
Haemophilia

Summary

- Haemophilia is an inherited condition where the blood doesn't clot properly
 - It is caused when blood does not have enough clotting factor.
 - The lack of clotting factor means that people with haemophilia tend to bleed internally into their joints and muscles.
 - Haemophilia is treated by injecting clotting factor concentrate into a vein – which either prevents or reduces bleeding.
 - Once you have been trained by health professionals, you can do this at home.
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Haemophilia is an inherited bleeding disorder where blood doesn't clot properly. It is caused when blood does not have enough clotting factor. A clotting factor is a protein in blood that controls bleeding.

In Australia there are more than 2,800 people diagnosed with haemophilia (mostly males). Severe haemophilia in females is very rare.

Haemophilia is a lifelong condition. It can't be cured, but with current clotting factor treatments it can be managed effectively.

Internal bleeding episodes or 'bleeds' are the main problem for most people with haemophilia. Bleeds are most commonly into joints or muscles. They can happen without an obvious cause, or as a result of injury. Over time, repeated bleeding into joints and muscles can cause permanent damage, such as arthritis in the joints, and chronic pain.

What causes haemophilia?

Haemophilia is an inherited condition and occurs in families. It is caused by a genetic variation (change) in one of two genes (the F8 and F9 genes) that make blood clotting factor. Changes in these genes can alter or reduce the blood clotting process.

A change in the F8 gene, which makes blood clotting factor VIII (8), causes haemophilia A. A change in the F9 gene, which makes blood clotting factor IX (9), causes haemophilia B.

These genes are located on the X chromosome, which is one of the two sex chromosomes (X and Y). Haemophilia is inherited in an '**X-linked recessive pattern**'.

Men who carry the genetic change (and therefore have haemophilia) will pass it on to their daughters but not their sons. These daughters are said to 'carry' the genetic change. They may or may not have symptoms of haemophilia.

Women who carry the genetic change may pass it on to their sons and daughters:

- There is a 50 per cent chance at each birth of a son that the son will have the genetic change and therefore have haemophilia.
- There is a 50 per cent chance at each birth of a daughter that the daughter will carry the gene. Daughters with the genetic change may or may not have symptoms of haemophilia.

Males (XY) only have one X chromosome, so only one copy of the genetic change is needed to cause the condition. Females (XX) have two X chromosomes and therefore have two copies of these genes. Women with a changed F8 or F9 gene usually have another copy that is not changed. Having one normal copy of the gene is often enough to control bleeding.

Sometimes there is no family history of haemophilia

In about one third of people born with haemophilia, there is no history of the disorder in the family. This happens when a genetic change in the F8 or F9 gene occurs randomly during reproduction and is passed on at conception.

Once haemophilia appears in a family the genetic change is then passed on from parents to children following the usual pattern for haemophilia.

If your child has been diagnosed with haemophilia, it may be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand haemophilia and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

Type and severity of haemophilia

There are two major types of haemophilia:

- Haemophilia A is the most common form and is caused by having reduced levels of clotting factor VIII (8). This is caused by a change in the F8 gene.
- Haemophilia B, also known as Christmas Disease, is caused by having reduced levels of clotting factor IX (9). This is caused by a change in the F9 gene.

Both types have the same symptoms.

There are three levels of severity in haemophilia: mild, moderate and severe. The level of severity depends on the amount of clotting factor in the person's blood. The normal range of factor VIII and factor IX in a person's blood is between 50 and 150 per cent.

People with **mild haemophilia** (5 – 40 per cent of normal clotting factor):

- usually only have bleeding problems after having teeth taken out, surgery or a bad injury or accident
- might never have a bleeding problem.

People with **moderate haemophilia** (1 – 5 per cent of normal clotting factor):

- might have bleeding problems after having teeth taken out, surgery or a bad injury or accident
- may have bleeding problems after minor injuries, such as sporting injuries
- rarely have a bleed for no obvious reason.

People with **severe haemophilia** (less than 1 per cent of normal clotting factor):

- often have bleeds into joints, muscles and soft tissues
- can have bleeds for no obvious reason
- can have bleeds after surgery, dental work or injuries, including minor bumps or knocks.

