Q. What is hemophilia?

A. Hemophilia is a rare type of bleeding disorder that occurs when there is a deficiency or absence of a particular protein (clotting factor) needed for blood to clot. As a result, a person with hemophilia will experience longer bleeding after an injury because the clot formed is not strong enough to stop the bleeding. A person with hemophilia will not bleed any faster than a person without hemophilia, but he or she will bleed slower and longer. This type of bleeding disorder is inherited and occurs almost exclusively in males.

Q. What is clotting factor? What does it do?

A. Clotting factor is a series of blood proteins that go to work after there’s an injury that damages blood vessels. When a blood vessel is torn, it tightens to reduce the flow of blood. Tiny cells in the blood called platelets stick together at the injury site to form a platelet “plug,” and thread-like strands called fibrin bind the platelets together and form a fibrin net. The fibrin net entrap the plug, which has developed into a blood clot through a series of interactions among certain proteins in the blood (clotting factor). There are thirteen clotting factors and they all work in sequence to help form a clot.

The type of clotting factor that is missing determines what kind of hemophilia you have. The two most common types are hemophilia A and hemophilia B. In hemophilia A, the missing clotting factor is factor VIII. Hemophilia A is sometimes referred to as classical hemophilia. In hemophilia B, the missing clotting factor is factor IX. Hemophilia B is sometimes referred to as Christmas disease.

Q. How does a person “get” hemophilia?

A. Hemophilia is an inherited disease, meaning that a gene responsible for causing hemophilia is passed from parent to offspring. In the United States, about one or two in every 10,000 male infants is born with it. Approximately 80 percent have hemophilia A, and 20 percent have hemophilia B. Hemophilia affects mostly males. The chances of having a child with hemophilia are the same for all racial and socioeconomic groups. Although females rarely have hemophilia, they may carry the gene that causes hemophilia and pass it on to their children.

Q. How is hemophilia inherited?

A. During fertilization, a baby will inherit one sex chromosome from each parent. A daughter will receive one X chromosome from her mother and another X chromosome from her father (XX), while a son will receive one X chromosome from his mother and one Y chromosome from his father (XY).

The gene that is responsible for making clotting factor is found only on the X chromosome. This is called a sex-linked gene. If the gene is abnormal, the result is hemophilia unless there is a normal gene on a matching X chromosome to offset the abnormal gene.

A male infant receives one X chromosome from his mother and one Y chromosome from his father. As a result, if the gene on the X chromosome that is responsible for making clotting factor is abnormal, that son will be born with hemophilia.

In females, a daughter will receive one X chromosome from her mother and one from her father. If the mother contributes an X chromosome with normal genes and her father passes on an X chromosome that carries the abnormal gene for hemophilia, then the daughter will not have hemophilia. However, the daughter will still “carry” the gene that causes hemophilia and may pass that abnormal gene on to her own children. Females who carry the gene for hemophilia but are not affected with hemophilia are called “carriers.”

The hemophilia gene can also occur in a male or a female as a result of spontaneous gene mutation. If this occurs, the abnormal gene may be passed from female “carriers” for several generations before hemophilia appears. Women with fathers who have hemophilia are often referred to as “obligate carriers.”
**Q. How does a woman know if she is a carrier?**

**A.** There are two ways to test a woman to tell if she is a hemophilia carrier. A doctor may send a small amount of blood to the laboratory to measure circulating factor VIII or factor IX levels. Many women who carry the gene for hemophilia have lower than normal levels of clotting factor circulating in their blood. A doctor will examine the blood tests and the woman’s family history to help determine her chances of being a carrier. This type of testing cannot be done when a woman is pregnant or taking birth control pills because these conditions can cause false readings.

The second way to determine if a woman is a carrier is through DNA analysis. This type of testing is more accurate. A small sample of blood will be needed from the woman, from a family member who has hemophilia (the woman’s parents and other relatives). The blood is then sent to a special laboratory. The doctor will examine the results of the DNA analysis and the family tree in order to provide answers.

There are some women who are considered definite carriers and do not necessarily need to be tested. These women include: women who have fathers with hemophilia, mothers who have more than one son with hemophilia, or mothers who have one child with hemophilia and who have at least one other blood relative with the disorder.

