Understanding haemophilia

The Haemophilia Society

Contents

Introduction

This booklet is about haemophilia A and B. It gives a general overview of haemophilia and information on diagnosing, treating and living with the condition that we hope will answer your main questions. It has been written for people directly affected by haemophilia and for anyone interested in learning about haemophilia.

If you are a parent and your child has recently been diagnosed with haemophilia you may be feeling quite overwhelmed. Remember, you're not alone and many families are facing the same concerns and issues. Please do get in touch – we have lots of support and information available as well as services for parents and children. You can find out more via our website or Facebook pages, by emailing **info@haemophilia.org.uk** or calling us on **020 7939 0780**.

The outlook is now the best it has ever been for people with haemophilia in the UK. Scientific advances in understanding haemophilia have led to the development of effective treatment. Modern treatment allows children to grow up with the opportunity of a good quality of life and every prospect of fulfilling their potential.

Each person with haemophilia has their own experience with the disorder. What happens with your child may be different to another child's experience.

Haemophilia and your child

Having just found out your child has haemophilia can leave you feeling shocked and overwhelmed. You may experience many different feelings, including worry and sadness, and you may wonder how you will cope. There's also a lot of information to take in, often all at the same time, about their condition and treatment. You may find it useful to write down questions that you want to ask at your next haemophilia centre appointment.

Your haemophilia team know that this learning takes time and will help you to prioritise the most important information. For example, at first you really need to know the main signs of bleeding that you should be looking out for and who to contact for advice, day and night. With time and experience you will soon learn to recognise the signs of bleeding and judge what to do, though the haemophilia team is always there to offer support.

Because haemophilia is rare, you and your child may feel isolated and alone and it can be helpful to be put in touch with others in a similar situation. It's important to hold onto the fact that with modern treatment a child with haemophilia has every chance of growing up as an active, fit child who can participate fully in family, school and working life. It's also important to remember that your child's siblings can feel they aren't getting as much attention, and can become distressed, angry or isolated, so they need to feel included and able to have time with you too.

It's natural to want to protect your child as much as possible, especially when they are very young. But letting them find their own boundaries, within reasonable limits, is essential for building their self-esteem. Most parents find they become more relaxed over time as they learn more about haemophilia and become more confident in managing it effectively.

Brothers and sisters should be included in discussions about haemophilia and trips to the centre. This makes haemophilia less scary for them too, which is important when thinking about home therapy. Sisters may also be carriers and having a good experience with their brother's haemophilia may help them when they think about having children themselves.

Call your haemophilia centre if you have any concerns about your child. Make sure you:

- have the contact numbers in your phone
- give the contact information to others involved with your child's care.

Each person with haemophilia has their own experience with the disorder. What happens with your child may be different to another child's experience.

What is haemophilia?

Haemophilia is a lifelong inherited bleeding disorder. In haemophilia one of the clotting factor proteins important for blood clotting is either partly or completely missing. People with haemophilia take longer than normal for bleeding to stop. They may have bleeding into joints and muscles without having had an injury, so treatment is aimed at reducing spontaneous bleeding.

There are two types of haemophilia:

- haemophilia A is a deficiency of factor VIII (8)
- haemophilia B (also known as Christmas Disease) is a deficiency of factor IX (9).

Both types of haemophilia have the same symptoms and are inherited in the same way, though treatment is different depending on which clotting factor is missing. Specialist blood tests are needed to measure the clotting factors to show whether factor VIII or factor IX is affected and how much is missing.

What causes haemophilia?

Haemophilia is an inherited condition. The genes responsible for producing factor VIII and IX are on the X chromosome.

Females have two copies of the X chromosome and males have one X chromosome and one Y chromosome. The mother produces an egg containing one X chromosome. The father produces sperm, which could contain either an X or a Y chromosome. If the father contributes his X chromosome, a girl is conceived. If he contributes his Y chromosome, a boy is conceived. If a man has an altered haemophilia gene on his X chromosome, then he will be affected with haemophilia. If a female has an altered haemophilia gene on only one of her X chromosomes, then she is known as a carrier.

The term carrier can be misleading because some females who are carriers of haemophilia may have significantly reduced factor levels, which means they have a mild form of haemophilia themselves. You may hear doctors use the term heterozygote instead of carrier. The pattern of inheritance is known as sex- or X-linked recessive.

In some cases of haemophilia there is no known family history. This may be because the alteration to the haemophilia gene is new, known as a spontaneous mutation, or that no affected males have been known in the family.

Can females have haemophilia?

Females **can** also be affected with haemophilia. Many females often don't show symptoms of haemophilia. Some females who are carriers of haemophilia have reduced factor levels, which means they have a form of haemophilia themselves. They can also have symptoms such as frequent and prolonged nosebleeds, heavy or prolonged periods (menorrhagia), prolonged bleeding from cuts and easy bruising.

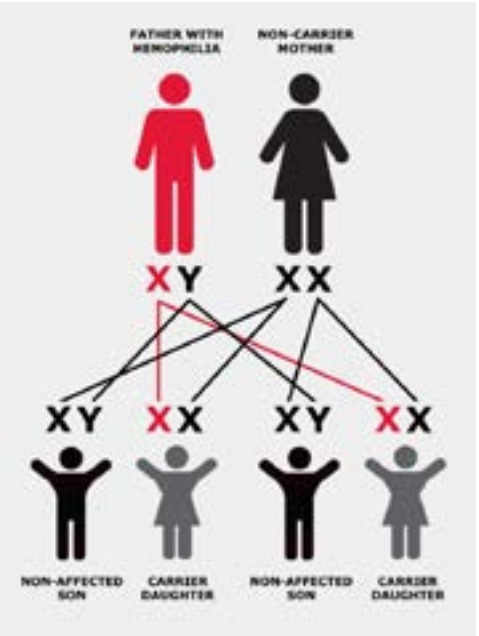
Symptomatic females should be defined as having haemophilia of a specified severity, like males with haemophilia.

