Living with Haemophilia

A guide to mild & moderate haemophilia

The information provided is not a substitute for professional and/or medical advice. Please consult a healthcare professional for further advice.

This brochure is intended for people with haemophilia, their carers and parents of children with haemophilia. It has been developed and funded by Takeda UK Ltd.

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Is this book for me?

This guide has been written for anyone who needs to learn about living with mild or moderate haemophilia. Whether you are a person with mild or moderate haemophilia, or you look after an adult or a child with mild or moderate haemophilia, we hope that this guide will enable you to understand a little more about the condition.

People with mild haemophilia often don't have many problems compared with those with severe haemophilia. In fact, many are totally unaware that they have haemophilia at all. Those with moderate haemophilia might bleed about once a month, although spontaneous bleeding is rare. In both mild and moderate cases, even though the blood clots more slowly than normal, the condition is only likely to cause concern after serious injury or during major surgery.

Remember, whatever your haemophilia type, whether you are male, female, experience symptoms or are a carrier, you are not alone. Haemophilia is a well-understood and manageable condition.

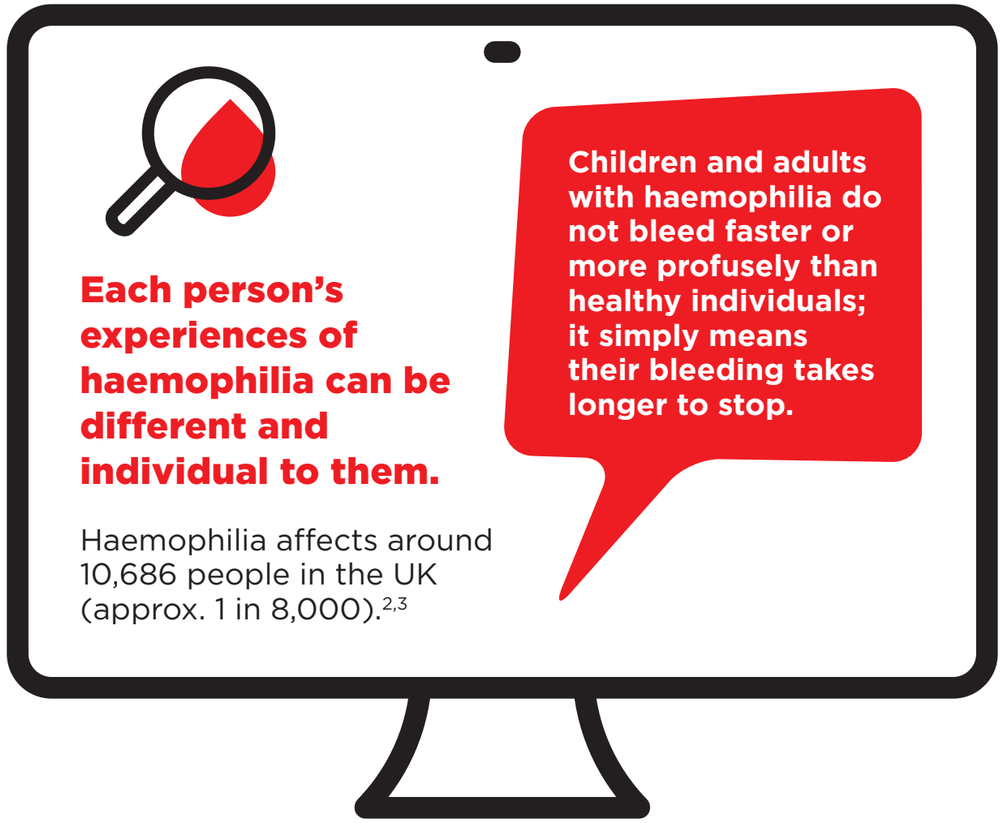
Contents

What is haemophilia?

