



Home → Health Topics → Hemophilia

URL of this page: <https://medlineplus.gov/hemophilia.html>

Hemophilia

Also called: Christmas disease, Factor IX deficiency, Factor VIII deficiency

What is hemophilia?

Hemophilia is a rare bleeding disorder [<https://medlineplus.gov/bleedingdisorders.html>] in which the blood does not clot properly. This can lead to problems with bleeding too much after an injury or surgery. You can also have sudden bleeding inside your body, such as in your joints, muscles, and organs.

Your blood contains many proteins called clotting factors that can help form clots to stop bleeding. People with hemophilia have low levels of one of these factors, usually either factor VIII (8) or factor IX (9). How severe the hemophilia is depends on the amount of factor in the blood. The lower the amount of the factor, the more likely it is that bleeding could happen and might lead to serious health problems.

What are the types of hemophilia?

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

- Hemophilia A (classic hemophilia), which is caused by a lack or decrease of clotting factor VIII (8)
- Hemophilia B (Christmas disease), which is caused by a lack or decrease of clotting factor IX (9)

What causes hemophilia?

Most types of hemophilia are inherited. They are caused by change in one of the genes [<https://medlineplus.gov/genetics/condition/hemophilia/>] (also called a mutation) that provides instructions for making the clotting factor proteins. The change may mean that the clotting proteins don't work properly or that they are missing altogether.

These genes are on the X chromosome. You may have one or two X chromosomes:

- People who are born male have one X chromosome (from the mother) and one Y chromosome (from the father). They can get hemophilia if their one X chromosome has the gene change.
- People who are born female have two X chromosomes, one from the father and one from the mother. They usually only get hemophilia if:
 - Both X chromosomes have the gene change **OR**
 - One X chromosome has the gene change and the other X chromosome is missing or inactive.

People who are born female who have the gene change on one X chromosome are a "carrier" of hemophilia.

Sometimes they may have some symptoms of hemophilia. They can pass the gene change on to their children.

Hemophilia that is not inherited is called acquired hemophilia. It is rare. It happens when your body makes specialized proteins called autoantibodies that attack and disable a clotting factor. This can happen because of pregnancy, immune system disorders [<https://medlineplus.gov/immunesystemanddisorders.html>] , cancer, or allergic reactions [<https://medlineplus.gov/drugreactions.html>] to certain medicines. Sometimes the cause is unknown.

Who is at risk for hemophilia?

Hemophilia is much more common in people who were born male since they can get it with a change to the gene on one X chromosome. People who have a family history of hemophilia are also at higher risk.

What are the symptoms of hemophilia?

The signs and symptoms of hemophilia are:

- Bleeding into the joints. This can cause swelling and pain or tightness in the joints. It often affects the knees, elbows, and ankles.
- Bleeding into the skin (which is bruising [<https://medlineplus.gov/bruises.html>]).
- Bleeding into the muscle and soft tissue, which can cause a build-up of blood in the area (called a hematoma).
- Bleeding of the mouth and gums, including bleeding that is hard to stop after you lose a tooth.
- Bleeding after circumcision [<https://medlineplus.gov/circumcision.html>] .
- Bleeding after having shots, such as vaccinations [<https://medlineplus.gov/vaccines.html>] .
- Bleeding in the head of an infant after a difficult delivery.
- Blood in the urine or stool.
- Frequent and hard-to-stop nosebleeds.

In some cases, severe hemophilia may cause bleeding in the brain. This may cause brain damage and can be life-threatening.

How is hemophilia diagnosed?

To find out if you have hemophilia, your health care provider will:

- Ask about your medical history, including your symptoms and other health conditions you may have.
- Ask about your family history, to find out if you have relatives who have or had hemophilia.
- Do a physical exam to look for signs of hemophilia, such as bruising.
- Do certain blood tests to show if your blood is clotting properly. If it does not, then you will have clotting factor tests [<https://medlineplus.gov/lab-tests/coagulation-factor-tests/>] to diagnose the cause of the bleeding disorder. These blood tests would show the type of hemophilia and the severity.

There is genetic testing for the factor VIII (8) and factor IX (9) genes. This testing may be used in people who have a family history of hemophilia to:

- Identify people who are carriers before they make decisions about pregnancy
- Test a fetus for hemophilia during pregnancy
- Test a newborn for hemophilia

What are the treatments for hemophilia?

The best way to treat hemophilia is to replace the missing clotting factor so that your blood can clot properly. This is usually done by injecting replacement clotting factor into a vein. The replacement clotting factor may be made from donated human blood. Or it may be made in a lab; this kind is called a recombinant clotting factor.

Replacement clotting factor can help treat a bleeding episode. In more severe cases of hemophilia, you might get the factor on a regular basis to prevent bleeding. You can learn how to inject the factor so that you can do it yourself at home.

There are other medicines to treat hemophilia. They may work by releasing factor VIII (8) from where it is stored in the body tissues, replacing the function of factor VIII (8), or preventing clots from breaking down.

If bleeding has damaged your joints, physical therapy may help them function better.

Good quality medical care from healthcare professionals who know a lot about the disorder can help prevent some serious problems. Often the best choice for care is to visit a hemophilia treatment center (HTC) [<https://www.cdc.gov/hemophilia/treatment/treatment-centers.html>] .