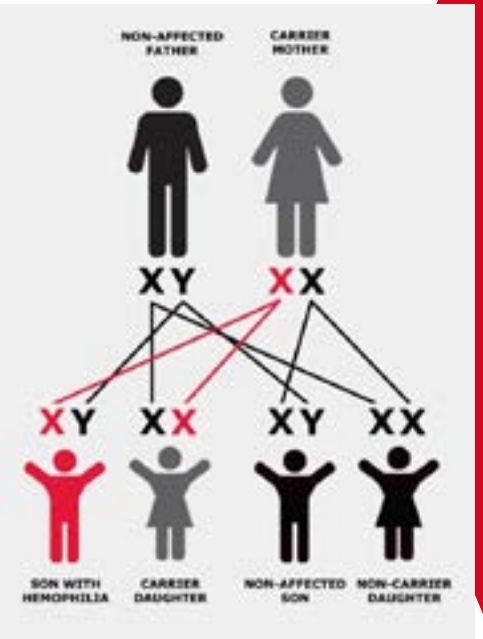
What is a chromosome?

Each cell of the body contains structures called chromosomes. A chromosome is a long chain of chemicals known as DNA. This DNA is arranged into hundreds of units called genes that hold the instructions for making proteins such as clotting factors and such things as the colour of a person's eyes.

Each cell contains 46 of these chromosomes arranged in 23 pairs. One pair is known as the sex chromosomes because they determine a person's sex.

Sex inheritance pattern





Summary of inheritance chances

Chance of a carrier having a **son** with haemophilia	Each of her **sons** will have a 50% (1 in 2) chance of having haemophilia
Chance of a carrier having a **daughter** who is a carrier	Each of her **daughters** will have a 50% (1 in 2) chance of being a carrier
Chance of a man with haemophilia having a **son** with haemophilia	None – unless the mother of his **son** is a carrier
Chance of a man with haemophilia having a **daughter** who is a carrier	All his **daughters** will be carriers

Who might be a carrier of haemophilia?	
Obligate carriers	**Possible**
Any biological daughter of a father with haemophilia Any biological mother of a child with haemophilia who also has at least one other family member with haemophilia (brother, maternal grandfather, uncle, nephew, or male cousin) or who is a known carrier of haemophilia (mother, sister, maternal grandmother, aunt, niece, or female cousin) Any biological mother of two or more children with haemophilia	Any biological daughter, sister, mother, maternal grandmother, aunt, niece or female cousin of a carrier of haemophilia A biological mother of a child with haemophilia and no known family history of haemophilia or carriers of haemophilia

Carriers

A propotion of females will have low levels of factor VIII or IX which can result in mild, moderate or even severe haemophilia in rare instances.

It shouldn't be assumed that a female who has grown up with a father with haemophilia understands that she is a carrier, or that a sister or cousin of a male with haemophilia is aware that she might be a carrier. As they grow up females need to be given information relevant to their age using language they can understand.

There are two different types of blood testing for haemophilia carriers. There are also different issues involved in deciding when they should be done.

Testing factor VIII or factor IX levels

This test is done to find out if a female has a low factor level and will therefore tend to bleed more than normal. This is important information for her own health and safety if she has an accident or needs an operation. She can then have the right advice and treatment to prevent bleeding. It's also helpful to know before she starts her periods so that advice and support can be in place if she has heavy periods.

Testing the factor level is therefore recommended for all females who are or may be carriers. A low factor level on its own suggests that a female may be a carrier, but the test doesn't show genetically whether or not she is. Likewise, a normal level doesn't mean that a female isn't genetically a carrier. Until the factor level test is done it should be assumed that a female may have a low factor level and any doctor consulted should be told this as action may need to be taken when having surgery or with a major injury.

Tests to find out if a girl or woman is a carrier

In most cases the type of alteration in the haemophilia gene that has caused haemophilia in any family can be found by examining the DNA in a person with haemophilia. This then makes it possible to see whether a related female carries the same gene alteration. If the alteration is found, then she is a carrier.

Unlike factor testing, it's less clear when this test should be done. Views differ on the advantages and disadvantages of knowing at an early age. This is an issue that should be discussed with the haemophilia team. However, it is important for young females to know before pregnancy.

Who is affected by haemophilia?

Haemophilia A affects about 1 in 5,000 of the male population. Haemophilia B is rarer, affecting around 1 in 30,000 of the male population. Around 30-50% of females who are carriers may have milder forms of haemophilia. Haemophilia affects people of all ethnic origins and from all parts of the world.

How severe is haemophilia?

Haemophilia is classed as severe, moderate or mild depending on how much clotting factor is missing. The level of factor VIII or factor IX in the blood is measured by a specialist laboratory. In general, the lower the level, the more bleeding problems the affected person will have without treatment.

