

# What is Hemophilia

## About Bleeding Disorders

Hemophilia is a genetic bleeding disorder that prevents the blood from clotting normally. The main symptom is uncontrolled, often spontaneous bleeding. Internal bleeding into the joints can result in pain, swelling and, if left untreated, can cause permanent damage

Hemophilia results from the deficiency of one more proteins known as blood clotting factors. There are 13 clotting proteins in the body, all of which contribute to the formation of a clot. The two main forms are hemophilia A (factor VIII deficiency) and hemophilia B (factor IX deficiency). Hemophilia occurs in 1 in 5,000 live male births, of these 80% are hemophilia A and 20% are hemophilia B. The worldwide incidence of hemophilia is estimated at more than 400,000 people. Approximately 70% of people around the world do not have access to treatment.

- Currently, there is no cure for hemophilia. While treatment exists, it is costly and may require lifelong infusion of replacement clotting factor that is manufactured either from human plasma or using recombinant technology. Hemophilia occurs predominately in males. Women are carriers of the defective gene and may experience mild symptoms. In about one-third of cases there is no known family history of hemophilia. Instead, the disorder results from a spontaneous genetic mutation.

- Nearly 90% of Americans with severe hemophilia became infected with HIV in the 1980s when the nation's blood supply was contaminated by blood pooled from people infected by HIV/AIDS. More than 50% of people with hemophilia infected with HIV have died. Since 1986, there have been no reported cases of HIV transmission through factor concentrates in the U.S. Current donor screening measures and improved viral inactivation methods have been integrated into the manufacturing process to dramatically improve the safety of these plasma-derived products.

von Willebrand disease is another genetic bleeding disorder that prevents the blood from clotting normally. It is caused by a deficient or defective blood protein known as von Willebrand factor. It occurs with equal frequency in men and women. It is estimated to affect more than two million people in the U.S. Of the three main types, type I (the mildest form of the disease) accounts for 70% of cases. Symptoms include frequent nosebleeds, a tendency to bruise easily, and excessive bleeding following surgery. In women, the disease may also cause heavy, prolonged bleeding during menstruation and excessive bleeding following childbirth. VWD is often undiagnosed or incorrectly attributed to a gynecologic condition.

## About Clotting Disorders

Clotting disorders are conditions in which the blood clots excessively. More than 600,000 Americans are affected by abnormal blood clots. People with these conditions also have the potential to develop dangerous clots, known as deep vein thrombosis or DVT. If left undiagnosed or untreated, life-threatening complications can occur. Approximately 146,000 people are affected by DVTs each year.

More than 11 million people in the U.S. have one of several inherited clotting disorders, known as thrombophilia. Factor V Leiden is the most common inherited form of thrombophilia.

Not everyone who has thrombophilia will experience a blood clot. The development of a blood clot is called thrombosis, which is a common medical problem. Some people only need treatment when recovering from surgery, during pregnancy or when immobile for long periods in a car or airplane. Others need to take anti-clotting medications for their entire lives.

For further information, please visit the Indiana Hemophilia and Thrombosis Center website at [www.ihtc.org](http://www.ihtc.org) and National Hemophilia Foundation [www.Hemophilia.org](http://www.Hemophilia.org)