A woman with only one son who has hemophilia may or may not be a carrier because a genetic mutation could have occurred in the mother’s genes, or in one of her ancestors or in her son. If the mutation occurs on the son’s genes, the mother is not a carrier and her other children will not be affected with hemophilia.

**Q. What are the symptoms of hemophilia?**

**A.** The signs and symptoms of hemophilia can vary from person to person, but the hallmarks of the disorder are easy bruising and abnormal bleeding. Other common symptoms include swelling and pain from bleeding into joints, blood in the urine and stool, and nosebleeds with no apparent cause.

Most children with hemophilia are diagnosed early in life. A doctor may suspect hemophilia if a newborn experiences severe bruising after receiving shots that are given after a baby’s birth, or if an infant experiences prolonged or severe bleeding after a heel stick to obtain blood or circumcision. If a baby is not diagnosed at birth, other symptoms that may prompt a family to suspect there may be a problem include easy bruising in places that are not normally seen on infants, such as the face, back or stomach, delayed healing after an injury, or swollen joints and muscles. If there is a strong family history of hemophilia, a newborn should be tested as soon as possible to determine if the baby has the disorder.

**Q. How is hemophilia diagnosed?**

**A.** Hemophilia is diagnosed through a blood test to measure the factor VIII or factor IX levels that circulate in the blood. Factor VIII and factor IX levels are measured as a percentage. The average factor level in the population is 100% but a level between 50 and 150 % are considered to be normal.

**Q. What’s the difference between mild, moderate and severe hemophilia?**

**A.** Hemophilia is classified as mild, moderate and severe. The severity is dependent on the person’s factor level.

A person with mild hemophilia will have a factor VIII or IX level greater than 5 percent but less than 50 percent. People with mild hemophilia may have problems with bleeding only after serious injury, trauma, or surgery. Some people with mild hemophilia are not diagnosed until later in life, when a surgery, dental procedure or serious injury causes prolonged bleeding.

A person with moderate hemophilia will have a factor VIII or IX level between 1 and 5 percent. People with moderate hemophilia are more likely to experience prolonged bleeding after a slight injury. A person may have a bleeding episode one to two times per month. In general, the diagnosis is usually made earlier in life, especially if there is a family history of hemophilia.

A person with severe hemophilia will have a factor VIII or IX level less than 1%. A person with severe hemophilia can experience bleeding without an injury and may bleed one to two times per week. Approximately 60 percent of people with hemophilia have severe hemophilia. The diagnosis for severe hemophilia occurs relatively early in life.
Q. What is a bleeding episode? Does it hurt?

A. A bleeding episode occurs when a person with hemophilia experiences an injury to the tissues in the body, which results in bleeding. Without the correct amount of clotting factor, the fibrin net that is made is not strong enough to stay together and the clot falls apart, causing re-bleeding in the injured area.

Minor scrapes usually don't cause pain or cause a problem, but many bleeding episodes can result in considerable pain if they're not treated promptly. Immediate treatment is the best way to prevent pain and injury.

Bleeding episodes can be grouped according to severity:

A minor bleeding episode is any bleeding that is caught just as it starts. These types of bleeding episodes heal quickly and sometimes without much treatment.

A major bleeding episode is any bleeding caused by an injury that involves swelling and pain or any bleeding that cannot be stopped on its own. Major bleeding episodes include muscles and joints. Bleeding episodes should always be treated promptly to prevent damage to joints or other complications.

A severe bleeding episode is a potentially life threatening injury or bleeding that can cause permanent damage or death. Severe bleeding episodes may include bleeding from surgery or dental procedures, deep muscle bleeding, bleeding inside the head, abdominal bleeding, or bleeding around the throat or neck.

Q. How is hemophilia treated?

A. The standard treatment for hemophilia is to give an infusion of factor VIII or factor IX concentrate into the veins. This is known as factor replacement therapy. Once the factor is infused, the normal clotting events can occur and a fibrin net and clot will be formed. This type of treatment is often referred to as "on-demand factor replacement therapy" and has been used for several years.