Classification	Level of factor VIII or factor IX in the blood (normal: 50 to 150%)	Typical bleeding tendency
Severe	Less than 1% of normal level	• Easy bruising including from mouth and nose • Bleeding into joints and muscles, which can be without obvious cause • Bleeding after dental or surgical procedures or injuries including minor bumps and knocks
Moderate	1 to 5% of normal level	• Easy bruising • Bleeding because of minor injury • Occasional spontaneous bleeding • Likely to have problems after having dental or surgical procedures and/ or a bad injury
Mild	Over 5% of normal level	• Easy bruising • Bleeding usually only occurs following injury, surgical or dental (tooth extraction) procedure • Might never have a bleeding problem requiring medical attention • Might not be diagnosed until later in life if not playing contact sports orhave not had any injuries or operations • Females may also have heavy or prolonged periods (menorrhagia)

By understanding the severity of your child's haemophilia and their pattern of bleeding, over time you will know what is likely to cause bleeds, how they will affect them and how best to help.

Signs and symptoms of haemophilia

People with haemophilia don't cut more easily or bleed more quickly than normal. They do bleed for longer.

Cuts and scratches

In most cases minor cuts and scratches aren't a problem. A little pressure is usually enough to stop the bleeding.

Bruises

Bruises are common when children start to crawl or walk. They may get bruises on their knees and elbows as they progress to crawling and standing, or if they fall or roll onto a hard toy or surface.

Once they begin to stand and walk, they may get bruising on their buttocks if they fall or sit down suddenly. The bruises may look serious, but they don't usually need any treatment. However, if the bruise is swelling and painful then treatment may be helpful (see Joints and muscles below).

Prolonged bleeding

People with haemophilia commonly have prolonged bleeding following larger cuts or minor surgery such as having a tooth out or a circumcision. This can last for several days. There's no reason why anyone with haemophilia shouldn't have surgery with the correct treatment.

Joints and muscles

In severe haemophilia the main problem is internal bleeding into joints and muscles. We all damage our tissues in small ways in the activities of everyday life and most people repair that damage automatically.

With severe haemophilia, the tiny breaks in the blood vessels in joints and muscles may continue to bleed. These bleeds are sometimes described as 'spontaneous' because it's impossible to identify an obvious reason such as a bump or a fall.

Small children may not be able to describe their pain, so some signs to watch for include:

- crying

- favouring a limb – a baby may hold their bottle with the opposite hand from usual; a toddler may use their opposite hand to eat
- irritability
- refusing to walk – they may not want to move or may walk trying to avoid putting weight on the affected leg

Older children may describe:

- discomfort
- difficulty moving
- pain
- stiffness
- swelling
- tingling inside the joint
- warmth

Joint bleeding

- Knees, ankles and elbows are most affected.
- Usually starts by feeling stiff, tingly, bubbly or warm – these sensations are felt before there are any external signs.
- Becomes increasingly painful as the joint fills up with blood.
- May become swollen, warm and difficult to straighten.

Bleeding into the joint has a damaging effect on the joint. Once a joint becomes damaged bleeding may occur more frequently and damage can be permanent. This is known as a target joint.

Muscle bleeding

- An affected arm or leg may become swollen, tender and painful.
- Bruising near the surface may not be obvious at first.
- In deeper muscles swelling can press on nerves or arteries causing numbness and pins and needles. This needs urgent treatment and medical attention as there is a risk of permanent damage.
- May also happen to someone after an accident or sporting injury.
- May not be noticed or be uncomfortable at first.

Blood in the urine

Blood in the urine may be red or brown. It may go away with drinking plenty of fluid.

However, it may be a sign of infection and treatment may be needed to stop the bleeding.

Serious bleeding

Some types of bleeding are serious, such as head, neck and face injury. These need immediate treatment and should be assessed at hospital.

A head injury is always serious if a child is knocked unconscious. Bleeding into the brain is uncommon but can occur without an obvious injury. Symptoms include:

- confusion
- dilated or unequal pupils
- drowsiness
- headache
- irritability
- lethargy
- nausea and/or vomiting
- unsteady walk
- weakness in an arm or leg.

Rarely, babies with severe haemophilia can develop bleeding in their brain. It's therefore important to be aware that the following are possible signs:

- difficulty with feeding
- irregular breathing
- irritability
- seizures
- sleepiness
- vomiting.

Neck bleeding is serious because of the possibility of blocking the airway. Pain and tenderness should be attended to promptly.

Other bleeding

- Vomiting blood
- Coughing up blood
- Blood in their poo (that may look like blood or be black and tar-like) is a sign of bleeding in the gastrointestinal tract.

It is important to contact your haemophilia team straightaway if there are signs or symptoms of any of the above.

How is haemophilia diagnosed and treated?

Diagnosis

The diagnosis of haemophilia may be expected or suspected where there is a family history, or it may be completely unexpected. The following investigations lead to the diagnosis:

- history, signs and symptoms of bleeding
- family history of bleeding
- family history of haemophilia
- blood tests – a general test of blood clotting called a clotting screen which can be performed at all hospitals may suggest haemophilia and lead to referral for specific tests for factor VIII and factor IX.

Known haemophilia in the family

If there is a history of haemophilia in the family, it's possible that parents will have had contact with the haemophilia team at their nearest centre and will have had theopportunity to talk about the options available to them before or during pregnancy. They will also often have known their baby's sex before delivery.

If the haemophilia is severe in the family, then the delivery should have been carefully planned and a sample of blood taken from the baby's umbilical cord shortly after birth to measure the factor VIII or factor IX level. Even with the knowledge that there is a 1 in 2 chance of a baby boy having haemophilia, it can take time to get used to the diagnosis.

While the baby's mother is likely to have a good understanding of the haemophilia in her family, this will be affected by family experience and may not be up to date with current treatment. The baby's father may not have full knowledge of haemophilia and will seek guidance. The haemophilia team will aim to answer questions and give clear explanations to both parents.

