What is hemophilia?

The word “hemophilia” comes from two Greek words: haima, meaning blood, and philia, meaning affection. It is pronounced “he-mo-feel-ia”.

Hemophilia is not a condition you can “catch” – it is hereditary, meaning it is passed on from one or both parents to the child, who then carries the gene for hemophilia, and has the condition from birth.

People with hemophilia, commonly referred to as hemophiliacs, have a problem with clotting in their blood. Clotting is the process by which your blood forms a solid plug to help stop bleeding. Because people with hemophilia are lacking one of several blood-clotting proteins, called factors, they bleed for a longer time than others. This doesn’t mean that they bleed more profusely or quickly than others, just that they don’t stop bleeding as quickly.

Hemophiliacs don’t have a problem with minor cuts, which is a common myth. The real danger is from internal bleeding, or hemorrhaging. Bleeding in joints like the knees elbows, and ankles, or into tissues and muscles, can be a real problem and can lead to swelling and pain in the affected area, and even permanent damage. When bleeding happens in a vital organ, especially the brain, it can put the person’s life in danger.

The most common type of hemophilia is called hemophilia A, or classical hemophilia. Since it is caused by the lack of factor 8 in the blood (written factor VIII), it is also referred to as factor VIII deficiency hemophilia. Hemophilia B, or Christmas Disease, was first diagnosed in a Canadian named Steven Christmas in 1952. Caused by a lack of factor 9 (written factor IX), it is also referred to as factor IX deficiency hemophilia. Hemophilia C, which is the rarest form, is caused by a lack of factor XI (11), and is also referred to as factor XI (11) deficiency hemophilia. Unlike A and B, which can only be passed on by the mother, hemophilia C is caused by a gene from both the mother and father.

How common is hemophilia?

Hemophilia is a very rare disorder. The most common type, hemophilia A, affects fewer than 1 in 10,000 people, or about 2,500 Canadians. Hemophilia B affects about 1 in 50,000 people, or about 600 Canadians. And the rarest type, hemophilia C, affects about 1 in every 100,000 people.

Who gets hemophilia?

Hemophilia A and B can affect people of any race and colour. The most severe cases of these conditions are seen almost exclusively in males. For women to be affected by a severe case, they must receive the gene from both mother and father, which is extremely rare. Women are usually carriers of one gene, and the other gene they have, which is most often normal, means that they will only have symptoms of mild hemophilia, if any. Since the disorder is lifelong, it is usually diagnosed in the first year of life, meaning most people begin treatment for hemophilia as infants.

Hemophilia C occurs in both males and females, since it is inherited from both mother and father. It is most common among Ashkenazi Jews of Eastern European origin, with about 8% of these people being carriers of the gene.

It is also possible for a spontaneous mutation of a gene to occur, meaning that the person born with hemophilia or as the carrier of hemophilia did not get the defective gene from either parent, and that there is no family history of the disease. This is thought to be what happens in about 30% of cases – which means that any family can be affected by this relatively rare condition.
What is hemophilia?

How to prevent hemophilia

Since hemophilia is a hereditary condition, it cannot be prevented by the person who has it. However, testing for blood clotting factors and even genes in women with a family history of the disease can help them know if they are a carrier of the disease and whether they suffer from a mild form of hemophilia. Knowing this will not only help with any future bleeding issues, it will also tell them that their sons will have a 50% chance of having hemophilia A or B, and their daughters a 50% chance of being a carrier.

What are the signs and symptoms?

The signs and symptoms of hemophilia will vary depending on a person’s lack of clotting factors due to the condition. When levels are only slightly to moderately low, bleeding may only occur after a trauma or surgery. When levels are very low, a person may experience spontaneous bleeding.

The first signs of hemophilia usually occur in infants when their teeth are coming in, with excessive bleeding, or when a sharp tooth cuts the tongue or gums. Also common are surface bruises from falls when an infant begins to move around. As the child becomes more active, this kind of bleeding into soft tissue may become more frequent, especially after the age of two, when bleeding into joints is also seen more frequently.

