Living life with haemophilia

Information about haemophilia. How it can affect you. What treatments are available, as well as suggestions and advice to enable you to live your life in the best way possible.

Haemophilia

Haemophilia is a form of bleeding disorder. Bleeding disorders is a collective term for several different diseases where the blood's ability to coagulate does not work in the way that it should. Haemophilia means that a person is completely deficient in or has too little of a clotting factor in their blood. Clotting factors are proteins that cause the blood to clot.

In haemophilia A, which is the most common variant, a person is deficient in clotting factor VIII. Whereas with haemophilia B, a person is deficient in factor IX. Haemophilia A is five times more common than haemophilia B.

Haemophilia is classified as severe, moderate or mild depending on the amount of clotting factors that are present in the blood.

There are approximately 900 men and a small number of women with haemophilia in Sweden. The disease almost exclusively affects men, while women are carriers of the disease. If haemophilia is not treated either preventively (prophylactically) or when needed (on-demand), the disease can lead to bleeding in the muscles, joints, abdomen and brain.

From mild to severe haemophilia

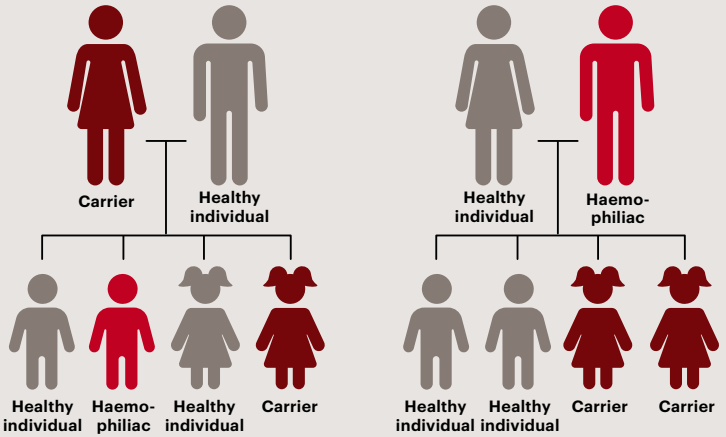
Haemophilia A means that a person has a deficiency or absence ofclotting factor VIII, and Haemophilia B means that a person has a deficiency or absence of clotting factor IX. The severity of the disease is dependent on the concentration of clotting factor in the blood.

How haemophilia is inherited

Of the new cases of haemophilia, about half are due to the disease being inherited, and half are due to new mutations where the disease was not previously present in the family. Haemophilia is caused by a mutation on the X chromosome, which means that clotting factors are not produced at all or in very small quantities. The X-chromosome and the Y-chromosome determine a person's sex and are therefore also called the sex chromosomes. Females have two X chromosomes. Males have an X chromosome and a Y chromosome.

Because males only have a single X chromosome, the mutation on the X chromosome means that they will always develop haemophilia. In women, on the other hand, who have two X chromosomes, the mutation is usually compensated for by the healthy gene on the other X chromosome. Therefore, it is almost exclusively boys and men who have haemophilia, whereas it is very rare for women to have haemophilia.

A woman with a mutation on her X chromosome will instead be a carrier and pass the mutated gene on to her children. Even women who are carriers can experience bleeding, because they will have reduced levels of factor VIII or factor IX.



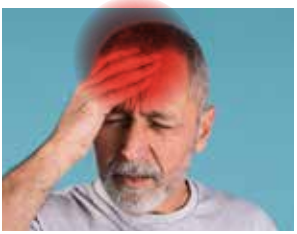
A woman who is a carrier and a healthy man: Daughters of a mother who is a carrier and a healthy father will receive a healthy X chromosome from their father, and from their mother, 50 percent will receive a mutated X chromosome and 50 percent a healthy X chromosome. The sons will receive a Y chromosome from their father and either a healthy X chromosome or the mutated X chromosome from their mother. Here too, the numbers are 50 percent for each chromosome.

A man with haemophilia and a healthy woman: Daughters of a father with haemophilia and a healthy mother will be carriers because they will all inherit the defective X chromosome from their fathers. The sons will be healthy because they receive a Y chromosome from their father and can only inherit healthy X chromosomes from their mother.

