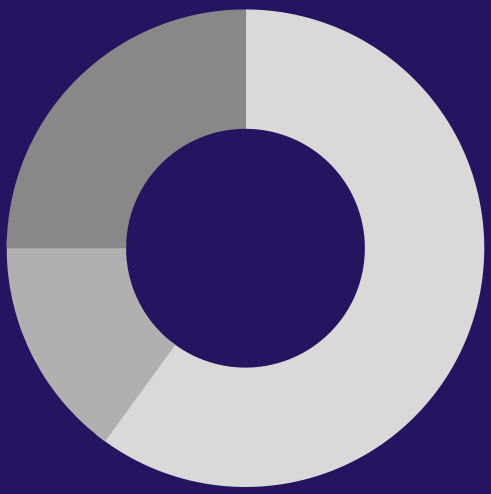
HEMOPHILIA ASSOCIATION OF NEW JERSEY

Hemophilia Fast Facts

There are between 30,000 - 33,000 people living with hemophilia in the US



60%
have severe hemophilia

25%
have moderate hemophilia

15%
have mild hemophilia

The most common symptoms of hemophilia are:

Bleeding into joints

Prolonged bleeding from cuts or injuries

Bruising

Nosebleeds

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What is Hemophilia?

Hemophilia is a rare genetic bleeding disorder caused by a shortage of certain clotting factors. Blood-clotting factors are needed to help stop bleeding after a cut or injury and to prevent unplanned bleeding. The hemophilia gene can contain many different errors, leading to different degrees of abnormality in the amount of clotting factor produced.

There are two types of hemophilia. Hemophilia A is caused by a deficiency of active clotting factor VIII. Approximately 80 percent of all people with hemophilia have type A, and most of these cases are severe. Approximately 1 out of every 5,000 male babies is born with hemophilia A. Hemophilia B (Christmas Disease) is caused by a lack of active clotting factor IX. It is less common, occurring in 1 out of every 30,000 male babies.

What causes the disease?

Hemophilia A and B are caused by an inherited defect in a pair of chromosomes. The defect affects how much clotting factor a person will produce and how the factor will function. The less normal the function and amount of clotting factor, the more severe a case of hemophilia can be.

What are the symptoms of hemophilia?

Symptoms of hemophilia are generally first noticed during infancy or childhood. However, some people with milder forms of the disease may not develop symptoms until later. The following signs of hemophilia may be noticed shortly after birth:

- Bleeding into the muscle, resulting in a deep purple bruise after receiving a routine Vitamin K shot.
- Prolonged bleeding after a male child is circumcised.

Other symptoms include:

- Bleeding into a joint or muscle that causes pain and swelling.
- Abnormal bleeding after an injury or surgery.
- Easy bruising.
- Bleeding after dental work.

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How is Hemophilia Diagnosed?

Blood tests can help determine whether your child has hemophilia. Hemophilia is usually classified by its severity. There are three levels of hemophilia. The severity of the disease is defined by how much clotting factor is produced and in what situations bleeding most often occurs.

Mild hemophilia factor levels > 5 % might not be recognized unless there is excessive bleeding after a major injury or surgery.

Moderate hemophilia factor levels 1 - 5 % bleeding usually follows a fall, sprain or strain.

Severe hemophilia factor levels< 1 % bleeding may occur one or more times a week for no apparent reason.

The percentage of clotting factor in the blood remains the same throughout a person's life. All family members who have hemophilia will have similar forms.

What is the treatment for hemophilia?

Most people with hemophilia can successfully manage their bleeding problems with clotting factor replacement therapy. Clotting factors may be injected when needed or on a regular basis to prevent bleeding episodes. On demand therapy is used before participating in activities with high risk for injury or once it is suspected that bleeding has begun.

TREATMENT OF HEMOPHILIA

For many people, treatment products are injected, or infused, to stop bleeds or to prevent bleeds. There are two main ways to treat hemophilia.

Episodic care is used to stop a bleeding episode when it occurs.

Prophylacticic care is used to prevent bleeding episode from occurring.

Source: CDC

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What is von Willebrand disease (VWD)?

Von Willebrand disease (VWD) is a bleeding disorder. People with VWD have a problem with a protein in their blood that helps control bleeding. They do not have enough of the protein or it does not work the way it should. It takes longer for blood to clot and for bleeding to stop.