Acquired haemophilia

Acquired haemophilia is another bleeding disorder. It is not inherited like the classical form of haemophilia. It is a very rare condition where a person's immune system develops antibodies against one of their body's own clotting factors. This results in a reduced factor level in their blood. Acquired haemophilia usually develops when people are older, and can affect both men and women.

How is haemophilia diagnosed?

Haemophilia is usually diagnosed through:

- physical signs that a person has unusual bleeding problems, and
- checking the family history for bleeding problems, and
- blood tests for a person's clotting factor levels and
- genetic testing that identifies a change in the F8 or F9 gene that causes haemophilia – Haemophilia Foundation Australia has more information about [what genetic testing may involve](#).

Depending on whether there is a known family history of haemophilia, and the severity of the person's symptoms, haemophilia may be diagnosed before birth, in early infancy, or not until later in life.

Mild or moderate haemophilia might not be diagnosed until children are older, or sometimes until they are adults. In a person with mild haemophilia, minor injuries may heal normally because there is enough clotting factor activity in their blood. Therefore, a bleeding problem might not be noticed until the person has surgery, a tooth taken out or a major accident or injury.

Testing for haemophilia before birth

If you or your partner are pregnant and you have a known genetic change in the family that you want to test for before your baby is born, there are two main genetic testing options:

- prenatal diagnostic testing – the genetic change identified in the family can be tested for using **chorionic villus sampling (CVS)** or **amniocentesis**
- IVF with pre-implantation genetic diagnosis (PGD) – if you are undergoing **IVF**, IVF technology can be used to test embryos for the gene change identified in the family before embryo implantation and pregnancy.

A genetic counsellor can explore these genetic testing options with you in further detail, along with any implications. There may be substantial costs involved for these tests and procedures and it may be valuable to clarify this and options for support beforehand.

If you are pregnant and there is a history of haemophilia in your family but you do not know whether you carry the gene, it is important to let the obstetrics team know so that they can plan the safe delivery of your baby with the haemophilia team.

Testing for haemophilia in babies

If you have a known family history of haemophilia, a sample of your baby's blood can be tested after birth to check the blood clotting factor levels and see whether they have haemophilia. Testing is repeated when the baby is six months of age to confirm the results.

Where there is no known family history of haemophilia, children with severe haemophilia are usually diagnosed in their first year of life when their parents or health professionals notice unusual bruising or bleeding problems.

Most babies with haemophilia do not have bleeding problems at birth. However, some bleeding problems may appear at birth or soon after. Haemophilia may be suspected if babies:

- have internal bleeding or unusual swelling or bruising after delivery
- continue to bleed after a heel prick (or after circumcision, if this is performed)
- have excessive bruising after immunisation.

Other signs may include:

- bruising easily
- painful swelling
- reluctance to use an arm or a leg.

Growing up with haemophilia

With treatment and support from their haemophilia treatment centre, most people with haemophilia can live relatively normal healthy lives. Unless there are complications, young people can expect to grow up with few or no joint problems from their haemophilia. With sensible precautions, they can play most sports, exercise and look forward to a full and productive life.

As children grow they learn to recognize that bleeding may be occurring. Even before pain or swelling becomes obvious they may recognise the 'funny feeling' that is one of the earliest signs of a joint bleed.

The specialist nurses and physiotherapists at your nearest haemophilia treatment centre can advise parents and children on haemophilia issues during normal childhood stages.

Immunisations and haemophilia

Children with haemophilia can have all the normal immunisations at the usual age. However, informing the nurse or doctor giving the immunisation that the baby or toddler has haemophilia is important.

Injections can be given into the fatty tissue under the skin (subcutaneously), rather than into the muscle, and

pressure put on the skin where the child was injected. This reduces the risk of bruising and bleeding. However, changing the way of giving immunisations isn't necessary for all children with haemophilia.

If you have a child with haemophilia, contact your haemophilia treatment centre for advice on how your child should be immunised.