Symptoms of spontaneous bleeding include:

- Many large or deep bruises
- Joint pain and swelling (caused by bleeding)
- Unexplained bruises or bleeding
- Blood in urine or stool
- Bleeding for longer than normal from a cut or injury, or after a lost tooth or surgery
- Nosebleeds for no apparent reason

Emergency signs and symptoms may include:

- Sudden pain, swelling and warmth in large joints, such as the knees, elbows, hips and shoulders, as well as in arm and leg muscles
- Bleeding from an injury, especially for those with severe hemophilia
- Bleeding into the brain, which can be associated with:
  - A painful, lasting headache
  - Repeated vomiting
  - Extreme tiredness or a change in normal behaviour
  - Sudden weakness or clumsiness of an arm or leg
  - Stiffness or pain in the neck
  - Double vision
  - Developing crossed eyes
  - Balance problems when walking
  - Convulsions or seizures (fits)
How is the disease diagnosed?

For people with a family history of hemophilia, it is possible to have the fetus tested during pregnancy (generally after 16 weeks) to see if it will be affected by the disease. If the mother is a carrier, simply performing an ultrasound to determine the sex of the baby will help – a girl might be a carrier, while a boy might actually have hemophilia A or B.

Fetal gene testing can also be done in one of two ways. The earlier of the two is called chorionic villus sampling, or CVS for short, which can be done after 11 weeks. It is a test of the cells from a membrane that surrounds the fetus. The test can be done like a Pap smear, or by using a needle passed through the abdomen. The second test is called an amniocentesis, and is performed after 15.5 weeks. This test involves using an ultrasound to find a pocket of amniotic fluid, which contains cells shed by the fetus. A needle is inserted through the abdomen and into the uterus, and a small amount of amniotic fluid is removed. The DNA testing for both procedures takes up to 3 or 4 weeks, and both involve a slight risk (0.5 to 1%) of miscarriage.

Another method involves testing the fetal blood, which can be done at 18 weeks. Blood is taken from the umbilical vein, using ultrasound and a needle inserted through the abdomen. Factor levels of the blood can be tested immediately, once it is determined that the affected blood is not from the mother. However, the procedure is difficult, and there is a higher risk (up to 5%) of miscarriage.

Testing in children involves a few simple lab tests. A blood sample is taken, and then the levels of factor VIII (hemophilia A), factor IX (hemophilia B) or factor XI (if hemophilia C is suspected) are measured. These tests will tell doctors what kind of bleeding disorder the patient has, if any, as well as how severe it is. One of the tests will test the blood to measure levels of von Willebrand factor (VWF) which is a protein that acts like glue in the clotting process. If the VWF level is low, then the child has von Willebrand disease and not hemophilia. These tests usually take one or two weeks to complete.

Hemophilia A and B are easier to classify than hemophilia C.

<table>
<thead>
<tr>
<th>Hemophilia A and B Classification</th>
<th>Level of Factor VIII or IX in the Blood</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild</td>
<td>5 to 30% of normal</td>
</tr>
<tr>
<td>Moderate</td>
<td>1 to 5% of normal</td>
</tr>
<tr>
<td>Severe</td>
<td>Less than 1% of normal</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Hemophilia C Symptoms (varies from person to person)</th>
<th>Level of Factor XI in the Blood</th>
</tr>
</thead>
<tbody>
<tr>
<td>Problems after surgery or trauma</td>
<td>15 to 70% of normal</td>
</tr>
<tr>
<td>Mild to moderate bleeding</td>
<td>Less than 15% of normal</td>
</tr>
</tbody>
</table>
How is hemophilia treated?

There is currently no cure for hemophilia, though treatments are helping most people with the condition lead fairly normal lives. The basic treatments involve replacing the missing clotting factors by infusing them into the patient, using an injected hormone to increase the efficiency of factor VIII in the body (only for hemophilia A), or prescribing drugs to help hold clots in place once they form.