There are different types of VWD. All are caused by a problem with the von willebrand factor (VWF) protein when a blood vessel is injured and bleeding occurs. VWF helps cells in the blood, called platelets, mesh together and form a clot to stop the bleeding. VWD is the most common bleeding disorder that people have. It affects both males and females. VWD is generally less severe than other bleeding disorders. Many people with VWD may not know that they have the disorder because their bleeding symptoms are very mild. For most people with VWD, the disorder causes little or no disruption to their lives except when there is a serious injury or need for surgery. However, with all forms of VWD, there can be bleeding problems.

How do people get VWD?

VWD is usually inherited. It is passed down through the genes from either parent to a child of either sex. Sometimes there is evidence of a family history of bleeding problems. However, bleeding symptoms can vary a lot within a family. Sometimes there is no family history and VWD occurs due to a spontaneous change in the VWD gene before the baby is born.

What are the symptoms?

The main symptoms of VWD are:

- Easy bruising
- Frequent or prolonged nosebleeds
- Bleeding from the gums
- Prolonged bleeding from minor cuts
- Heavy or prolonged menstrual bleeding
- Bleeding in the upper and lower gastrointestinal tract
- Prolonged bleeding following injury, surgery, dental work, or childbirth

Many people with VWD have few or no symptoms. People with more or serious VWD may have more bleeding problems. Symptoms also change over time. Sometimes VWD is discovered only when there is heavy bleeding after a serious accident or a dental procedure.

More women than men show symptoms of VWD. Women with VWD often bleed more, or longer than normal, with menstruation and following childbirth. Some women with VWD have a lot of menstrual pain or irregular menstruation.

Blood type can play a role. People with Type O blood often have lower levels of VWF than people with Types A, B or AB. This means people with VWD and Type O blood may have more problems with bleeding.

How is VWD Diagnosed?

VWD is not easy to diagnose. People who think they have a bleeding problem should see a hematologist who specializes in bleeding disorders. Proper tests can be done at a bleeding disorder treatment center. Since the VWF protein has more than one function, more than one lab test should be used to diagnose VWD.

Laboratory testing for VWD is also difficult. VWD cannot be diagnosed with routine blood tests. Testing involves measuring a person's level and activity of VWF, and that of another blood clotting protein, factor VIII (FVIII). Testing is often repeated because a person's VWF and FVIII levels can vary at different times.

Different types of VWD

There are three main types of VWD. Within each type of VWD the disorder can be mild, moderate, or severe. Bleeding symptoms can be quite variable within each type depending in part on the VWF activity. It is important to know which type of VWD a person has, because treatment is different for each type.

Type 1 VWD is the most common form. People with Type 1 have lower than normal levels of VWF. Symptoms are usually very mild. Still, it is possible for someone with Type 1 VWD to have serious bleeding.

Type 2 VWD involves a defect in the VWF structure. The VWF protein does not work properly, causing lower than normal VWF activity. There are different Type 2 VWD defects. Symptoms are usually moderate.

Type 3 VWD is usually the most serious form. People with Type 3 VWD have very little or no VWF. Symptoms are more severe. People with Type 3 VWD can have bleeding into muscles and joints, sometimes without injury.

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How is VWD treated?

VWD can be treated with a synthetic drug called desmopressin, a clotting factor concentrate that contains VWF, or other drugs that help control the bleeding. The type of treatment depends in part on the type of VWD a person has. People with mild forms of VWD often do not require treatment for the disorder except for surgery or dental work.

Desmopressin is generally effective for treating Type 1 VWD, and helps prevent or treat bleeding in some forms of Type 2 VWD. It is used to control bleeding in an emergency or during surgery. It can be injected or taken by nasal spray, and raises VWF and FVIII levels to help blood clot. Desmopressin does not work for everyone. A doctor needs to do tests to find out if an individual responds to the drug. Ideally, tests should be done before treatment is needed.

Factor concentrates are used when desmopressin is not effective or when there is a high risk of major bleeding. Factor concentrates contain VWF and FVIII. This is the preferred treatment for Type 3 VWD, most forms of Type 2 VWD, and for serious bleeding and major surgery in all types of VWF. Bleeding in mucus membranes (inside the mouth, nose, intestines, or womb) can be controlled by drugs such as tranexamic acid (Cyklokapron), aminocaproic acid (Amincar), or by fibrin glue. However, these products are used to maintain a clot, and do not actually help form a clot.

Hormone treatment, such as oral contraceptives, help to increase VWF and FVIII levels and control menstrual bleeding. If hormone treatment is not prescribed, antifibrinolytic agents may be effective for treating heavy menstruation.
These treatments may have side effects, so people with VWD should talk to their physician about possible side effects of treatment.

