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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 (8), 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 (9). Both are genetic, heritable 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 and chronic loss of function in the joints. 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

- Although exact numbers are not known, approximately **33,000** people live with hemophilia in the United States.
- About one in 5,000 males born in the United States will have hemophilia A.
- About **60%** of those with hemophilia A have the most severe form.

Risk Factors

Hemophilia is inherited in about two-thirds of cases, caused by a passed-down defect in the gene to create blood clotting factor VIII or IX, which is located on the X chromosome. This leads to a deficiency of the amount of factor VIII or IX in the blood, so the blood does not clot normally after an injury. Women (XX) who are carriers of the hemophilia gene on one of their X chromosomes can pass it on to their children, with each child having a 50% chance of inheriting the X chromosome that carries the hemophilia gene. A man (XY) with hemophilia will pass his X chromosome with the hemophilia gene to all of his daughters (XX), who will become carriers of hemophilia on one of their X chromosomes; none of his sons (XY) will have hemophilia, because sons get their X chromosome from their mother and their Y chromosome from their father. Sons (XY) will be born with hemophilia if they inherit an X chromosome that carries the hemophilia gene. Only rarely would a daughter (XX) be affected by hemophilia, because having the gene with hemophilia on one X chromosome and having a healthy gene on the other X chromosome usually results in being able to create enough clotting factor to allow the blood to clot normally. A daughter could have hemophilia if she receives a hemophilia gene on an X chromosome from both parents, or if one X chromosome is inactive.

However, around one-third of babies born with hemophilia have no genetic history and are the first in their family to develop the gene mutation. They can then pass it on to their children.

Symptoms and Signs

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

- Bleeding into the head as an infant after arduous birth
- Umbilical stump bleeding or after circumcision
- A tendency to bruise or bleed easily from minor injuries
- Frequent nosebleeds
- Heavy menstrual bleeding
- Excessive bleeding after giving birth
- Bleeding into the joints, especially the elbows, knees, and ankles
- Bleeding into the muscles after injections
- Petechiae: small red, purple, or brown spots caused from bleeding under the skin
- Blood in the urine or stool
- Bleeding in mouth and gums, such as excessive bleeding after having a tooth pulled
- Bleeding in the brain – key signs are long-lasting headaches, seizures, or paralysis

Treatment

While there is currently no permanent cure for hemophilia A or B, people with hemophilia can enjoy a normal life with proper treatment. The primary mode of treatment for hemophilia is called replacement therapy, in which replacement

clotting factor is infused into the blood stream. Depending on the severity of hemophilia, some patients may receive regularly scheduled replacement therapy to prevent bleeding; other patients may receive only on-demand therapy in which infusions are given as needed for injuries, surgeries, or procedures. Both types of treatments can be done at home. Mild cases of hemophilia A may also be treated with certain synthetic hormones and other medicines.

About Texas Oncology

Texas Oncology is an independent private practice with more than 525 physicians and 220 locations across the state and southeastern Oklahoma. 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, Texas Imaging & Infusion Center, 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, National Library of Medicine, and National Organization for Rare Disorders

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