HAEMOPHILIA

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Definition

Haemophilia is a disorder of your blood-clotting system. Clotting is the process by which your blood changes from a liquid to a solid state.

There are several types of haemophilia. All types can cause prolonged bleeding. If you have haemophilia and you have a cut, you'll bleed for a longer time than you would if your blood clotted normally. Small cuts usually aren't such of a problem. The greater health concern, particularly with haemophilia A and B, is deep internal bleeding and bleeding into joints.

Haemophilia is a lifelong disease, but with proper treatment and self-care, most people with haemophilia can maintain an active, productive lifestyle.

Symptoms

Signs and symptoms of haemophilia vary depending on how deficient you are in clot-forming proteins called clotting factors. If levels of your deficient clotting factor are very low, you may experience spontaneous bleeding. If levels of your deficient clotting factor are slightly to moderately low, you may bleed only after surgery or trauma.

Signs and symptoms of spontaneous bleeding may include:

- Many large or deep bruises
- Joint pain and swelling caused by internal bleeding
- Prolonged bleeding from cuts or injuries or after surgery or tooth extraction
- Nosebleeds without a known cause
- Unexplained and excessive bleeding or bruising
- Blood in your urine or stool
- Tightness in your joints
- In infants, unexplained irritability
- Unusual bleeding after immunizations

Emergency signs and symptoms of haemophilia may include:

- Sudden pain, swelling, and warmth of large joints, such as knees, elbows, hips and shoulders, and of the muscles of your arms and legs
- Bleeding from an injury, especially if you have a severe form of haemophilia
- Repeated vomiting
- Extreme fatigue
- Neck pain
- Double vision
- Painful, lasting headache

Babies with haemophilia

At first, because of limited mobility, a baby with haemophilia usually won't have many problems related to haemophilia. But as your baby begins to move around, falling and bumping into things, superficial bruises may occur. This bleeding into soft tissue may become more frequent the more active your child becomes.

When to see a doctor

If you're pregnant or considering a pregnancy and have a family history of haemophilia, talk to your doctor. You may be referred to a medical genetics specialist or a specialist in bleeding disorders, who can help you determine if you are a carrier of haemophilia. If you are a carrier, it's possible to test the fetus during pregnancy to determine if it is affected by the disease.

If you have a baby boy, prolonged bleeding following circumcision may be the first indication of haemophilia. In girls and in boys who aren't circumcised, easy bruising when the child becomes more mobile may lead to the diagnosis. The first episode of bleeding generally occurs by the time a child is 2 years old. If your baby bruises easily as he or she becomes more mobile, see your doctor.

Causes

When you're cut or bleeding internally, your body normally pools blood cells together to form a clot to stop the bleeding. This process is called coagulation. Coagulation involves blood particles called platelets and plasma proteins that encourage clotting (clotting factors). The cause of haemophilia is a deficiency of one of these clotting factors.

Which type of haemophilia you have depends on which clotting factor is deficient:

- Haemophilia A: The most common type, haemophilia A is caused by lack of enough clotting factor 8 (VIII).
- Haemophilia B: This second most common type is caused by lack of enough clotting factor 9 (IX).
- Haemophilia C: This type is caused by a lack of clotting factor 11 (XI), and symptoms are often mild with this type of haemophilia.

Haemophilia inheritance

Everyone has two sex chromosomes, one from each parent. Females inherit an X chromosome from their mother and an X chromosome from their father. Males inherit an X chromosome from their mother and a Y chromosome from their father.

- The gene that causes haemophilia A or B is located on the X chromosome, so it can't be passed from father to son. Haemophilia A or B almost always occurs in boys and is passed from mother to son through one of the mother's genes. Most women who have the defective gene are simply carriers and exhibit no signs or symptoms of haemophilia. It's also possible for haemophilia A or B to occur through spontaneous gene mutation.
- The gene that causes haemophilia C can be passed on to children by either parent. Haemophilia C can occur in both boys and girls.

Complications

Complications may occur from the condition or from the treatment for the condition:

- Deep internal bleeding: Heamophilia may cause deep muscle bleeding that leads to swelling of a limb. The swelling may press on nerves and lead to numbness or pain. This may result in a reluctance to use that limb.
- Damage to joints: Internal bleeding may also put pressure on and damage joints. Pain sometimes may be severe, and you may be reluctant to use a limb or move a joint. If bleeding occurs frequently and you don't receive adequate treatment, the irritation may lead to destruction of the joint or the development of arthritis.
- Infection: People with haemophilia are more likely to receive blood transfusions and are at greater risk of receiving contaminated blood products. Until the mid-1980s, it was more likely for people with haemophilia to become infected with the human immunodeficiency virus (HIV) or with hepatitis through contaminated blood products. Since then, blood products are much safer because of steps taken to screen the supply of donated blood. The risk of infection through blood products has decreased substantially since the introduction of genetically engineered clotting products called recombinant factors, which are free of infection. However, it's still possible for people who rely on blood products to contract diseases. If you have haemophilia, consider receiving immunization against hepatitis A and B.
- Adverse reaction to clotting factor treatment: In some people with haemophilia, the immune system sees these clotting factor treatments as foreign. When this happens, the immune system develops proteins that inactivate the clotting factors used to treat bleeding. Researchers are investigating treatments to dampen the immune system's response and allow continuing treatment with clotting factors.