Women who carry the haemophilia gene

Many girls or women who carry the genetic change do not have signs or symptoms of a bleeding disorder. However, some do. This is sometimes called 'having a bleeding tendency'. Symptoms of having a bleeding tendency may include:

- bruising easily
- heavy menstrual bleeding, which may lead to low iron levels or anaemia
- excessive bleeding after dental surgery or extractions, other surgery and medical procedures, injuries or accidents
- prolonged bleeding after childbirth
- joint or muscle bleeds (in females with very low clotting factor levels).

All females who carry the genetic change should have periodic testing for their clotting factor levels as they are unpredictable and can vary over a woman's lifetime. Factor levels can also vary between family members.

Women whose factor level is low will need a treatment plan to prevent bleeding problems and manage any situations that occur.

How is haemophilia treated?

Specialist haemophilia treatment services have a team of health professionals with expertise in providing treatment and care to people with bleeding disorders including haemophilia. They can work with you to make a treatment plan and advise on ways to live well with haemophilia.

The team includes:

- haematologists: doctors who specialise in blood disorders
- haemophilia nurses
- social workers or counsellors
- physiotherapists
- other specialist health professionals.

There is at least one specialist haemophilia treatment centre in every Australian state or territory, located in a major public hospital.

Your local haemophilia treatment centre team will help you learn how to recognize a bleed and deal with it promptly, and how to prevent and rehabilitate injuries and bleeds.

Your haemophilia physiotherapist will develop an exercise program for you that will help you to prevent bleeds and protect your joints and maintain a healthy body and mind.

Products for treating haemophilia

There are a variety of treatment products used to treat haemophilia. These are broadly classified as **preventative treatments** (where medicine is used to prevent bleeds – this is also known as prophylaxis) and **on-demand treatments** (where medicine is used to treat a bleed, or is administered just before an event such as surgery, dental treatment or childbirth).

Treatment options available include:

- **desmopressin** – a synthetic hormone that releases the body's stored factor VIII into the bloodstream to help blood clot. It can be given as a slow injection into a vein; as an injection into the fatty tissue under the skin; or as a nasal spray. Desmopressin is not suitable for everyone. It is used for treating some people with mild haemophilia A and some women with bleeding disorder symptoms
- **clotting factor concentrates** – these are concentrated amounts of clotting factor that can be injected (or

'infused') directly into a vein at home, or at a haemophilia treatment centre. They can be given on demand or as a preventative measure. There are two types – recombinant factor concentrate and plasma-derived factor concentrate:

- **recombinant factor concentrate** is the most widely used type of clotting factor concentrate. It is made by genetic engineering and contains little or no material from human blood or animals
- **plasma-derived factor concentrate** is made from the plasma (pale yellow fluid part) in human blood and is produced from blood donations. It is used when desmopressin is not suitable, or when it is likely the person will need treatment for more than two to three days. It is injected into a vein in the arm
- **tranexamic acid** is a medicine that stops blood clots being dissolved once they have formed. It can help to treat mouth or nosebleeds, gut bleeding or bleeding after dental work. Most commonly it is taken as tablets, syrup or in a mouthwash.
- **fibrin glue** is a medical gel made from fibrinogen and thrombin, which are proteins in the body that help blood to clot. It can be applied directly onto a wound to stop bleeding
- **hormone treatment**, such as oral contraceptives (birth control pills), can help women who have heavy menstrual bleeding. The hormones can increase factor VIII levels.

Inhibitors

After treatment with a clotting factor product, some people with haemophilia may develop antibodies – known as 'inhibitors' – which make treatment less effective. There are a number of ways to treat inhibitors. Many people are successful in overcoming them, although others have ongoing problems.

How often do people need treatment?

Current treatment for people with moderate or severe haemophilia aims to prevent bleeding, pain and joint damage.

In Australia the usual treatment for most children and young people with severe haemophilia is prophylaxis, where factor concentrate is injected two to three times a week to keep factor levels high enough to prevent 'spontaneous' bleeds.