Haemophilia is a rare genetic disorder, typically inherited from parents, although sometimes acquired (acquired haemophilia – AH), which makes it difficult for the body to form a blood clot and control bleeding. It is caused by a faulty gene that means the body is unable to make the important proteins, called clotting factors, it needs to form blood clots.[1]

This means:

- Blood takes longer to clot following an injury
- Knocks and bumps can also cause internal bleeding under the skin or into joints
- In the event of a bang to the head, bleeding can occur into the skull, placing pressure on the brain



Haemophilia is more often seen in males, but affects both sexes

100% of males with the faulty gene develop mild, moderate or severe haemophilia

30–50% of females with the faulty gene develop mild haemophilia[3]

Haemophilia runs in families

of haemophilia cases have a family history

of cases may occur spontaneously, or the mother may be unaware she carries a faulty gene[4]

There are two main types of haemophilia[1]

Haemophilia A – missing or reduced levels of a blood protein called Factor VIII (eight)

Haemophilia B – missing or reduced levels of a blood protein called Factor IX (nine)

Although haemophilia is a lifelong disorder, people with haemophilia can lead long, active and healthy lives.

Haemophilia severity and clotting factor levels[4]

The level of haemophilia severity depends on how many of these proteins, or clotting factors, are in your blood compared with somebody without haemophilia:

Mild: 6–40% of normal levels

Moderate: 1–5% of normal level

Severe haemophilia has less than 1% of normal levels

Once established, the severity of haemophilia does not usually change during a person's lifetime. People in the same family usually inherit haemophilia of the same severity.[1]

How is haemophilia inherited?

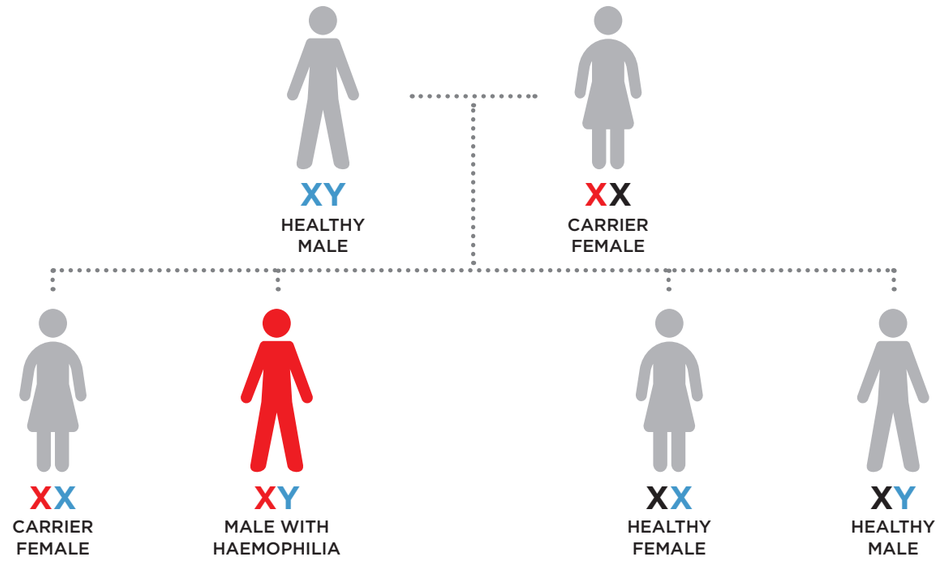
Haemophilia is an X-linked genetic disorder passed from mother to son on the X chromosome. If the mother carries the gene for haemophilia on one of her X chromosomes, each of her sons will have a 50% chance of having haemophilia.[5]

Less frequently a spontaneous mutation within the X chromosome of the foetus may cause the condition.[5]

This genetic basis means that haemophilia runs in families. If a child has haemophilia, any brothers or cousins may also be affected.[5] If the child has sisters, they may have inherited one affected X chromosome and so 'carry' the condition. Haemophilia carriers may occasionally show signs of mild haemophilia themselves, but this is unusual.[5]

