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 46,719,201 to 46,739,506 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (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


• 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
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2147
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P00734

Sources for This Summary

- OMIM: COAGULATION FACTOR II
  http://omim.org/entry/176930
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12421139

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16024978

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15262854

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