FGB gene

fibrinogen beta chain

Normal Function

The *FGB* gene provides instructions for making a protein called the fibrinogen B beta (Bβ) chain, one piece (subunit) of the fibrinogen protein. This protein is important for blood clot formation (coagulation), which is needed to stop excessive bleeding after injury. To form fibrinogen, the Bβ chain attaches to two other proteins called the fibrinogen A alpha (Aα) and fibrinogen gamma (γ) chains, each produced from different genes. Two sets of this three-protein complex combine to form functional fibrinogen.

For coagulation to occur, another protein called thrombin removes a piece from the Aα and the Bβ subunits of the functional fibrinogen protein (the pieces are called the A and B fibrinopeptides). This process converts fibrinogen to fibrin, the main protein in blood clots. Fibrin proteins attach to each other, forming a stable network that makes up the blood clot.

Health Conditions Related to Genetic Changes

Congenital afibrinogenemia

Mutations in the *FGB* gene can lead to congenital afibrinogenemia, a condition that causes excessive bleeding due to the absence of fibrinogen protein in the blood. Most *FGB* gene mutations that cause this condition lead to an abnormally short blueprint for protein formation (mRNA). If any fibrinogen Bβ chain is produced, it is nonfunctional. Some mutations in the *FGB* gene result in the formation of a protein that cannot be released from the cell, making the protein effectively nonfunctional. Because this condition occurs when both copies of the *FGB* gene are altered, there is a complete absence of functional fibrinogen Bβ chain. Without the Bβ subunit, the fibrinogen protein is not assembled, which results in the absence of fibrin. As a result, blood clots do not form in response to injury, leading to excessive bleeding.

Other disorders

Mutations in one or both copies of the *FGB* gene can cause other bleeding disorders known as hypofibrinogenemia, dysfibrinogenemia, or hypodysfibrinogenemia.

Hypofibrinogenemia is a condition characterized by decreased levels of fibrinogen in the

blood. This condition is caused by mutations that reduce but do not eliminate the production of the fibrinogen Bβ chain. People with hypofibrinogenemia can have bleeding problems that vary from mild to severe. Generally, the less fibrinogen in the blood, the more severe the bleeding problems are.

Dysfibrinogenemia is a condition characterized by abnormally functioning fibrinogen, although the protein is present at normal levels. This condition is usually caused by mutations that change a single protein building block (amino acid) in the fibrinogen Bβ chain. These mutations alter the function of the fibrinogen protein and, depending on the functional change, can lead to excessive bleeding or abnormal blood clotting (thrombosis).

Hypodysfibrinogenemia is a condition characterized by low levels of abnormally functioning fibrinogen protein in the blood. As in dysfibrinogenemia, this condition can result in excessive bleeding or thrombosis.

Other Names for This Gene

- FIBB_HUMAN
- fibrinogen beta chain isoform 1 preproprotein
- fibrinogen beta chain isoform 2 preproprotein
- fibrinogen, B beta polypeptide

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FGB (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2244[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FGB%5BTIAB%5D%29+OR+%28fibrinogen+beta+chain%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

- FIBRINOGEN, B BETA POLYPEPTIDE; FGB (https://omim.org/entry/134830)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2244)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FGB[gene])

References

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Genomic Location

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

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