F9 gene

coagulation factor IX

Normal Function

The *F9* gene provides instructions for making a protein called coagulation factor IX. 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 IX is made in the liver. This protein circulates in the bloodstream in an inactive form until an injury that damages blood vessels occurs. In response to injury, coagulation factor IX is activated by another coagulation factor called factor XIa. The active protein (sometimes written as coagulation factor IXa) interacts with coagulation factor VIII and other molecules. These interactions set off a chain of additional chemical reactions that form a blood clot.

Health Conditions Related to Genetic Changes

Hemophilia

Mutations in the *F9* gene cause a type of hemophilia called hemophilia B. More than 900 alterations in this gene have been identified. The most common mutations change single DNA building blocks (base pairs) in the gene. A small percentage of mutations delete or insert multiple base pairs or rearrange segments of DNA within the gene.

Mutations in the *F9* gene lead to the production of an abnormal version of coagulation factor IX 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. Mutations that completely eliminate the activity of coagulation factor IX result in severe hemophilia. Mutations that reduce but do not eliminate the protein's activity usually cause mild or moderate hemophilia.

Several mutations near the beginning of the *F9* gene sequence cause an unusual form of hemophilia known as hemophilia B Leyden. People with these mutations are born with very low levels of functional coagulation factor IX, but hormonal changes cause the levels of this protein to increase gradually during puberty. As a result, adults with hemophilia B Leyden rarely experience episodes of abnormal bleeding.

Warfarin sensitivity

MedlinePlus Genetics provides information about Warfarin sensitivity

Other disorders

Several rare mutations in the *F9* gene cause an increased sensitivity (hypersensitivity) to a drug called warfarin. This medication is an anticoagulant, which means it is used to prevent the formation or growth of abnormal blood clots. Warfarin works by reducing the amount of active factor IX and three other coagulation proteins.

The mutations responsible for warfarin hypersensitivity each change a single base pair in the *F9* gene. These mutations do not cause hemophilia B, and people with these genetic changes only have bleeding problems if they are treated with warfarin. Warfarin reduces the amount of coagulation factor IX to very low levels in these individuals, which prevents the blood from clotting normally and can lead to recurrent, severe bleeding problems. To avoid these complications, people with warfarin hypersensitivity can be treated with other anticoagulant medications.

Other Names for This Gene

- Christmas factor
- coagulation factor IX (plasma thromboplastic component, Christmas disease, hemophilia B)
- FA9_HUMAN
- Factor 9
- FIX
- HEMB
- Plasma thromboplastin component
- PTC

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of F9 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2158[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28F9%5BTI%5D%29+OR+%28factor+IX%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+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- COAGULATION FACTOR IX; F9 (https://omim.org/entry/300746)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2158)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=F9[gene])

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Genomic Location

The *F9* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

Last updated May 1, 2010