



F2 gene

coagulation factor II, thrombin

Normal Function

The *F2* gene provides instructions for making a protein called prothrombin (also called coagulation factor II). Coagulation factors are a group of related proteins that are essential for normal blood clotting (hemostasis). After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

Prothrombin is made chiefly by cells in the liver. The protein circulates in the bloodstream in an inactive form until an injury occurs that damages blood vessels. In response to injury, prothrombin is converted to its active form, thrombin. Thrombin then converts a protein called fibrinogen into fibrin, the primary protein that makes up blood clots.

Thrombin is also thought to be involved in cell growth and division (proliferation), tissue repair, and the formation of new blood vessels (angiogenesis).

Health Conditions Related to Genetic Changes

Prothrombin deficiency

More than 50 mutations in the *F2* gene have been found to cause prothrombin deficiency. Most of these mutations change one protein building block (amino acid) in prothrombin. Some mutations drastically reduce the activity of prothrombin and can lead to severe bleeding episodes. Other mutations allow for a moderate amount of activity of prothrombin, typically causing mild bleeding episodes. None of the mutations identified eliminate prothrombin function. Researchers believe that people cannot live with a complete absence of prothrombin.

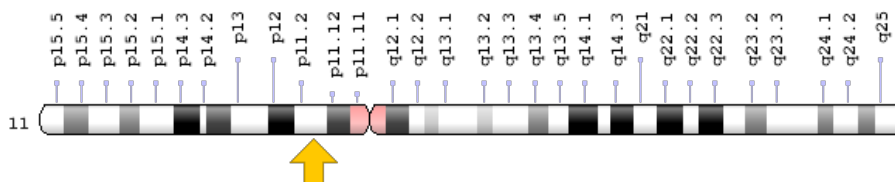
Prothrombin thrombophilia

The mutation that causes most cases of prothrombin thrombophilia changes one DNA building block (nucleotide) in the *F2* gene. Specifically, it replaces the nucleotide guanine with the nucleotide adenine at position 20210 (written G20210A or 20210G>A). This mutation, which occurs in a region of the gene called the 3' untranslated region, causes the gene to be overactive and leads to the production of too much prothrombin. An abundance of prothrombin leads to more thrombin, which promotes the formation of blood clots.

Chromosomal Location

Cytogenetic Location: 11p11.2, which is the short (p) arm of chromosome 11 at position 11.2

Molecular Location: base pairs 46,719,166 to 46,739,508 on chromosome 11 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Blood Coagulation Factor II
- coagulation factor II
- coagulation factor II (thrombin)
- prothrombin B-chain
- PT
- Q7Z7P3_HUMAN
- serine protease

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Prothrombin Is Readied for Activation by a Vitamin K-Dependent Modification
<https://www.ncbi.nlm.nih.gov/books/NBK22589/#A1406>
- Guide to Peripheral and Cerebrovascular Intervention (first edition, 2004): Common hypercoagulable states: PT G20210A
<https://www.ncbi.nlm.nih.gov/books/NBK27369/#A1096>
- National Center for Biotechnology Information: Mutations and Blood Clots
<https://www.ncbi.nlm.nih.gov/books/NBK2318/>

Clinical Information from GeneReviews

- Prothrombin-Related Thrombophilia
<https://www.ncbi.nlm.nih.gov/books/NBK1148>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28F2%5BTI%5D%29+OR+%28coagulation+factor+II%5BTI%5D%29+OR+%28prothrombin%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+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- COAGULATION FACTOR II
<http://omim.org/entry/176930>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_F2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=F2%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3535
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:2147>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2147>
- UniProt
<https://www.uniprot.org/uniprot/P00734>

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<https://ghr.nlm.nih.gov/gene/F2>

Reviewed: September 2008

Published: June 11, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services