The standard treatment for hemophilia is called factor replacement therapy. Treatment involves infusions (injections into the bloodstream) of the missing clotting factor to help stop or prevent bleeding. These injections are effective for only a short period of time, with the blood returning to its original clotting capability within two or three days. For people with mild hemophilia, factor replacement therapy may only be needed after an injury or before surgery. For people with moderate to severe hemophilia, a person may require more regular infusions, such as several times a week or even daily, to prevent serious bleeding. The same is true for children who are very active. This type of treatment is called prophylaxis therapy. Patients and caregivers can be trained to perform these infusions at home.

People with mild to moderate hemophilia A can also be treated with a drug called desmopressin, which is a copy of a natural hormone that releases von Willebrand factor (VWF) into the blood. It is thought that this helps because VWF helps transport factor VIII to the site of damaged blood vessels. This therapy can be infused, injected under the skin, and even taken as a nasal spray. Since it stimulates the use of VWF in your system, and your body needs to rebuild its stores of this clotting factor, it cannot be used more than once a day.

There are also drugs called antifibrinolytics that help hold a clot in place. These are useful before dental work, in cases of nose and minor intestinal bleeds, or for carriers with heavy, prolonged menstrual bleeding. They don’t actually help form a clot, so they are not used instead of desmopressin or factor replacement therapy. They come in tablet form.

Patients should know that any medication comes with a risk of side effects, and should consider speaking with their healthcare team if they have any questions or concerns about a medication or its potential unwanted effects.

People with hemophilia C can also receive fresh, frozen plasma infusions for mild bleeding episodes.

For people with joint damage due to bleeding, physical therapy may help the joint work better by preserving movement and preventing frozen or deformed joints. In cases where damage is severe, an artificial joint may be necessary.

Living with hemophilia

Living with any chronic condition can be a challenge, whether it is mild, moderate or severe. Here are some tips that may help you avoid excessive bleeding and joint damage as you work to better manage hemophilia.

1. **Exercise as regularly as possible.** Taking part in activities such as swimming, bike riding and walking can help you strengthen muscles while protecting joints. Of course, avoiding contact sports such as football, hockey or wrestling is also important for your health and safety.

2. **Avoid certain medications.** Certain drugs can increase your risk of bleeding, such as aspirin and nonsteroidal anti-inflammatory drugs (NSAIDs) like ibuprofen. A pain reliever like acetaminophen is a safe alternative. Blood-thinning medications, such as heparin and warfarin, should also be avoided, since they prevent blood from clotting. Even certain herbal supplements contain ingredients that may cause bleeding if you have hemophilia, so
What is hemophilia?

be sure to talk to your doctor or other healthcare professional before taking any new ones.

3. **Take care of your teeth.** Good oral hygiene can help prevent the need dental surgery, which can lead to unnecessary bleeding.

4. **Avoid injuries that could cause bleeding.** If you or your child has hemophilia, ask your doctor to help find ways to stay active while avoiding injury. Protective devices such as kneepads, elbow pads, helmets and safety belts may help prevent injuries from falls and other accidents. Also, be sure to keep your home free of furniture with sharp corners and to keep any sharp objects out of reach or locked away.

Learning as much as possible about your disease and actively working with your healthcare professionals are effective ways to regain control over your life. There is a great deal of information out there that can help. Check out some of the websites listed below to get started.

**Empowering both the patient and the caregiver**

Patients as well as their physicians can access the following sites to find out more information about the major forms of hemophilia and other bleeding disorders, their symptoms, treatment options as well as recent related scientific discoveries.

**Resources**

Helpful information and support networks on the internet:

Canadian Hemophilia Society ([www.hemophilia.ca](http://www.hemophilia.ca))

World Federation of Hemophilia ([www.wfh.org](http://www.wfh.org))

National Hemophilia Organization ([www.hemophilia.org](http://www.hemophilia.org))

Canadian Blood Services ([www.bloodservices.ca](http://www.bloodservices.ca))

Association of Hemophilia Clinic Directors of Canada (AHCDC) ([www.ahcdc.ca/publications.html](http://www.ahcdc.ca/publications.html))

[www.livingwithhemophilia.ca](http://www.livingwithhemophilia.ca)