If there is a family history of moderate or mild haemophilia, a blood sample can still be taken from the baby's cord. However, since factor VIII and factor IX levels may not reach their normal baseline until the baby is six months old, the blood tests may need to be done again at that stage. This will give a clearer picture of how the child is likely to be affected by haemophilia.

No history of haemophilia in the family

For at least one-third of people newly diagnosed with haemophilia there is no family history. The diagnosis may have taken time and can be a traumatic experience. Severe haemophilia will tend to be revealed by bruising when an affected baby starts to crawl or is learning to stand.

However, it may come to light earlier, such as in these examples:

- May have had bleeding outside his skull or within his brain after the birth, particularly if the delivery was long or complicated. Haemophilia will have been diagnosed during the many tests he had. For the family trying to take in this information while managing their anxieties around his general condition, this will have been very difficult.
- May have needed surgery or a procedure that caused an unexpected amount of bleeding. This may have led to further investigation and a diagnosis of haemophilia.
- May have had bruising with no obvious cause, which led the family to take him to their GP or the hospital for advice. This may have led to blood tests, diagnosis of haemophilia and referral to the nearest haemophilia centre. But as bruising in a baby who is not yet crawling raises concern about the possibility of non-accidental injury (when an adult is harming a child) sometimes children's doctors and social workers become involved before the blood tests reveal the haemophilia diagnosis. If there is a delay in reaching the diagnosis and child protection proceedings have started, the family will not only have to come to terms with the diagnosis but also with the anger and distress following the suggestion of non-accidental injury.

Moderate and mild haemophilia may not be diagnosed until later in childhood or in some cases in adulthood. The diagnosis may only be suspected if there is bleeding after surgery or having a tooth out, or after an injury.

Treatment

Different types of treatments are available for managing haemophilia and may vary depending on how severe it is. Prompt, effective treatment and prevention of bleeding significantly reduces the risk of complications and disruptions to school, work and family life.

Clotting factor concentrates (CFCs)

Bleeding can be controlled or prevented by replacing the missing clotting factor in the blood through an infusion of clotting factor concentrate. The level of factor VIII or factor IX is increased temporarily so infusions need to be repeated.

For some minor bleeding episodes one infusion may be enough to stop bleeding. For more serious bleeding or where the first infusion has been delayed, treatment may be needed once or twice a day until the bleeding stops.

Recombinant factor VIII and factor IX concentrates are made using genetic technology and aren't made from blood.

Treatment can be given in two ways		
On demand	Treatment is given when bleeding occurs such as after an injury	• Very young children • Some adults with severe haemophilia • People with mild or moderate haemophilia
Prophylaxis	Treatment is given regularly to prevent bleeding before it starts	• Children with severe haemophilia • Some adults with severe haemophilia • Some people with moderate haemophilia who have frequent bleeding problems

Treatment will be needed by everyone with haemophilia if they are bleeding. It will also be necessary to prevent bleeding if they need an operation or have an injury where there is a risk of bleeding.

Clotting factor concentrate is given intravenously (into a vein) into the bloodstream through a needle. Children can have anaesthetic cream rubbed onto the skin before an injection to reduce any pain. The puncture caused by the needle is small and does not result in excessive bleeding.

Standard half-life (SHL) treatment

Standard half-life factor concentrates stay in the blood for a relatively short time and require repeat injections for prophylaxis – every 2-3 days for haemophilia A and twice a week for haemophilia B.

Extended half-life (EHL) treatment

These are factor VIII or FIX products that can stay in the bloodstream for longer. If this treatment is suitable for your child, they may be able to have injections to prevent bleeds (prophylaxis) less often or may stay on the same dose/schedule and have higher trough levels. They are likely to need fewer injections to control any bleeds they have. Half-lives are shorter in children and young people than adults.

Subcutaneous treatment

Emicizumab is a subcutaneous (under the skin) treatment used to prevent or reduce bleeding in people with severe haemophilia A that is given weekly, two weekly or once a month. It is an antibody which mimics the action of factor VIII.

Emicizumab can be prescribed for very young babies.

However, the clotting system doesn't fully mature until 6 months of age, so you will need to discuss with your haemophilia team what the best option for you and your child is. Emicizumab is not a treatment for bleeds, serious injury or major surgery; factor VIII would need to be given to treat bleeds.

For more information please visit our website:
https://haemophilia.org.uk/resources/publications/factsheets/

Moderate and mild haemophilia may not be diagnosed until later in childhood or in some cases in adulthood. The diagnosis may only be suspected if there is bleeding after surgery or having a tooth out, or after an injury.

Prophylaxis

Treatment with regular infusions of clotting factor aims to prevent the long-term damage caused by bleeding into joints and muscles. Prophylaxis works by stopping the bleeding that occurs without any obvious injury, often called spontaneous bleeding. It also reduces the risk of bleeding from minor injuries.

Research has shown that prophylaxis gives children the best chance to reach adulthood without damage to their joints. How often the infusions need to be given is decided for the individual but is typically every other day for haemophilia A and 2-3 times a week for haemophilia B because factor IX lasts a bit longer in the blood.

Home treatment

Most people with severe haemophilia and some with moderate haemophilia learn to treat themselves/their children with clotting factor at home. Home treatment has many advantages:

- makes regular treatment possible
- treatment can be given as soon as possible after bleeding starts
- there is less disruption to home, school and working life
- greater independence and control for the parent/person with haemophilia
- reduced need for hospital attendances
- early treatment reduces recovery time and risk of permanent damage.

At first parents are taught by a specialist haemophilia nurse how to give treatment to their child. In time children will learn how to self-treat – most can do this by the time they reach secondary school.

