F11 gene

coagulation factor XI

Normal Function

The *F11* gene provides instructions for making a protein called factor XI. This protein plays a role in the coagulation cascade, which is a series of chemical reactions that forms blood clots in response to injury. After an injury, clots seal off blood vessels to stop bleeding and trigger blood vessel repair.

Factor XI is made primarily by cells in the liver. The protein circulates in the bloodstream and is normally turned off (inactive) until the coagulation cascade is turned on (activated) by an injury that damages blood vessels. When factor XI is activated, it interacts with other coagulation factors, resulting in conversion of an important coagulation protein called prothrombin to its active form, thrombin. Thrombin then converts a protein called fibrinogen into fibrin, which is the material that forms blood clots.

Health Conditions Related to Genetic Changes

Factor XI deficiency

About 250 mutations in the *F11* gene have been found to cause a rare bleeding disorder called factor XI deficiency. This disorder, while usually mild, can cause nosebleeds, easy bruising, bleeding under the skin, bleeding of the gums, and prolonged or excessive bleeding following surgery, dental procedures, or trauma. The *F11* gene mutations that cause factor XI deficiency reduce the amount of factor XI in the bloodstream or result in the production of a factor XI protein with impaired function. A deficiency of functional factor XI slows blood clotting, causing episodes of abnormal bleeding.

The amount of functional factor XI remaining varies depending on the particular mutation and whether one or both copies of the *F11* gene in each cell have mutations. However, the severity of the bleeding problems in affected individuals does not necessarily correspond to the amount of factor XI in the bloodstream, and can vary even within the same family. Other genetic and environmental factors likely play a role in determining the severity of this condition.

Other Names for This Gene

- coagulation factor XI preproprotein
- FXI
- plasma thromboplastin antecedent
- PTA

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of F11 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2160[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28F11%5BTIAB%5D%29+OR+%28factor+XI%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- COAGULATION FACTOR XI; F11 (https://omim.org/entry/264900)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2160)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=F11[gene])

References

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Genomic Location

The *F11* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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