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Hemophilia A. Factor VIII Deficiency

Hemophilia A is a hereditary bleeding disorder caused by a lack of blood clotting factor VIII. Without enough factor VIII, the blood cannot clot properly to stop bleeding

Causes, incidence, and risk factors

Hemophilia A is caused by an inherited X linked recessive trait, with the defective gene located on the X chromosome. Females have two copies of the X chromosome, so if the factor VIII gene on one chromosome doesn't work, the gene on the other chromosome can do the job of making enough factor VIII. Males, however, have only one X chromosome, so if the factor VIII gene on that chromosome 13 is defective, they will have hemophilia A. Thus, most people with hemophilia A are male. If a woman has a defective factor VIII gene, she is considered a carrier. This means the defective gene can be passed down to her children. In a woman who carries the defective gene, any of her male children will have a 50% chance of having hemophilia A, while any of her female children will have a 50% chance of being a carrier. All female children of men with hemophilia carry the defective gene. Genetic testing is available for concerned parents.

FACTS ABOUT HEMOPHILIA

There are many different types of hemophilia.
The most common types are:

Hemophilia A:
people with low levels of clotting factor VIII (8)

Hemophilia B:
people with low levels of clotting factor IX (9)

Source: CDC

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Hemophilia B. Factor IX Deficiency

Hemophilia B is a hereditary bleeding disorder caused by a lack of blood clotting factor IX. Without enough factor IX, the blood cannot clot properly to stop bleeding.

Causes, incidence, and risk factors

Hemophilia B is caused by an inherited X linked recessive trait, with the defective gene located on the X chromosome. Females have two copies of the X chromosome, so if the factor IX gene on one chromosome doesn't work, the gene on the other chromosome can do the job of making enough factor IX. Males, however, have only one X chromosome, so if the factor IX gene on that chromosome is defective, they will have hemophilia B. Thus, most people with hemophilia B are male. If a woman has a defective factor IX gene, she is considered a carrier. This means the defective gene can be passed down to her children. In a woman who carries the defective gene, any of her male children will have a 50% chance of having hemophilia B, while any of her female children will have a 50% chance of being a carrier. All female children of men with hemophilia carry the defective gene. Genetic testing is available for concerned parents.

HEMOPHILIA B FACTS

5%

Inhibitors occur in approximately 5% of patients with hemophilia B

75%

Moderate and severe accounts for 75% of the Hemophilia B pool

Around 3 in 100

Individuals with hemophilia B produce an antibody to the factor IX

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Symptoms of Hemophilia

The severity of symptoms varies. Bleeding is the main symptom of the disease and sometimes, although not always, occurs if an infant is circumcised. Additional bleeding problems are seen when the infant starts crawling and walking. Mild cases may go unnoticed until later in life when they occur in response to surgery or trauma. Internal bleeding may happen anywhere, and bleeding into joints is common. Symptoms may include:

- Bleeding into joints, with associated pain and swelling
- Bruising
- Prolonged bleeding from cuts, tooth extraction, and surgery

Treatment of Hemophilia

Standard treatment involves replacing the missing clotting factor. The amount of factor concentrates needed depends on the severity of the hemophilia, severity of the bleeding, the site of the bleeding, and the size of the patient. People with severe/moderate forms of the Hemophilia may need regular preventive treatment. To prevent a bleeding crisis, families of people with hemophilia can be taught to give factor concentrates at home at the first signs of bleeding. Depending on the severity of the disease, DDAVP or factor VIII concentrate may be given before having dental extractions or surgery in order to prevent bleeding People with mild hemophilia may be treated with desmopressin (DDAVP), which helps the body release factor that is stored within the lining of blood vessels.

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Prognosis of Hemophilia

The outcome for hemophiliacs is usually good with treatment. Most people with hemophilia are able to lead relatively normal lives. Patients with hemophilia should establish regular care with a hematologist, especially one who is associated with a hemophilia treatment center.

Diagnostic Tests

If the patient is the first person in the family to have a suspected bleeding disorder, he will undergo a series of tests called a coagulation study. Tests to diagnose hemophilia include:

- Prolonged Partial Thromboplastin Time (PTT)
- Normal Prothrombin Time (PT)
- Normal bleeding time
- Normal fibrinogen level
- Low serum factor activity

DIAGNOSTIC TESTS

If the patient is the first person in the family to have a suspected bleeding disorder, he will undergo a series of tests called a coagulation study.

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The Hemophilia Association of New Jersey is dedicated to improving the lives of people with bleeding disorders and their families by providing access to resources, education, and advocacy.

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HANJ is a great resource for all of your questions related to your bleeding disorder. Our website has news, updates, as well as educational program and event information. Scan the QR code to view our website.