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Hemophilia

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

Statistics

- In the United States, **more than 30,000** people have hemophilia.
- Of those with the disease, hemophilia A represents **four times** as many patients as hemophilia B.
- Among U.S. born males, about **one in 5,000** will have hemophilia.
- About **60 percent** of those with hemophilia A have the most severe form.

Risk Factors

Hemophilia is usually inherited and is caused by a defect on a gene, located on the X chromosome, that affects the blood's clotting proteins. 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, a female carrier can still have symptoms of hemophilia if she has low levels of clotting factors. Although rare, girls can be born with hemophilia if both the father and mother pass down affected X chromosomes, or if one inherited X chromosome is affected and the other is inactive.

However, around 30 percent of babies born with hemophilia have no genetic history and are the first in their family to develop the gene mutation.

Symptoms

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

- Excessive bleeding, especially in the knees, ankles, and elbows
- A tendency to bruise easily
- A tendency to bleed very easily
- Excessive bleeding from wounds
- Frequent nosebleeds
- Heavy menstrual bleeding
- Excessive bleeding after childbirth
- Umbilical stump bleeding or after circumcision
- Unexplained internal bleeding
- Heavy bleeding after vaccinations
- Petechiae (less common) – small red, purple, or brown spots caused from bleeding under the skin
- Blood in the urine or stool (sign of internal bleeding)
- Bleeding in the muscles
- Bleeding in mouth and gums, excessive bleeding after tooth loss, or after having injections such as vaccinations
- Bleeding into infant head after arduous birth
- Bleeding in the joints – key signs are joint tightness, swelling, redness, and pain
- Bleeding in the brain – key signs are long-lasting headaches, seizures, or paralysis

Treatment

While there is no cure for hemophilia, people with the blood disorder can enjoy a normal life with the proper treatment. The primary mode 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.

About Texas Oncology

Texas Oncology is an independent private practice with more than 500 physicians and 210 locations across the state. Meeting the oncology needs of Texans for more than 35 years, the practice includes Texas Center for Proton Therapy, Texas Breast Specialists, Texas Oncology Surgical Specialists, Texas Urology Specialists, and Texas Center for Interventional Surgery. As a lead participant in US Oncology Research, Texas Oncology played a role in the development of more than 100 FDA-approved therapies. For more information, visit www.TexasOncology.com.

Sources: Centers for Disease Control and Prevention, National Heart, Lung and Blood Institute, National Hemophilia Foundation, and National Organization for Rare Disorders

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888-864-4226

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