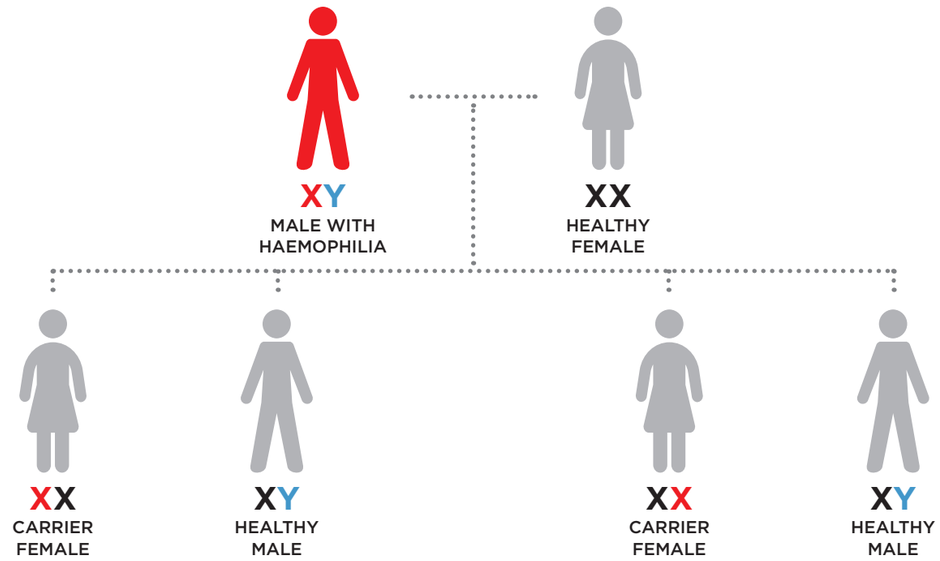
It is theoretically possible for a girl to also have haemophilia, but this can only happen if she is the child of a carrier mother and a father with haemophilia. This is very rare.[5]

These are the possibilities if a female carrier has children:



Adapted from: Cleveland Clinic. Hemophilia. Available at: https://my.clevelandclinic.org/health/diseases/14083-hemophilia. Accessed August 2022.[5]

And these are the possibilities if a man with haemophilia has children:



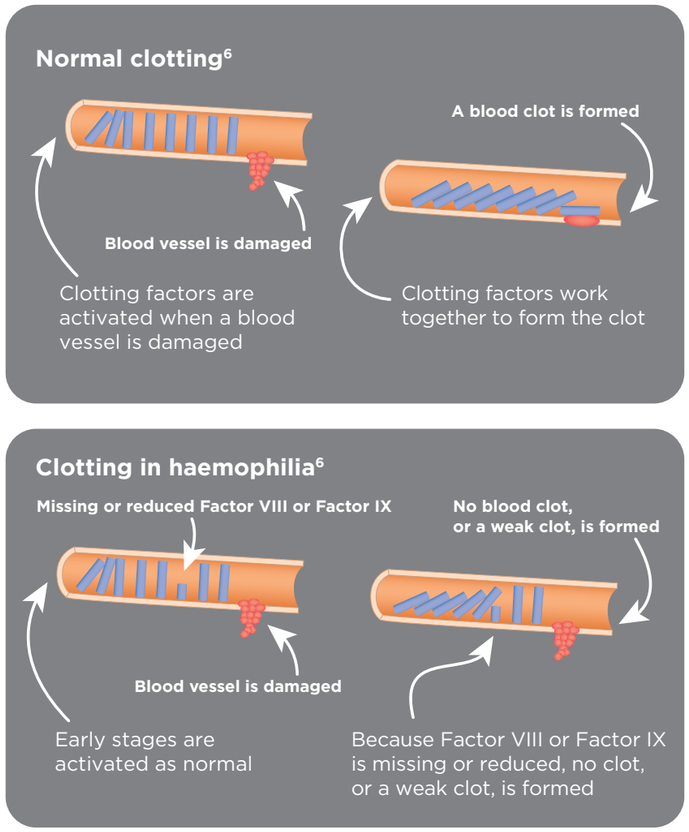
Adapted from: Cleveland Clinic. Hemophilia. Available at: https://my.clevelandclinic.org/health/diseases/14083-hemophilia. Accessed August 2022.[5]

All the daughters of a man with haemophilia will be carriers but none of the sons will have haemophilia. If you are not aware of any previous history of haemophilia in your family your doctor may recommend that you are all tested. This will not just help identify any carriers in the family; it will also spot any other not yet diagnosed with haemophilia.

