doi: 10.1186/1471-2105-8-S1-S11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17430555>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1885840/>)
- Goldenberg NA, Manco-Johnson MJ. Protein C deficiency. *Haemophilia.* 2008 Nov; 14(6):1214-21. doi: 10.1111/j.1365-2516.2008.01838.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19141162>)

- Knoebl PN. Severe congenital protein C deficiency: the use of protein C concentrates (human) as replacement therapy for life-threatening blood-clotting complications. *Biologics*. 2008 Jun;2(2):285-96. doi: 10.2147/btt.s1954. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19707361>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2721356/>)

## **Genomic Location**

The *PROC* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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