People with mild haemophilia will not need prophylaxis and will only need treatment for a bleed when it occurs, usually after trauma or injury, or when they are preparing for surgery or some dental work.

Haemophilia and planning a family

For people with haemophilia or women who carry the gene, planning a family can raise a number of questions, such as:

- Will my children have haemophilia or carry the gene? And if so, how will this affect them?
- What are my options for planning a family?
- How can a mother who carries the gene plan for a safe pregnancy and delivery?
- Who will help with all of this?

Haemophilia treatment centres can help you with all your questions about having children.

It may also be helpful to speak to a genetic counsellor.

Haemophilia, pregnancy and childbirth

With good management, women who carry the altered gene causing haemophilia have no more problems with delivering a healthy baby than other mothers. However, it is very important to plan and prepare as much as possible.

This involves:

- ideally, if you are planning a pregnancy, talk to a haemophilia specialist, who may refer you to a genetic counsellor
- if a pregnancy is confirmed, contact your haemophilia treatment centre for advice on local obstetric services with experience of haemophilia
- ask your haemophilia and obstetrics teams to consult with each other to plan for a smooth and safe

pregnancy and delivery and care for your newborn

- check with your haemophilia treatment centre before having any invasive procedures, such as chorionic villus sampling or amniocentesis
- discuss suitable choices for anaesthesia, especially an epidural, with your haemophilia treatment centre and obstetrics teams. A normal vaginal delivery is usually recommended unless there are obstetric complications.

Identifying the sex of the baby

If you are pregnant and you know you carry the gene change causing haemophilia, it is recommended that you find out the sex of the baby before birth, preferably with a non-invasive test with minimal risk to the foetus, such as an ultrasound or prenatal screening test.

A male baby is more likely to be affected by haemophilia than a female. The woman and her partner can choose if they want to be told the sex of the baby or not before delivery even if the haemophilia and obstetric teams are aware.

If neither the couple nor the medical staff know the sex of the baby and no other genetic testing of the foetus has been carried out, the pregnancy and birth will be managed as if the baby is a male with haemophilia to ensure the baby is delivered safely.

Travel and haemophilia

Everyone has individual needs when travelling. If you are planning a trip, contact your haemophilia treatment centre a few months before departure to plan ahead. They can:

- help with preparations tailored to your situation
- advise on documentation, medication and travel insurance – for example, documentation to carry medication and treatment equipment through security and customs may be required
- advise you on what to do if you need treatment while travelling
- provide you with contact details of haemophilia treatment services along your route or at your destination in case of an emergency.

Be sure to get travel insurance for overseas travel.

You may also consider telling travelling companions what to do to help in case of an emergency.

Physical activity and haemophilia

Physical activity is good for everyone. For people with haemophilia it can strengthen joints and muscles and help prevent bleeds.

Speak to your doctor about what sorts of physical activity may best suit you. Everyone is different and has a different 'bleed history', so advice about physical activity needs to be individually tailored.

Think about what appeals to you and might not aggravate any injuries or problem joints you have. By choosing an activity that suits your needs, you will be able to perform better, reduce potential frustrations, and just enjoy your sport.

Rough, high-contact sports such as football and boxing aren't usually recommended as they may cause bleeds. Generally low impact or non-contact sports are most recommended. Some ideas include:

- swimming or hydrotherapy
- gym-based programs
- Wii Fit
- rock climbing (with a safety rope!)
- hiking
- rowing
- paddle boarding
- dance
- pilates or yoga

- surfing
- cycling.

Medical identification tags for haemophilia

People with haemophilia are advised to wear a Medical ID tag. This is a small tag worn on a bracelet, neck chain, or on your clothing. The aim is to alert people, in an emergency situation, that you have haemophilia and might require urgent medical attention.

Support for people and families living with haemophilia

Haemophilia treatment centres provide treatment and support for people living with haemophilia and their families.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by haemophilia.

Where to get help

- Your **GP (doctor)**
- **Your haemophilia treatment centre**
- **Haemophilia Foundation Australia** Tel. **1800 807 173**
- **Haemophilia Foundation Victoria** Tel. **03 9555 7595**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**

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