A butterfly needle is inserted into a vein in the hand or arm, the clotting factor is infused and the needle is removed. Alternatively, in very small children or those whose veins are quite difficult to access, an implantable port-a-cath can be inserted to make having the injections easier.

This is a small device inserted under the skin (under general anaesthetic in the operating theatre). Part of the port has a small container called a reservoir and this is placed under the skin. A special needle can be put into the port through the skin so the factor can be injected.

Other treatments

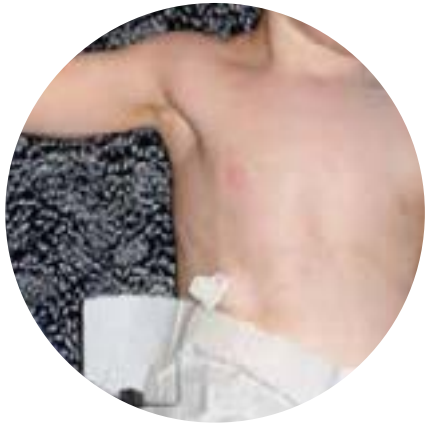
Desmopressin (DDAVP®)

DDAVP® is a synthetic drug that can be suitable for some people with milder forms of haemophilia A. It releases factor VIII stored in the lining of blood vessels, increasing the amount of factor VIII circulating in the blood.
This increase can be enough to control minor bleeding episodes and to prevent bleeding from minor operations including dentistry. It is given as a subcutaneous injection (under the skin like a vaccination). It can't work for severe haemophilia as there are no stores of factor VIII.

Tranexamic acid

Tranexamic acid is a medicine that helps to hold a clot in place once it has formed. It comes as a liquid or tablet and can also be used in a mouthwash. It can be particularly helpful for bleeding in the mouth, nosebleeds or heavy periods. It's often used at the same time as clotting factor or DDAVP® but can be used on its own.

Port-a-cath



Port-a-caths are used in children with severe bleeding disorders. This is a small device surgically inserted under the skin. This device is a more comfortable way for children to receive their treatment rather than into a vein.

Haematomas or bruises can occur on the surface of the port after treatment is given. Children with ports are closely monitored if they have a fever, especially if it is not due to a childhood illness, as there is a risk of infection. If a port infection is identified, the child will be given a course of antibiotics. The infection is likely to be treated in hospital for a few days.

Children can play sports with a port, though it is best to avoid contact sports as a knock to the port could be painful or could cause damage. Swimming is allowed with a port-a-cath.

Managing joint bleeds with PRICE

It is very important not to ignore the signs of a joint bleed. Early treatment with factor concentrate is crucial to reduce the risk of joint damage.

Alongside the clotting factor infusion and pain relief (paracetamol), the following steps help with recovery and comfort.

- **Protection** Try to keep the weight off the joint or muscle for a couple of days. If it's the ankle or knee, try to use crutches.
- **Rest** Stay off it or don't use it. This helps with healing.
- **Ice** For 10-15 minutes every two hours. This can help with pain and swelling. Wrap an ice pack in a tea towel or soft cloth so it doesn't directly touch the skin because it can cause ice burns.
- **Compression** Use the right size of elasticated bandage if this feels comfortable. It can help reduce swelling.
- **Elevation** Raise and support the affected arm or leg to reduce swelling.

As your child grows older, they will come to know the sensation associated with bleeds and will usually be able to tell when they are having a bleed. If you are unsure your child has a bleed check with your haemophilia team. It will give you peace of mind even if you find out it was a false alarm.

Gene therapy

Gene therapy is a treatment technique that uses genes or genetic material to treat or prevent disease. Following extensive trials for gene therapy in haemophilia this has now become available, but only for adults.

Children are not yet included in trials of gene therapy. Researchers need to understand the long-term impact of gene therapy in adults before it is given to children, as the effect could last for their entire lives.

Children are at earlier stages of development and some processes, like the immune response or how proteins are produced, may be accelerated or different in a child. It's possible that at some stage in the future clinical trials involving children will be developed and become available.

Prophylaxis is also recommended for children with severe haemophilia in order to decrease the number of bleeding episodes and to help protect against joint disease. Gene therapy will not cure joint damage, so it is vital that all joint bleeds are managed and rehabilitated to prevent long-term damage such as a target joint.

Possible complications of haemophilia

Inhibitors

An inhibitor is an antibody that the immune system (the body's defence mechanism) develops when it recognises the clotting factor being infused as something foreign. The inhibitor removes the clotting factor before it can work to stop any bleeding. Inhibitors usually affect people with severe haemophilia A in early childhood, but they can also occur later in life in milder forms of haemophilia. They are much more unusual in people with haemophilia B.

Sometimes higher doses of clotting factor can have some effect in stopping bleeding but usually an alternative treatment is needed.
There is also treatment called:

Immune Tolerance Therapy (ITT)

Very high frequent doses of factor are given, which aims to get rid of the inhibitor so that the person with haemophilia can return to standard treatment. People who develop an inhibitor can get specialised advice and support from a haemophilia centre team that has expertise in this area.

Emicizumab (for people with an inhibitor)

This is available for people with haemophilia A who have a factor VIII inhibitor. It can be used as prophylaxis to prevent bleeds. It is not a treatment for bleeds, injury or major surgery. It is given as an injection just under the skin either weekly, fortnightly or every 4 weeks.

Joint and muscle damage

Bleeding can cause permanent damage to muscles and joints. This is why regular prophylaxis and prompt treatment of any bleeding is so important. When bleeding has affected a joint or muscle, physiotherapy is crucial for good recovery.

