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My46 Trait Profile

Hemophilia B

Other Names: Factor IX deficiency; Factor 9 deficiency

Hemophilia B is a bleeding disorder in which the blood does not clot normally. It is by mutations in the F9 gene which makes the factor 9 protein.

Characteristics of Hemophilia B

Individuals with hemophilia bleed longer than individuals without hemophilia. Hemophilia B is characterized by a wide range of signs and symptoms. These can include but are not limited to bleeding into joints and muscles, spontaneous bleeding (bleeding for no apparent reason), bleeding after trauma or surgery, easy bruising, intracranial bleeding (i.e., bleeding around the skull), gastrointestinal bleeding, and hematuria (blood in the urine). The degree of bleeding often depends on the severity of hemophilia present in the individual. Individuals with hemophilia B can have a variety of complications resulting from their bleeding disorder. These complications include varying degrees of joint disease due to a history of bleeding into the joint. The degree of joint disease depends on a variety of factors including the number of bleeds into the joint, past treatment for bleeding, and age.

Diagnosis/Testing

Hemophilia B is caused by a changes or mutations in a gene called F9. The F9 gene makes the clotting factor IX protein. Clotting factors are proteins that control bleeding. Mutations in the F9 gene do not allow the factor IX protein to work properly, and this leads to the bleeding problems seen in affected individuals. Hemophilia B can be also be diagnosed by a laboratory finding of a decreased factor IX activity level in blood. The normal range of factor IX is generally between 50 and 150%. Levels below 40% are consistent with hemophilia. The bleeding disorder is divided into three categories of severity: severe (5-30% factor IX).

Management/Surveillance

Hemophilia B is treated with intravenous (IV) replacement of the missing clotting factor. Thus, individuals with hemophilia B are given replacement of factor IX protein. Individuals with hemophilia B are treated with regular IV infusion of the missing clotting factor or by an “on-demand” schedule meaning they are infused when they experience a bleed. Which regimen an individual is prescribed depends on multiple factors including severity of hemophilia, age, lifestyle, number of bleeds, etc. When possible, aspirin and aspirin-containing products should be avoided in individuals with hemophilia B.

Mode of inheritance

Hemophilia B is inherited in an X-linked recessive pattern. This means that in females, both copies of the F9 gene (i.e., one on each X chromosome) must have a change or mutation, whereas in males, only one copy of the F9 gene must have a mutation to be affected. A female with a mutation in one copy of the F9 gene is said to be a “carrier” of Hemophilia B, and is typically not affected.

Risk to family members

If a father is affected with hemophilia B, his daughters will be carriers of hemophilia B and his sons will be unaffected. If a mother is a carrier of hemophilia B, each daughter has a 1 in 2 chance (i.e., 50%) of being a carrier and each son has a 1 in 2 chance (i.e., 50%) of being affected with hemophilia B.

A variety of reproductive options are available to individuals with a family history of hemophilia including natural conception, egg/sperm donation, prenatal diagnosis, and preimplantation diagnosis. Additional information about these options can be discussed with a genetic counselor.

Special considerations

Unlike many X-linked recessive disorders, female carriers of hemophilia B can have symptoms related to having a decreased factor IX level. For this reason it is important that females as well as males with a family history of hemophilia B have their clotting factor activity levels checked.

Resources

National Hemophilia Foundation
http://www.hemophilia.org
Genetics Home Reference: Hemophilia
http://ghr.nlm.nih.gov/condition/hemophilia
World Federation of Hemophilia
http://www.wfh.org
Canadian Hemophilia Society
http://www.hemophilia.ca
Haemophilia Society
http://www.haemophilia.org.uk

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