Hemophilia-a genetic disease.

Domer Bach*

Department of Hematology, Fordham University, New York, United States

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Description

Hemophilia is a blood clotting condition that makes it feasible for blood to clot. Following an injury or surgery, people with this illness have persistent bleeding or oozing. Many proteins known as clotting factors are found in blood and can aid in the prevention of bleeding. Low levels of factor VIII (8) or factor IX are found in people with hemophilia.

Mechanism

Hemophilia A is an inheritable condition, which means it can be handed down from one generation to the next. Sex chromosomes are the X and Y chromosomes of a person. The X chromosome is where the hemophilia gene is found. Hemophilia is a recessive X-linked disorder. Females receive two X chromosomes from their mothers and one from their fathers (XX). Males have an X chromosome inherited from their mother and a Y chromosome inherited from their father. That means that if a boy receives an X chromosome through his mother with haemophilia, he will develop the disease. It also means that fathers cannot infect their sons with haemophilia. Daughters, on the other hand, have two X chromosomes, so even if they get the haemophilia gene from their mother, they will almost certainly inherit a healthy X chromosome from their father and will not develop haemophilia. A carrier is a daughter who receives an X chromosome that has the haemophilia gene. She has the ability to pass the gene on to her children. If a woman is heterozygous for haemophilia and the father is not, each kid has a one-in-two (50%) chance of inheriting his mother's haemophilia allele and developing haemophilia. Each daughter has a one-in-two (50%) chance of inheriting her mother's haemophilia allele and becoming heterozygous for the disease. Overall, each pregnancy has a one-in-four (25%) probability of producing a hemophilic son and a one-in-four (25%) chance of producing a heterozygous daughter. There is a 50% probability that the infant (either a son or a daughter) will not inherit the haemophilia allele and so will not be able to transmit it down to his or her children.

Signs and symptoms

The joints start oozing blood. It commonly affects the knees, elbows, and ankles, causing swelling, pain, and tightness in the joints. Bleeding into the skin (bruising) or muscle and soft tissue, resulting in a blood build-up in the area (called a hematoma). Bleeding in the mouth and gums, as well as bleeding that is difficult to stop after a tooth is lost. Circumcision-related bleeding (surgery performed on male babies to remove the foreskin, covering the head of the penis). An infant's head is bleeding after a tough delivery. Blood in the faeces or urine. Nosebleeds that is difficult to stop.

Cause

Hemophilia is caused by a mutation or change in one of the genes that gives instructions on how to make the clotting factor proteins that help blood clot. This alteration or mutation can cause the clotting protein to stop operating or go missing entirely.

Treatment

Hemophilia is effectively treated by replacing the missing blood clotting factor, which allows the blood to clot normally. Commercially produced factor concentrations are infused (delivered through a vein) to achieve this. People with haemophilia can learn to inject these infusions on their own to halt bleeding episodes and, by doing so on a regular basis (called prophylaxis), even prevent most bleeding episodes.

*Correspondence to

Domer Bach

Department of Hematology

Fordham University

New York

United States

Email: dobach@gamil.com