F8 gene

coagulation factor VIII

Normal Function

The *F8* gene provides instructions for making a protein called coagulation factor VIII. Coagulation factors are a group of related proteins that are essential for the formation of blood clots. After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

Coagulation factor VIII is made chiefly by cells in the liver. This protein circulates in the bloodstream in an inactive form, bound to another molecule called von Willebrand factor, until an injury that damages blood vessels occurs. In response to injury, coagulation factor VIII is activated and separates from von Willebrand factor. The active protein (sometimes written as coagulation factor VIIIa) interacts with another coagulation factor called factor IX. This interaction sets off a chain of additional chemical reactions that form a blood clot.

Health Conditions Related to Genetic Changes

Hemophilia

Mutations in the *F8* gene cause hemophilia A, the most common form of this bleeding disorder. More than 1,300 alterations in this gene have been identified. Some of these mutations change single DNA building blocks (base pairs) in the gene, while others delete or insert multiple base pairs. The most common mutation in people with severe hemophilia A is a rearrangement of genetic material called an inversion. This inversion involves a large segment of the *F8* gene.

Mutations in the *F8* gene lead to the production of an abnormal version of coagulation factor VIII or reduce the amount of this protein. The altered or missing protein cannot participate effectively in the blood clotting process. As a result, blood clots cannot form properly in response to injury. These problems with blood clotting lead to excessive bleeding that can be difficult to control. Some mutations, such as the large inversion described above, almost completely eliminate the activity of coagulation factor VIII and result in severe hemophilia. Other mutations reduce but do not eliminate the protein's activity, resulting in mild or moderate hemophilia.

Other Names for This Gene

- AHF
- antihemophilic factor
- coagulation factor VIII, procoagulant component
- coagulation factor VIII, procoagulant component (hemophilia A)
- DXS1253E
- FA8_HUMAN
- Factor VIIIF8B
- FVIII
- HEMA
- procoagulant component

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of F8 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2157[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28F8%5BTIAB%5D%29+OR+%28coagulation+factor+VIII%5BTI%5D%29+OR+%28factor+VIII%5BMAJR%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 VIII; F8 (https://omim.org/entry/300841)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2157)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=F8[gene])

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Genomic Location

The *F8* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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