A common type of factor replacement therapy is a type of preventive care known as prophylaxis. Prophylaxis is the infusion of clotting factor at scheduled times to maintain higher factor VIII or factor IX levels in order to prevent spontaneous bleeding and decrease the number of bleeding episodes. The purpose of this therapy is to prevent bleeding that causes joint damage. This type of therapy is commonly used for children who have severe hemophilia and significantly improves the child’s quality of life because the child has fewer bleeding episodes (and possibly none) and no permanent joint damage.

Although there is no cure for hemophilia, with proper treatment, most people with hemophilia lead full, active lives.

Q. Are there any other medications that can help stop or prevent bleeding?

A. Before surgery or dental work, people with mild hemophilia are sometimes prescribed a drug called desmopressin acetate (commonly know as DDAVP). This synthetic hormone stimulates the release of existing factor VIII stores. It can be given through a vein (DDAVP) or via a nasal spray (Stimate).

Your doctor may also prescribe a medication known as an anti-fibrinolytic agent. This drug, commonly called Amicar (aminocaproic acid) slows the normal breakdown of blood clots and is helpful for treating oral bleeding.

People with hemophilia should avoid drugs that can exacerbate bleeding problems, such as aspirin, heparin, warfarin, and nonsteroidal anti-inflammatory drugs (NSAIDs).
Q. Does a person with hemophilia have to go to the hospital every time he or she needs treatment?

A. No. Years ago, people with hemophilia had to rely on hospitals and clinics each time they needed clotting factor, which meant frequent absences from school and work and long hours spent in emergency rooms or clinics. Today, through the support of hematologists, hemophilia treatment centers, local and national hemophilia foundations, and specialized home care companies, people with hemophilia and their families can learn to recognize bleeding episodes early and infuse their own factor at home. This has decreased the amount of time families have had to spend in emergency rooms and clinics and has contributed to an increased quality of life. Children with hemophilia today have fewer hospitalizations, spend more time in school, are better integrated into their peer groups, and experience significantly less joint damage when compared to previous generations of hemophiliacs.

Home infusion therapy has allowed many families with hemophilia to lead a more normal lifestyle and has also improved the long-term prognosis for adult hemophilia patients, according to Haemophilia journal. Home infusion, it reported, "significantly decreased hospital admissions for bleed complications and decreased pain, dysfunction and long-term disability."

Of course, along with the freedom home infusion therapy offers, comes responsibility. For all its benefits, experts caution that hemophilia home infusion is complicated and time-consuming, and might not be for everyone.

However, by receiving ongoing education and support from your hemophilia team, home infusion therapy will put you on the frontline as your child’s caregiver and insure that your child receives the best care possible.

Q. Does having hemophilia restrict a person’s lifestyle?

A. For the most part, living with hemophilia today is much better than it was 30 to 40 years ago. New and improved treatments allow most people with hemophilia to lead normal, active lives. Research has shown that being physically active is extremely important for a person with hemophilia because the benefits of exercise can actually reduce or prevent bleeding episodes by strengthening the muscle surrounding vulnerable joints. In fact, the National Hemophilia Foundation strongly encourages regular exercise for people with hemophilia of all ages in order to preserve joint function.

Of course, there are some activities that people with hemophilia should avoid. According to a report published in Clinical Orthopaedics and Related Research, some sports such as boxing, rugby, and football are considered too high risk and should be avoided. Hematologists who specialize in hemophilia care recommend that people with severe hemophilia avoid high contact sports like football, wrestling, and other sports that are considered high risk for head or severe injury.

However, there are many different types of sports and activities that a person with hemophilia can safely participate in. Some of the most widely recommended activities include swimming, walking, golfing, and bicycling. People with hemophilia are strongly encouraged to talk with their hemophilia team about the best activities to participate in, so that they can maintain a healthy and active lifestyle as much as possible.

-- The article was reviewed by Kim Schafer, a pediatric hemophilia nurse at the hematology/oncology division of the Department of Pediatrics at the University of California at Davis, a nationally recognized Hemophilia Treatment Center (HTC).

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