A bleeding disorder is a condition in which a person tends to bleed longer (not faster) before a blood clot forms to stop the bleeding. Hemophilia is an inherited bleeding disorder in which the blood does not clot properly. Currently, an estimated 20,000 people in the United States have hemophilia. Read below for more information about the cause, diagnosis, and treatment of hemophilia.

What Is Hemophilia?
Hemophilia is a bleeding disorder in which one of the proteins that cause the blood to clot (called clotting factors) is missing or there isn’t enough of it to work properly. The lower the factor level, the more serious the hemophilia.

There are different types of hemophilia:
- hemophilia A (sometimes called classic hemophilia), which is caused by a lack or deficiency of clotting factor VIII
- hemophilia B (sometimes called Christmas disease), which is caused by a lack or deficiency of clotting factor IX

Hemophilia A and B are the most common types of hemophilia, with hemophilia A being four times as common as hemophilia B. Also, more than half of patients with hemophilia A have the severe form of hemophilia.

What Happens When a Person Has Hemophilia?
People with hemophilia often bleed longer than other people. Bleeds can occur internally, into joints and muscles, or externally, from minor cuts, dental procedures, or trauma. Internal bleeding into the joints can cause pain and swelling. If left untreated, it can lead to permanent damage.

People with hemophilia can’t make a firm fibrin clot. The first two steps to stop bleeding usually work normally in a person with hemophilia: the blood vessel narrows and the platelets make a plug. A person with hemophilia has a problem when a fibrin clot is needed to stop the bleeding. He or she does not have enough of certain clotting factors to do the job. Because of this, the fibrin clot is not made or is so thin that it’s not strong enough to stop the bleeding.

Symptoms of hemophilia include:
- Bleeding into joints, muscles and soft tissues
- Excessive bruising
- Prolonged, heavy menstrual periods (menorrhagia)
- Unexplained nosebleeds
- Extended bleeding after minor cuts, blood draws, vaccinations, surgery, or dental procedures

How is Hemophilia Diagnosed?
Hemophilia is an inherited blood disorder, meaning it is passed from the parent to the child through mutations (alterations in the DNA sequences) in genes. The gene that causes hemophilia is carried by women. However, in about one-third of cases, there is no known family history of hemophilia; instead, the disorder results when a spontaneous mutation occurs.

A medical health history is important to help determine if other relatives have been diagnosed with a bleeding disorder or have experienced symptoms. Tests that evaluate clotting time and a patient’s ability to form a clot may be ordered. A clotting factor test, called an assay, will determine the type of hemophilia and its severity.

The frequency and severity of a person’s bleeds depends on the amount of clotting factor in the plasma. The TABLE below outlines this relationship.

<table>
<thead>
<tr>
<th>Severity</th>
<th>Level of Factor VIII or IX in the Blood</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal (person who does not have hemophilia)</td>
<td>50%-100%</td>
</tr>
<tr>
<td>Mild hemophilia</td>
<td>6%-30%</td>
</tr>
<tr>
<td>Moderate hemophilia</td>
<td>1%-5%</td>
</tr>
<tr>
<td>Severe hemophilia</td>
<td>Less than 1%</td>
</tr>
</tbody>
</table>

How is Hemophilia Treated?
The goal of hemophilia treatment is to prevent bleeds and the damage associated with it.

Because many bleeding disorders are due to a lack of blood clotting factors, the mainstay of treatment is replacement of the deficient clotting factor. This is done by injecting commercially prepared clotting factor concentrates into a person’s vein. The two main types of clotting factor concentrates available are:
- plasma-derived factor concentrates
- recombinant factor concentrates

Unlike human plasma-derived factor products, recombinant factor products are developed in a lab through the use of DNA technology.

Other agents given to individuals with hemophilia include desmopressin acetate (DDAVP, a synthetic form of arginine vasopressin, the naturally occurring hormone that helps to release FVIII from where it is stored in the body’s tissues) and aminocaproic acid and tranexamic acid (chemicals that prevent clots from breaking down, resulting in a firmer blood clot). Usually these treatments are given orally or by nasal spray. Some of them are available in injectable forms.

Often the best place for patients with hemophilia to be diagnosed and treated is at one of the 130 or so federally funded hemophilia treatment centers (HTCs) that are spread throughout the country. HTCs provide comprehensive care from skilled hematologists and other professional staff, including nurses, physical therapists and social workers, and sometimes dentists, dieticians, and other health-care providers.

NHF Resources
The National Hemophilia Foundation (NHF) is dedicated to finding better treatments and cures for inheritable bleeding disorders and to preventing the complications of these disorders through education, advocacy, and research.

- Visit the NHF website (www.hemophilia.org) or their Steps for Living website (stepsforliving.hemophilia.org) for more information on living with a bleeding disorder.
- Patients or caregivers can also contact HANDI, NHF’s information resource center on hemophilia and other bleeding disorders, by emailing handheld@hemophilia.org or calling 1.800.42.HANDI.
- You can also find articles on hemophilia in HemAware, the NHF’s award-winning magazine, available at www.hemaware.org.