Common symptoms of bleeding

The most common symptom of haemophilia is bleeding anywhere in the body, such as in the joints, mucous membranes and muscles. Bleeding can start after a blow, a knock or through overexertion.

- **Muscle bleeding**
  The muscles in the calves, thighs and arms are often affected and the bleeding can continue for some time without any symptoms. After a while, pain and a tingling sensation often occur. Superficial bleeding causes bruising while some other types of bleeding can cause the arms and legs to swell and become tender.
- **Joint bleeding**
  Knees, elbows, ankles, shoulders, hips and wrists are prone to bleeding. The first sign of bleeding is that the joint will feel tender and warm. Then it will be difficult to move the joint.
- **Internal bleeding**
  Bleeding in internal organs is unusual, but bleeding in the brain and stomach can occur, for example in the event of major trauma. The symptoms include the person being pale, feeling unwell, having a severe headache and being difficult to wake up. This is an emergency situation that requires immediate hospital care.



Surgery and childbirth

People with severe haemophilia are usually undergoing treatment and are therefore prepared in the event of surgery, tooth extraction and childbirth. However, these types of situations can be dangerous for carriers or for people who have not yet been diagnosed.

Getting a diagnosis

If a mother knows that she is a carrier and she gives birth to a male child, we know that the child will have haemophilia. A simple blood test performed on the child will show the factor levels. If it is a new mutation, the disease will usually manifest itself by the child bruising easily or through spontaneous bleeding in the joints and muscles.

An investigation into suspected haemophilia is usually carried out at one of the country's three Coagulation Clinic. Diagnosis is based on the child's medical history and the results of a number of blood tests, which measure the blood's ability to clot and the levels of clotting factors in the blood.

In haemophilia, a person has either low levels of clotting factor VIII (haemophilia A) or clotting factor IX (haemophilia B). If there is less than one percent of these clotting factors present in the blood, a person is considered to have a severe bleeding disorder.

Remember!

People with haemophilia should avoid medication containing acetylsalicylic acid, for example Treo and Albyl. The same applies to NSAIDs, for example Ibuprofen and Ipren.

Reporting bleeding incidents

Tell your doctor or clinic about every incident of bleeding!

If you have haemophilia or help a relative who has the disease, it is important to write down when minor or major bleeding incidents occur. Give the bleeding incident report to your doctor or clinic. It is important for you and everyone else with haemophilia to receive the best possible treatment.

Treatment is preventative

People with severe haemophilia are treated preventively (prophylactically) often with factor concentrates that are injected into a blood vessel in the hand or in the crook of the arm. A recombinant protein is used to supply the clotting factor that is deficient. Today, there are also preparations that can be taken subcutaneously (under the skin) and are based on monoclonal antibodies instead of a human protein. They are only available for people with haemophilia A.

Those with mild haemophilia usually only need to take factor concentrates when needed (on-demand), for example before surgery or a tooth extraction.

There are also ongoing trials using gene therapy as a potential cure for haemophilia. The treatment entails that a healthy variant of the gene is introduced into the body in a modified adenovirus to replace the defective gene. Once inside the body it starts to produce the deficient clotting factor.

Pregnancy and childbirth

Women who are carriers of haemophilia should contact a Coagulation Clinic when they become pregnant. If you are pregnant and have a partner with haemophilia, your partner can contact his doctor if you think the child may be a carrier.

You should also inform your midwife that you are a carrier and are more susceptible to bleeding than normal. Then you can make a plan together for your pregnancy and childbirth.

In general, no special medical examinations are necessary during pregnancy, except for a blood test during the third trimester to check the levels of factor concentrate. If the level is low, you may need to take factor concentrate before delivery.

A baby with haemophilia does not need to be cared for any differently to other newborn babies. However, a blood sample will be taken to measure the factor levels if it is known that the child has haemophilia.

Haemophilia and quality of life

Today, most people with haemophilia who have access to treatment can live a full life. However, you have to make sure to follow your doctor's advice and have your medication with you at all times. However, despite access to good treatment, there are shortcomings in the quality of life of people with haemophilia, including considerable concern on the part of relatives, ignorance about the disease amongst healthcare professionals and under-reporting of bleeding incidents. More than six out of ten people with haemophilia state that they are often worried because of their disease.

Become a member. It makes a difference!

It is easy to register at fbis.se

Together we are stronger!

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