Damage to a joint can be caused by one serious bleed, though normally it's a result of repeated bleeding into the same joint over several years. The bleeding damages the synovium (the lining of the joint) as well as the cartilage and surrounding tendons and tissues.

This leads to arthritic pain and loss of movement and strength in the joint. In general, the older someone is the more likely they are to have joint damage. Orthopaedic surgery, including joint replacement, can be a successful option when a joint is badly damaged.

As your child gets older it is important to encourage them to be physically active to protect their muscles, bones and joints.

Medical and dental treatment

Surgery

People with haemophilia can have any surgery they need but careful planning is essential. The aim of the haemophilia team will be to provide safe management plan so that there's no greater risk of bleeding than usual for the type of surgery.This is important in supporting good healing and for the best possible results from the surgery. This means that clear communication with whoever is carrying out that surgery or procedure is vital. The haemophilia specialist will advise what treatment is required for the haemophilia.

Depending on the type of surgery and the person's condition, planning may include deciding the best place for the surgery to be carried out. If surgery is unplanned, the patient's care team need to know about their haemophilia and speak to their specialist team before the operation.

Circumcision

Circumcision is quite a common surgical procedure. It may be done for medical, social, cultural, personal or religious reasons. It is often the earliest surgery performed in a boy's life and may lead to a diagnosis of haemophilia. In haemophilia a circumcision is associated with complications including prolonged bleeding, infection and delayed skin healing. Some males will bleed when stitches are removed. Early diagnosis and factor replacement are key to stop bleeding.

A circumcision should only be carried out after consultation with your haemophilia team. Controlling bleeding after the surgery and factor replacement with monitoring from your haemophilia team is crucial.

Dental care

Good dental education and care is particularly important for people with haemophilia to prevent tooth decay and gum disease and avoid the need for dental surgery.

Gum bleeding is caused by gum disease but will be worse for someone with haemophilia. Regular brushing will help prevent the build up of plaque and the development of gum disease.

Regular check-ups with a dentist and dental hygiene sessions are also essential. Any dental treatment will need to be planned by the dentist and the haemophilia centre. Haemophilia treatment isn't always necessary before fillings or hygiene sessions but may be before having a tooth out.

Depending on the severity of the bleeding disorder, your haemophilia team may recommend factor replacement therapy or tablets before a tooth is extracted. It is important to speak to your haemophilia centre before any dental treatment is needed.

Medicines

Some medicines can affect blood clotting and so may not be suitable for someone with haemophilia. These include aspirin and ibuprofen, which should never be taken unless advised by a haemophilia specialist. Any new medicine, including ones that can be bought over the counter and herbal preparations, need to be considered for any increased risk of bleeding.

Paracetamol is a suitable painkiller for people with haemophilia and can be used for treating pain and fever in children.

If you are uncertain about whether any medication may affect blood clotting, it is important to check with your haemophilia team before taking it.

Vaccinations

If vaccinations are given at school or at the GP, they need to be administered subcutaneously (under the skin) rather than into a muscle. It is important the school and parents liaise with child's haemophilia centre for advice before any vaccination is given.

Bleeding disorder card

Children should have a 'bleeding disorder card' that is provided by their haemophilia centre. This will detail their bleeding disorder, treatment required and the haemophilia centre's contact details. Haemophilia centres should be able to provide you with as many cards as required, for example so that one can be left at school at all times (with medicines if appropriate).

Living with haemophilia

Sport and exercise

Exercise and sport have many benefits for health and can improve self-esteem, learning and concentration. There are some particular benefits for children and adults with haemophilia as strong muscles, good balance and posture can help to protect joints from bleeding. Maintaining a healthy weight helps to reduce stress on joints that have already been damaged by bleeding.

The choice of activity or sport will be individual, and the specialist haemophilia physiotherapist is a good source of advice. The haemophilia team will discuss the risks and benefits of different sports taking account of the person's condition. In general, sports involving a lot of physical contact and those where head and neck injuries occur carry the highest risk of injury and therefore risk of bleeding. Prophylaxis can be tailored around days of highest activity so that there is maximum protection from bleeding at these times.

School, college and work

With modern treatment, haemophilia shouldn't have a serious impact on a child's education. It's important that the school has a good understanding of the possible problems and knows what to do and who to contact – our guide on bleeding disorders and school will help. Your haemophilia nursing team will liaise with the school and if you choose, can visit in person to provide advice.

There may be times when a child has to miss school while they recover after a bleed. They may need support to catch up on missed work and get back into the daily routine of classes. Rarely, a child may temporarily need to use a wheelchair or crutches. The school should be aware of this and have the necessary arrangements in place.

In general, most school-based activities are suitable for children with haemophilia. Children should be encouraged to do all the things they can do rather than focusing on minor restrictions. What they can and can't do will not only depend on the severity of their haemophilia, but also on their age, interests and talents. However, there should be discussion between parents and the school about taking part in contact sports. Prophylactic treatment must be given on days when children have PE or sports sessions.

Nursery, pre-school and primary school

It's important that children join in all play activities with their classmates, especially at this age. Ordinary play activities aren't usually a problem beyond the occasional bruise. Cuts and grazes can generally be managed with standard first aid. Scissors and other sharp instruments don't need to be avoided – all children need to learn to use these safely.

Primary school sport isn't usually particularly competitive or rough. Unless a child has a particular problem or is recovering from a bleed, he they should be able to join in with all sports activities.

Secondary school

As young people get older, the range of sports and other activities they want to joinin gets wider. The sports and activities they enjoy also tend to get more competitive and rougher. It's important that young people have the right clothing, footwear and equipment for any activity.