Preparing for your appointment

Haemophilia is diagnosed at an average age of 9 months and almost always by age 2. If your child has heavy bleeding that can't be stopped after an injury, call 10111 or 112 from any cellphone for a local emergency number or go to an emergency room.

If your child's symptoms are less severe — such as bruising that seems excessive after minor injuries — call your family doctor or your child's pediatrician. In some cases when you call to set up your appointment, you may be referred to a doctor who specializes in bleeding disorders (hematologist).

Tests and diagnosis

For people with a family history of haemophilia, it's possible to test the fetus during pregnancy to determine if it is affected by the disease. However, such testing poses some risk to the fetus. Discuss the benefits and risks of testing with your doctor.

Analysis of a blood sample from either a child or an adult can show a deficiency of a clotting factor. Sometimes mild haemophilia isn't diagnosed until after a person has undergone surgery and excessive bleeding results.

Treatments and drugs

While there's no cure for haemophilia, most people with the disease can lead fairly normal lives. Haemophilia treatment varies depending on the severity of the condition:

- Mild haemophilia A: Treatment may involve slow injection of the hormone desmopressin (DDAVP) into a vein to stimulate a release of more clotting factor to stop bleeding. Occasionally, desmopressin is given as a nasal medication.
- Moderate to severe haemophilia A or haemophilia B: Bleeding may stop only after an infusion of clotting factor derived from donated human blood or from genetically engineered products called recombinant clotting factors. Repeated infusions may be needed if internal bleeding is serious.
- Haemophilia C: The clotting factor missing in this type of haemophilia (factor XI) is available only in Europe.

Ongoing treatment

Your doctor may recommend:

- Regular infusions of DDAVP or clotting factor: The infusions can help prevent bleeding. This approach may reduce time spent in the hospital and limit side effects such as damage to joints. Your doctor can show you how to perform the infusions.
- Clot-preserving medications (antifibrinolytics): These medications help prevent clots from breaking down.
- Fibrin sealants: These medications can be applied directly to wound sites to promote clotting and healing. Fibrin sealants are especially useful in dental therapy.
- Physical therapy: It can ease sign and symptoms if internal bleeding has damaged your joints. If internal bleeding has caused severe damage, you may need surgery.
- First aid for minor cuts: Using pressure and a bandage will generally take care of the bleeding. For small areas of bleeding beneath the skin, use an ice pack. Ice pops can be used to slow down minor bleeding in the mouth.

- Vaccinations: Although blood products are screened, it's still possible for people who rely on them to contract diseases. If you have haemophilia, consider receiving immunization against hepatitis A and B.

For minor cuts

If you or your child experiences a small cut or scrape, using pressure and a bandage will generally take care of the bleeding. For small areas of bleeding beneath the skin, use an ice pack. Ice pops can be used to slow down minor bleeding in the mouth.

Lifestyle and home remedies

These steps may help you avoid excessive bleeding and protect your joints:

- Exercise regularly: Activities such as swimming, bicycle riding and walking can build up muscles while protecting joints. Contact sports - such as football, hockey or wrestling, are not safe for people with haemophilia.
- Avoid certain medications: Drugs that can aggravate bleeding include aspirin and ibuprofen (Advil, Motrin, others). Instead, use acetaminophen (Tylenol, others), which is a safe alternative for mild pain relief. Also avoid certain blood-thinning medications, such as heparin and warfarin (Coumadin), which prevent blood from clotting. Certain herbal supplements also contain ingredients that may cause bleeding, especially if you have haemophilia. Talk to your doctor before taking any herbal supplements
- Practice good dental hygiene: This can help prevent the need to have a tooth pulled, which can lead to excessive bleeding
- Protect your child from injuries that could cause bleeding: If your child has haemophilia, ask your doctor for guidance that can help your child stay active while avoiding injury. Knee pads, elbow pads, helmets and safety belts all may help prevent injuries from falls and other accidents. In addition, keep your home free of furniture with sharp corners and keep sharp objects out of reach or locked away.

Coping and support

Get a medical alert bracelet for you or your child and be sure it's worn always. This bracelet lets medical personnel know that you or your child has haemophilia and the type of clotting factor that's best in case of an emergency.

These tips can help you and your child cope with haemophilia:

- Tap into resources at a designated haemophilia treatment center: Comprehensive haemophilia treatment centers are located throughout South Africa. A wide range of experts at these centers can work with you and your family doctor to create personalized plan for managing your haemophilia. These centers also provide support services, including education for you and your family. Ask your doctor or check the South African Haemophilia Foundation's website for a list of centers http://www.haemophilia.org.za/centres.html.
- Talk with a counselor: If your baby or child has haemophilia, you may be concerned about striking the right balance between keeping your child safe and encouraging as much normal activity as possible. A social worker or therapist with knowledge about haemophilia can help you cope with your concerns and identify the minimum limitations necessary for your child. Ask your doctor to recommend a mental health professional or a support group that can help.
- Let people know: Be sure to tell anyone who will be taking care of your child — baby sitters, workers at your child care center, relatives, friends and teachers — about your child's condition. Because it's also OK to let your child engage in non-contact organized sports, be sure to let coaches know too.
- Comfort your child: Stay calm and reassure your child during injections or infusions. Encourage and praise your child when the treatment is complete.

Source: The Mayo Clinic

Contact us

Please feel free to contact your Aon Healthcare Consultant if you have any concerns. You may also contact the **Aon Resolution Centre on 0860 835 272 or e-mail: arc@aon.co.za** for further information.

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