Start Here

- About Hemophilia [<https://www.cdc.gov/hemophilia/about/>] (Centers for Disease Control and Prevention)
- About Hemophilia [<https://www.genome.gov/Genetic-Disorders/Hemophilia>]  (National Human Genome Research Institute)
- Hemophilia [<https://elearning.wfh.org/elearning-centres/hemophilia/>] (World Federation of Hemophilia)
Also in Spanish [<https://elearning.wfh.org/es/centros-de-aprendizaje/hemofilia/>]
- What Are Bleeding Disorders [<https://www.nhlbi.nih.gov/health/bleeding-disorders>]
 (National Heart, Lung, and Blood Institute)
Also in Spanish [<https://www.nhlbi.nih.gov/es/salud/trastornos-hemorragicos>]

Diagnosis and Tests

- Coagulation Factor Tests [<https://medlineplus.gov/lab-tests/coagulation-factor-tests/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/pruebas-de-los-factores-de-la-coagulacion/>]
- Diagnosing Hemophilia [<https://www.cdc.gov/hemophilia/testing/>] (Centers for Disease Control and Prevention)
- Partial Thromboplastin Time (PTT) Test [<https://medlineplus.gov/lab-tests/partial-thromboplastin-time-ptt-test/>]
 (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-tpt-tiempo-parcial-de-tromboplastina/>]
- Prothrombin Time Test and INR (PT/INR) [<https://medlineplus.gov/lab-tests/prothrombin-time-test-and-inr-ptinr/>]
 (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-tiempo-de-protrombina-e-inr-ptinr/>]

Treatments and Therapies

- Treatments for Blood Disorders [<https://www.nhlbi.nih.gov/health/blood-bone-marrow-treatments>]
 (National Heart, Lung, and Blood Institute)
Also in Spanish [<https://www.nhlbi.nih.gov/es/salud/sangre-y-medula-osea-tratamientos>]

Living With

- Hemophilia: Handling Bleeds [<https://kidshealth.org/en/parents/hemophilia-bleeds.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/hemophilia-bleeds.html>]
- Living with Hemophilia [<https://www.cdc.gov/hemophilia/living-with/>] (Centers for Disease Control and Prevention)
- Travel Safe with a Bleeding Disorder [<https://www.cdc.gov/hemophilia/travel-safe/>] (Centers for Disease Control and Prevention)

Related Issues

- Testing for Inhibitors and Hemophilia [<https://www.cdc.gov/hemophilia/testing/testing-for-inhibitors-and-hemophilia.html>]
(Centers for Disease Control and Prevention)

Genetics

- Hemophilia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/hemophilia>]  (National Library of Medicine)

Statistics and Research

- Data and Statistics on Hemophilia [<https://www.cdc.gov/hemophilia/data-research/>] (Centers for Disease Control and Prevention)

Clinical Trials

- ClinicalTrials.gov: Blood Coagulation Disorders [<https://clinicaltrials.gov/search?cond=%22Blood+Coagulation+Disorders%22&aggFilters=status:not%20rec>]
 (National Institutes of Health)

Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: The attitudes and treatment practices of Hungarian primary care dentists regarding... [<https://www.ncbi.nlm.nih.gov/pubmed/40684000>]
- Article: Artificial Intelligence in the Management of Hereditary and Acquired Hemophilia: From... [<https://www.ncbi.nlm.nih.gov/pubmed/40649878>]
- Article: Prosthesis survival situation and complications following total hip arthroplasty in hemophilic... [<https://www.ncbi.nlm.nih.gov/pubmed/40634963>]
- Hemophilia -- see more articles [<https://pubmed.ncbi.nlm.nih.gov/?term=%22Hemophilia+A%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bdat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+>]

Find an Expert

- Find a Hematologist [<https://www.hematology.org/education/patients/find-a-hematologist>] (American Society of Hematology)
- National Heart, Lung, and Blood Institute [<https://www.nhlbi.nih.gov/>] 

Children

- Blood Test: Factor IX Activity (For Parents) [<https://kidshealth.org/en/parents/test-factor9.html>] (Nemours Foundation)
- What is Hemophilia (For Parents) [<https://kidshealth.org/en/parents/hemophilia.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/hemophilia.html>]

Women

- Information on Hemophilia for Women [<https://www.cdc.gov/hemophilia/about/information-for-women.html>] (Centers for Disease Control and Prevention)
- Women and Girls with Hemophilia [<https://www1.wfh.org/publications/files/pdf-2342.pdf>] (World Federation of Hemophilia) - PDF
Also in Spanish [<https://www1.wfh.org/publications/files/pdf-2350.pdf>]

Patient Handouts

- Hemophilia [<https://medlineplus.gov/ency/article/000537.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/000537.htm>]
- Hemophilia A [<https://medlineplus.gov/ency/article/000538.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/000538.htm>]
- Hemophilia B [<https://medlineplus.gov/ency/article/000539.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/000539.htm>]
- Partial thromboplastin time (PTT) [<https://medlineplus.gov/ency/article/003653.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/003653.htm>]



MEDICAL ENCYCLOPEDIA

Factor IX assay [<https://medlineplus.gov/ency/article/003679.htm>]

Factor VIII assay [<https://medlineplus.gov/ency/article/003678.htm>]

Hemophilia [<https://medlineplus.gov/ency/article/000537.htm>]

Hemophilia A [<https://medlineplus.gov/ency/article/000538.htm>]

Hemophilia B [<https://medlineplus.gov/ency/article/000539.htm>]

Partial thromboplastin time (PTT) [<https://medlineplus.gov/ency/article/003653.htm>]

Related Health Topics

Bleeding Disorders [<https://medlineplus.gov/bleedingdisorders.html>]

National Institutes of Health

The primary NIH organization for research on *Hemophilia* is the National Heart, Lung, and Blood Institute [<http://www.nhlbi.nih.gov/>]

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