

HEMOPHILIA FACT SHEET



In each person's body, a number of blood proteins, also known as "clotting factors," work with platelets in a series of biological steps to stop bleeding when a blood vessel becomes injured. If one of these clotting factors is missing or deficient, then the process of clot formation is hampered, and prolonged bleeding results. To have a missing or deficient amount of factor VIII (FVIII) protein is also known as **hemophilia A**. When the deficient protein involved is factor IX (FIX), that is known as **hemophilia B**.

Symptoms & Severity

Hemophilia A and B are conditions that have a varied degree of severity. While bleeds can still occur across severities, the frequency of bleeding episodes is related to an individual's clotting factor level. This level indicates the amount of clotting factor present in a person's blood. Individuals who have a factor level of less than 1% of normal are classified as having **severe hemophilia**. To have a factor level between 1-5% of normal is considered **moderate hemophilia** (normal being between 50-150%), and anyone with a FVIII or FIX level between 6 to 49% of normal is someone with **mild hemophilia**.

Some more common symptoms of hemophilia are easy bruising, bodily swelling, frequent nosebleeds, excessive bleeding after dental procedures, and frequent heavy menstrual periods among women. Bleeding can also be associated with pain, as is commonly the case when it occurs in the joints or muscles. If left untreated, repeated bleeds into a joint can lead to chronic pain and joint deterioration.

While bleeds can occur due to a traumatic event (i.e., injury, dental work, childbirth, surgery), prolonged bleeding episodes may be caused by minor injuries or arise spontaneously without clear cause.

Inheritance

Hemophilia can occur in men and women across all racial and ethnic groups. As a X-linked genetic disorder, hemophilia is commonly passed down from females who are "carriers" of the affected gene to their offspring. With each birth, a female hemophilia carrier will have a 1 in 4 chance of having a son with hemophilia.

A male with hemophilia will contribute an affected X-chromosome to his daughter, resulting in all daughters of a father with hemophilia being carriers.

Approximately one-third of all hemophilia cases are the result of a spontaneous gene mutation with no prior family history.

Currently, there are about 20,000 and as many as 33,000 people living with hemophilia A and B in the United States [1].

1 www.cdc.gov/ncbddd/hemophilia/facts.html

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Current Treatments

A wide range of biopharmaceutical products are used to treat people with hemophilia A and B. Various advanced technologies are utilized to manufacture these products and clear differences exist in how these products may be delivered into one's body.

Factor replacement therapy is a long-standing hemophilia treatment whereby the specific deficient clotting protein (FVIII or FIX) is replenished through an intravenous infusion. These factor replacement products are either sourced from human plasma or manufactured using DNA recombinant technology.

Non-factor therapy restores a person's blood clotting process without the need to replace the FVIII or FXI protein directly. These are monoclonal antibody therapies that work by activating other related proteins in the coagulation process. This type of treatment is delivered by an injection under the skin.

Gene therapy is a one-time FDA-approved treatment to deliver a working copy of the hemophilia A or B gene to correct the disease and prevent bleeding symptoms for a sustained period.

Today, individuals living with hemophilia benefit from preventative treatment to reduce bleeding episodes. Improved longer-acting therapies, comprehensive care, regular exercise, and physical therapy have also greatly improved health and quality of life.

Complications

A serious complication of hemophilia A or B is the development of an inhibitor (or antibody) to treatment. This occurs when an individual's own immune system no longer accepts the current therapy. Additional treatment options exist, and individuals experiencing an inhibitor may be prescribed a non-factor therapy (or some combination of products) to tolerize their body's immune system to an alternative therapy.

Older-aged individuals living with hemophilia and HIV or hepatitis viruses also face challenging treatment considerations in managing their other more prevalent age-related diseases (such as heart, liver, or cancer).

Links to additional resources:

To view a complete list of all available products to treat people with hemophilia -

www.hemophilia.org/healthcare-professionals/guidelines-on-care/products-licensed-in-the-us

To read more about future therapies for hemophilia - www.hemophilia.org/bleeding-disorders-a-z/treatment/future-therapies

To find a chapter organization near you - www.hemophilia.org/community-resources/resources-near-you/chapter-network, and to find a hemophilia treatment center - dbdgateway.cdc.gov/HTCDirSearch.aspx. For additional information or resources, contact HANDI.