F7 gene

coagulation factor VII

Normal Function

The *F7* gene provides instructions for making a protein called coagulation factor VII. Coagulation factors are a group of related proteins that are involved in the coagulation system, which is a series of chemical reactions that form blood clots. After an injury, clots seal off blood vessels to stop bleeding and trigger blood vessel repair.

Coagulation factor VII is made primarily by cells in the liver. The protein circulates in the bloodstream in an inactive form until the coagulation system is turned on (activated) by an injury that damages blood vessels. Activated coagulation factor VII helps turn on other coagulation factors in turn. This step-wise process ultimately promotes the conversion of an important coagulation protein called fibrinogen into fibrin, which is the material that forms blood clots.

Health Conditions Related to Genetic Changes

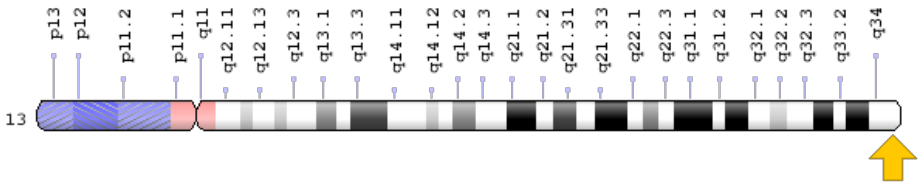
Factor VII deficiency

Almost 300 mutations in the *F7* gene have been found to cause a rare bleeding disorder called factor VII deficiency. This disorder commonly causes nosebleeds, easy bruising, bleeding of the gums, and prolonged or excessive bleeding following surgery or physical injury. In severe cases, life-threatening episodes of bleeding inside the skull or gastrointestinal tract can occur. Some affected individuals have no bleeding problems. The *F7* gene mutations involved in this condition reduce the amount of coagulation factor VII in the bloodstream. A shortage of coagulation factor VII prevents blood from clotting normally, causing episodes of abnormal bleeding that can be severe. What determines the severity of the condition is unclear; it does not appear to be related to the amount of coagulation factor VII in the blood.

Chromosomal Location

Cytogenetic Location: 13q34, which is the long (q) arm of chromosome 13 at position 34

Molecular Location: base pairs 113,105,773 to 113,120,681 on chromosome 13 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FVII coagulation protein
- proconvertin
- serum prothrombin conversion accelerator
- SPCA

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Diagram: Blood-Clotting Cascade https://www.ncbi.nlm.nih.gov/books/NBK22589/?rendertype=figure&id=A1401

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28F7%5BTIAB%5D%29+OR+%28coagulation+factor+VII%5BTIAB%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+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- COAGULATION FACTOR VII http://omim.org/entry/613878

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_F7.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=F7%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3544
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2155
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2155
- UniProt
  https://www.uniprot.org/uniprot/P08709

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