F2 gene

coagulation factor II, thrombin

Normal Function

The *F2* gene provides instructions for making a protein called prothrombin (also known as 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 damages blood vessels. In response to the 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

Several variants (also called mutations) in the *F2* gene have been found to cause prothrombin deficiency, a bleeding disorder that slows the clotting process. Most of these variants change one protein building block (amino acid) in prothrombin. Some variants drastically reduce the activity of prothrombin and can lead to severe bleeding episodes. Other variants allow for a moderate amount of prothrombin activity, which typically causes mild bleeding episodes. None of the variants that are known to cause prothrombin deficiency eliminate prothrombin function. Researchers believe that some prothrombin is necessary for life.

Prothrombin thrombophilia

Variants in the *F2* gene may also cause prothrombin thrombophilia, an inherited disorder that is associated with excessive blood clotting. Most cases of this condition are caused by a variant that changes one DNA building block (nucleotide) in the *F2* gene. Specifically, the nucleotide guanine is replaced with the nucleotide adenine at position 20210 (written as G20210A or 20210G>A). This variant, which occurs in a region of the gene called the 3' untranslated region, causes cells to produce too much

prothrombin. More prothrombin leads to more thrombin, which promotes the formation of blood clots in the absence of injury. When these clots block blood flow, it can cause serious health complications.

Other Names for This Gene

- coagulation factor II
- coagulation factor II (thrombin)
- PT
- RPRGL2
- THPH1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of F2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2147[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; F2 (https://omim.org/entry/176930)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2147)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=F2[gene])

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Genomic Location

The *F2* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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