The relative risks and benefits of different sports will vary for each individual and each sport. If a particular sport does cause a problem, the young person may need to find an alternative and seek advice from their parents, haemophilia centre and the school.

College and work

Most young people with haemophilia will go through school much as the rest of their peer group. This means that choosing to go to college or out to work won't be any different either.

College or a job brings new friends and workmates, employers and tutors. A young person with haemophilia has to decide who to tell and what to tell them about haemophilia.

This can be daunting – they may be worried that they will be treated differently if people know. This is an individual decision which means thinking about whether telling someone will make life easier or more difficult. It can be helpful to know there is some support and understanding if they have a bleed.

One good reason for telling a tutor or employer is so that a safe storage place can be available for some factor concentrate so that it's on hand in case of a bleed. If people don't know, it's worth considering how they will react if they find out later or from someone else.

Most people won't know much about haemophilia, so it can take time to explain and deal with their concerns. It may help to have some accurate written information to give them or refer them to our website.

More information is available in our 'bleeding disorders and school' booklet.

Travel

For anyone planning to travel, important considerations include the following.

- Taking up-to-date written medical information, including diagnosis details and the name and phone number of the haemophilia centre.
- Discussing travel plans with the haemophilia centre, including whether a supply of treatment should be taken and if so, how much.
- Getting a letter from the haemophilia centre explaining about carrying treatment in hand luggage and though security checks.
- Finding out the contact details of haemophilia centres in the places being visited. Information is available from the haemophilia centre, the World Federation of Hemophilia (**www.wfh.org/en/home**), or call us on 020 7939 0780.
- Getting travel insurance that covers haemophilia, including repatriationcosts.
- Carrying identification about haemophilia such as a bleeding disorders card or MedicAlert bracelet or necklace.

Our travel information section on our website has lots of tips on travelling with a bleeding disorder including travel insurance:
https://haemophilia.org.uk/support/day-day-living/travel/

Disability Living Allowance

What is DLA?

Disability Living Allowance (DLA) is a government benefit aimed to help you with the extra costs of raising a child with a medical condition or disability. It can be claimed for children under the age of 16 as long as your child has a disability, illness or health condition severe enough they

- need much more looking after than a child of the same age without a disability, or
- have walking difficulties or both

Your child must have had these difficulties for at least 3 months and expect them to last for at least 6 months.

You won't be awarded DLA because your child has a bleeding disorder. You will need to show how their bleeding disorder affects your lives and that they have additional care needs.

For more information our DLA Factsheet is available to download on our website:
https://haemophilia.org.uk/resources/publications/factsheets/

Transition to adult care

Transition is a gradual process that gives you, and everyone involved in your care, time to get you ready to move to adult services and discuss what your healthcare needs as adult are likely to be. Transition is about making plans with you and your child.

As your child grows older they will prefer to be seen in a more grown-up environment, rather than the usual children's departments or wards.

There is no exact time to move that is right for everyone. Your doctors and nurses may have an idea about when they feel you might be ready but it is important that you are involved in that decision.

Part of the transition process should be helping your child to look at where their ongoing healthcare needs can best be met and how this will fit with their future plans. The healthcare team at your haemophilia centre will be able to give you and your child information and support about moving on.

While transition is all about the young adult, it is important to realise that parents may also find the process difficult as they are now handing the responsibilty over to their child. Sharing how you are both feeling will help you all through the process.

Questions you may like to discuss with your healthcare team:

- What is the plan for my child to transition?
- When are they moving to adult services?
- Can we choose which adult service to move to?
- What is different about the adult service?
- Can we meet the adult staff before leaving the children's services?
- Can we visit the adult service to look around?
- Are there any young people they can talk to about moving to adult services?
- What do I need to know before my child moves to the adult service?
- When can my child start getting more involved in their health care?

By talking about transition early, you and your child should have enough time to preapre questions and have discussions, to make sure they are ready to move to adult services when the time arrives.

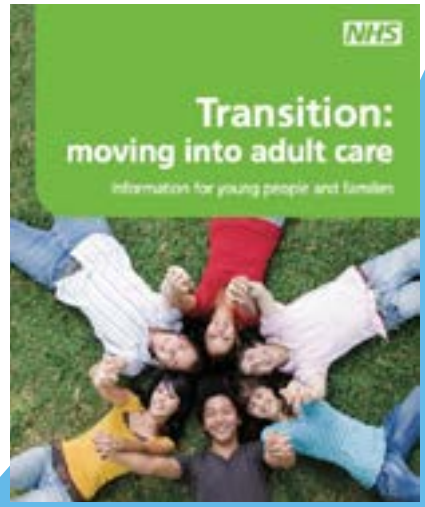
Challenges of transition

- Anxiety around meeting a new team of health professionals
- The logistics of finding your way around a new hospital
- Leaving behind the health care professionals with whom a strong bond has been made.

Benefits of transition

- Excitment for a new start
- Positive step for those who feel they have outgrown the children's hopsital setting
- A fresh start for your child who may have disengaged with the paediatric centre.

For more information on transition to adult services, please see the **Ready Steady Go programme, https://www.readysteadygo.net/**



Pregnancy and haemophilia

Prenatal diagnosis is usually offered to help with reproductive planning and risk assessment.

If you know that haemophilia runs in your family, you may wish to have a test during pregnancy to find out the sex of your baby.

Free foetal DNA (ffDNA) is a blood test arranged by your haemophilia centre to find out the sex of your baby during the early stages of pregnancy.

