Hemophilia

Hemophilia is a disorder in which the blood does not clot properly, leading to extensive bleeding. The bleeding can be external or internal, especially in the knees, ankles, and elbows. There are two types of hemophilia: type A and type B. Hemophilia A is the most common form in which the protein required for clotting, factor VIII, is missing or deficient. Hemophilia B, also known as Christmas disease, is four times less common and results from a deficiency in the factor IX clotting agent. Both are genetic disorders. Individuals with either form of hemophilia may bleed longer from sustained injuries and may bleed internally into the joints and muscles, which can result in organ and tissue damage. Hemophilia is more likely to be found in men and rarely found in women; however, women can be gene carriers of the disease.

Statistics

- In the United States, about 20,000 people have hemophilia.
- About 80 percent of those with hemophilia have type A.
- Among U.S. born males, about one in 5,000 will have hemophilia.
- About 70 percent of those with hemophilia type A have the most severe form.

Risk Factors

Hemophilia is usually an inherited disease caused by a defect on a gene located on the X chromosome. This gene carries the information about the blood clotting factor VIII or IX within the body. Women who are carriers of the gene can pass it on through birth, and therefore, have a 25 percent chance of having a son with hemophilia. A man with hemophilia will pass the gene on to all of his daughters who will become carriers of hemophilia because the father's X chromosome determines female gender. However, none of his sons will have hemophilia since the male gender is determined by the Y chromosome.

Since women have two X chromosomes, even if one of the genes carries the defect for hemophilia, women still have an additional X chromosome that would carry the normal clotting factors to prevent her from having clotting issues; however, carriers can still have symptoms of hemophilia if she has low levels of clotting factors. Girls can be born with hemophilia if the father has hemophilia and the mother is a carrier. Some males are born with hemophilia because of a gene mutation, not because the mother is a carrier.

Symptoms

The symptoms of hemophilia A and B are the same and can include:

- Profuse bleeding
- A tendency to bruise easily
- A tendency to bleed very easily
- Excessive bleeding from wounds
- Nose bleeds
- Heavy menstrual bleeding
- Unexplained internal bleeding
- Bleeding from a wound after the wound has appeared to clot
- Blood in the urine or stool (sign of internal bleeding)
- Bleeding in the joints – key signs are joint tightness, swelling, hot to touch, and pain
- Bleeding in the brain – key signs are long-lasting headaches or neck aches or stiffness, vomiting, sleepiness, behavior changes, double vision, convulsions, seizures, sudden weakness, and difficulty walking

Treatment

While there is no cure for hemophilia, people with the blood disorder can enjoy a normal life with the proper treatment. The primary course of treatment for hemophilia is called replacement therapy in which the clotting factor is infused into the blood stream. Depending on the severity of the disorder, some patients may receive preventive replacement therapy or regular treatments that help prevent bleeding. Other patients may receive demand therapy in which infusions are given only as needed. Both types of treatments can be done at home. In mild cases of hemophilia A, the disorder may be treated with man-made hormones and other medicines.

Sources: National Heart, Lung and Blood Institute, National Hemophilia Foundation, and World Federation of Hemophilia

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