

HAEMOPHILIA

This information sheet is for your general information and is not a substitute for medical advice. You should contact your doctor or other healthcare provider with any questions about your health, treatment or care.

What is haemophilia?

Haemophilia is a disorder of the blood-clotting system. Normally when you cut yourself, your blood plugs the wound by changing from a liquid to a solid and forming a blood clot. If you have haemophilia, your blood will not clot normally, and you may bleed for longer than normal or you may bleed internally, particularly into joints such as your knees, elbows and ankles.

Blood contains special proteins called 'clotting factors', which are involved in the clotting process. If you have haemophilia, your body does not produce enough of these clotting factors or does not produce it at all.

There are different types of haemophilia. If you have haemophilia A, you have a deficiency of clotting factor VIII, which is the cause of about 80% of haemophilia cases. If you have haemophilia B, you have a deficiency of clotting factor IX. Haemophilia can be mild, moderate or severe, depending on how much factor VIII or IX is missing from your blood.

Haemophilia is rare and almost always affects men.

Symptoms

Symptoms of haemophilia include:

- bleeding from cuts or injuries that continues for much longer than normal
- unexplained bleeding
- bruising or nosebleeds
- unusually large or deep bruising from an injury
- joint pain, swelling or a feeling of warmth in the joint
- blood in the urine or faeces.

Internal bleeding into the joints, muscles or other tissue can occur spontaneously (without an external cause), or from an injury. Repeated bleeding in a joint can lead to arthritis or permanent joint damage, causing stiffness and problems with mobility (ability to move around).

Bleeding can occur in or around the brain or spinal cord. This is rare and usually occurs only in those with severe haemophilia, but occasionally it can occur in those with the mild or moderate forms of the condition. How easily or badly a person with haemophilia bleeds depends on the severity of their condition.

Causes

Haemophilia is the result of a defective gene, which leads to inefficient production of the factors VIII or IX, in the blood. Haemophilia can be inherited because the defective gene can be passed on from parent to child. About 70% of haemophilia cases are inherited.

Women who have the defective gene on only one of their X chromosomes, don't usually develop symptoms of haemophilia and are known as carriers of the condition. Women who are carriers have a 50% chance that their sons will have haemophilia too.

In about 30% of cases there is no family history of haemophilia.

Diagnosis

Blood tests can identify whether the blood has low levels of any of the clotting factors or whether one of the factors is missing entirely. They can also measure the 'clotting time' of the blood. Normal clotting time is between four and seven minutes, but in a person with haemophilia, it will be longer. The results of these tests will show if the person has haemophilia, what type of haemophilia they have and whether it is mild, moderate or severe.

A woman who wants to become pregnant and who has a family history of haemophilia can be tested to determine whether she is a carrier. Women who are carriers and who are already pregnant can be given a chorionic villus sampling (CVS) test that will show whether their (male) child has haemophilia.

There is no known cure for haemophilia.

Treatment

Treatment for the condition is based on 'replacement therapy' – replacing the clotting factor that is too low or missing.

If necessary, mild haemophilia A can be treated with an injection of the hormone desmopressin, which stimulates the body's production of clotting factor VIII. Replacement therapy is not usually needed for mild haemophilia.

Moderate to severe haemophilia can be treated with an injection of the replacement clotting factor. Unfortunately, the replacement clotting factor remains active for only a short period and usually loses half its activity after only 12 hours. If the bleeding is serious, repeated injections are necessary. Injections can be given in hospital, but many people with haemophilia are taught to inject themselves at home.

Replacement clotting factors can be given as needed, or regular injections can be given two or three times a week to keep the levels of clotting factors in the blood high enough so that bleeding does not occur.

Synthetic alternatives called 'recombinant factors' are now used. Recombinant factors are regarded as free from the risk of blood infections.

Complications

The most common complication of haemophilia is arthritis due to repeated bleeding within the joint. This can cause long-term pain and loss of mobility.

Some people with haemophilia can develop antibodies to the replacement clotting factors used to treat the condition. The antibodies can destroy the clotting factors before it has a chance to work. This can be a serious problem as it means that the main treatment for haemophilia is no longer effective. When antibodies develop, doctors may use larger doses of the clotting factors or try different types of factors. Antibodies to clotting factors develop in about 20% of those with severe haemophilia A, and about 1% of those with haemophilia B.

Before the dangers of the AIDS epidemic were fully recognised, HIV-infected blood was used to produce replacement clotting factors, and many haemophiliacs acquired the disease between 1979 and 1985. Hepatitis has also been a problem in the past, with many haemophiliacs contracting the disease from blood carrying the virus.

Blood donors are now carefully screened and all donated blood products are tested for these viruses. Vaccination against hepatitis A and B is also offered to all haemophiliacs.

Your doctor will use the following information to monitor your progress:

- Clotting factor levels (%)
- Factor replacement required
- Factor batch number
- Site of bleeding

Reference

1. MOMENTUM HEALTH SOLUTIONS. 2021. *Haemophilia clinical policy*.

The content of this publication is the intellectual property of Momentum Health (Pty) Limited and any reproduction of this publication is prohibited unless written consent is obtained.