As early as nine weeks of pregnancy cells from your baby (foetal cells) can be detected in your blood and these are analysed to work out the baby's sex. The test results take about a week and your haemophilia centre will contact you as soon as they have the results.

If you don't want to know the sex of your baby your haemophilia centre can still do the test but inform your obstetric team without telling you the results. At this stage, some women will choose not to continue their pregnancy, this should be discussed with the haemophilia doctor.

If you are carrying a male foetus, you may then choose to have a test called chorionic villus sampling (CVS). CVS is a test where the doctor takes a sample of cells from the placenta. It is usually carried out between 11 and 14 weeks of a pregnancy.

To do the test, the doctor puts a fine needle through the wall of your abdomen or a thin tube into your vagina and up into the womb. They can then take a small sample of cells from the placenta for genetic testing.

An additional possible test is amniocentesis. This means taking a sample of fluid from the womb, from inside the membrane holding the baby. The fluid contains cells from the baby that can be genetically tested. Amniocentesis is usually done later in pregnancy than CVS, from between 15 and 20 weeks until near the end of pregnancy.

There is a risk of miscarriage with CVS and amniocentesis tests – about 1 in 100. Your doctor will talk you through all the possible risks and what the test can show before you decide whether you want to go ahead.

Management of care for all pregnant carriers should involve close cooperation between the haemophilia and obstetric teams.
It is important to have a clear plan for delivery that is shared with the mother to be and kept in her medical notes.

Preimplantation genetic diagnosis (PGD)

This may be an option available to couples who could have children affected by haemophilia. Couples can opt for PGD to make an informed decision before pregnancy.

PGD involves identifying the genes or chromosomes in embryos for gender and haemophilia. Embryos which are unaffected by haemophilia can then be transferred to the uterus.

Because the embryos need to be tested in a lab, IVF is needed, even if you and your partner have no fertility problems. Funding needs to be applied for and agreed before a referral can take place. The criteria to be eligible for NHS funding differs acorss UK regions so speak to your haemophilia team to get more information about this.

More information about pregnancy and bleeding disorders is available in our **'Women living with bleeding disorders'** booklet.

Other THS Publications

Additional literature is available and downloadable on our website - **https://haemophilia.org.uk/resources/publications/**. All our publications can also be posted to you throughout the UK.

- Understanding VWD
- Girls living with bleeding disorders
- Sex and bleeding disorders
- Ageing with a bleeding disorder - social care and support
- Rare bleeding disorders
- Dental care for adults with a bleeding disorder

Glossary of terms

Bleeds - Bleeding into the muscles and the space between joints

Circumcision - Surgically removing part or all the foreskin

Factor replacement - A treatment that replaces the low or missing factor in people with therapy haemophilia so that blood will clot if they have an injury

Haemophilia A - lifelong hereditary blood disorder in which people bleed more easily and bleeding lasts longer than normal. It is caused by a defect in a protein needed for blood clotting

Hereditary - Passed in the genes from parent to child. The basic unit of heredity is the gene

Inhibitor - Antibody to factor treatment

Joint - The place where two or more bones come together

Menorrhagia - Heavy periods with severe cramping

Obligate carrier - Daughter of a father with haemophilia

Prophylaxis - Treatment given regularly to prevent bleeding before it starts

Subcutaneous - A method to deliver medicine into the body that involves inserting a needle into the fatty tissue just beneath the skin

Target joints - Joint where bleeds occur most often.

Trough level - The lowest level that a medicine is present in the body

About The Haemophilia Society (THS)

We are the only UK-wide charity and free membership organisation for everyone affected by a genetic bleeding disorder.

We aim to empower people affected by a bleeding disorder to live life to the fullest; offering support, including events and local groups, the latest news and in-depth information resources, and campaigning and advocacy to demand the best possible care, safe and effective treatment, and equitable access for everyone affected by a bleeding disorder.

There are over 5,000 members of the Haemophilia Society, including people and families living with bleeding disorders, as well as healthcare professionals.
The charity's supporters help fundraise the costs that are vitally needed to be able to offer membership - and services such as events and printed publications - entirely free to all members.

What we do:
Support each other
We understand each other. We offer advice and support from personal experience. Our growing community is there for each other because we're in it together.

Raise awareness
We rally together because every little thing we do makes a difference and gives hope to people living with a bleeding disorder.

Make a lasting difference
We influence and advocate on what matters to our community. Health and social care policy, access to treatment and much more.

To find out more, or to become a member for free, visit our website at **haemophilia.org.uk** or call us on **020 7939 0780**.

Notes

This booked was reviewed by doctors from our Medical and Scientific Advisory Group.

The publication of this booklet has kindly been supported by Pfizer.

Special thanks to Haemophilia Nurse Specialists Julia Spires at Great Ormand Street Hospital for Children and Natlie Lawson at Birmingham Children's Hospital for their help with this booklet.

The Haemophilia Society makes every effort to make sure that its services provide upto- date, unbiased and accurate information about bleeding disorders. We hope that this information will add to the medical advice you have received and help you to take part in decisions related to your treatment and care. Please do continue to talk to your doctor or specialist nurse if you are worried about any medical issues.

Your Society: getting in touch
The Haemophilia Society
52b Borough High Street
London SE1 1XN
Phone: 020 7939 0780
Email: info@haemophilia.org.uk
Web: haemophilia.org.uk

HaemophiliaSocietyUK
HaemoSocUK
thehaemophiliasociety

Registered charity no. 288260 (Scotland SC039732)
Company limited by guarantee reg. no. 1763614
Members of the European Haemophilia Consortium and the World Federation of Hemophilia

Review date 2025