How does blood clot and what happens in haemophilia?

Typically, when the body is injured, blood clots and forms a scab. This stops the bleeding, providing protection while the body repairs itself. In mild and moderate haemophilia, this still happens – but sometimes the clot is weaker. If it breaks down before the repair process is finished, the bleeding restarts.

Clotting is a complex process that takes several steps and involves a series of clotting factors.[6]

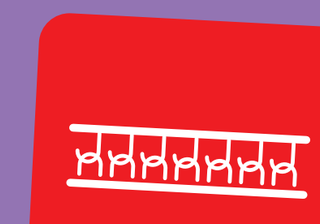


Figures showing how a blood vessel is damaged and how a blood clot is formed.

Why do clotting factors affect haemophilia severity?

The key difference between mild, moderate and severe haemophilia is the amount of clotting factor available. To illustrate this difference, imagine the amount of clotting factor is the same as the number of little hooks on a strip of Velcro®.

People with mild or moderate haemophilia often have enough clotting factor to cope with minor injuries and everyday life.[4] However, people with mild, moderate and severe haemophilia can experience repeated small, invisible bleeds into their joints that can cause damage over time.[7] That's why preventative treatment regimes, which regularly top-up clotting factor levels, may be recommended to prevent these bleeds and the joint damage they can cause.[7]



When there are a lot of hooks, the pieces stick together firmly and hold together well.

In mild and moderate haemophilia, however, there are fewer hooks and while the pieces can stick together, they just don't stick as well.

In severe haemophilia there are very few, or no hooks which means more hooks have to be added if the pieces are to stick together at all. This is why people with severe haemophilia need more regular treatment with factor than for mild or moderate haemophilia.

How might you feel about having haemophilia?

As a parent of a child with haemophilia

If your child has haemophilia, you are likely to worry about them. Some parents may feel guilty if they were carriers of a haemophilia gene. Don't worry, these are normal feelings and you are not alone.

Children need to be children
While you will need to be a little cautious, it is important to encourage your child to play and have fun. Bruises or bleeds can seem upsetting at first, but following the guidance from your haemophilia centre will help you to deal with incidents calmly and safely.

Talking can help
Perhaps approach the nurse at your haemophilia centre or other parents; your centre can give you contact numbers for the Haemophilia Society and local support groups. You can also find some useful haemophilia resources on page 25.

Involve the family
Helping your family to understand haemophilia, especially other children, can help avoid feelings of jealousy that may arise from the added attention. Try to help everyone understand that children with mild or moderate haemophilia can bruise more easily.

As a carer for someone with haemophilia

When caring for someone with haemophilia, keeping track of their treatment schedule and appointments can be challenging. You want to support them to stay active and healthy, and ensure they have access to the care they need. Luckily, their healthcare provider can point you towards resources and groups to help carers like you.

Get organised
Use a calendar or set digital reminders for all upcoming appointments and treatment doses. Ask the doctor or nurse at your haemophilia centre about local groups and resources for haemophilia carers. Coordinate with the patient's doctor to help them adapt their treatment to their usual activity levels.

As someone with haemophilia

If you have been recently diagnosed with haemophilia, there is a lot to learn and you may feel a little overwhelmed. This is not unusual as it takes time to get to know your condition and your limits. As you learn what to expect and how to manage problems as they arise, you should find it easier to refocus on the things you enjoy.

Find out as much as you can about haemophilia – your haemophilia centre and the Haemophilia Society will have plenty of information. The more you know, the easier everyday life will be.

How may haemophilia affect me, my child or someone that I am a carer for?

People with mild or moderate haemophilia do not usually have bleeding problems day-to-day. Bleeding only tends to happen after an injury, operation or dental treatment. That said, be aware that bleeds can happen from less severe injuries and that nose bleeds may be more frequent and last for longer.[4]

Remember, if in doubt, contact your haemophilia centre.

The term 'bleed' has a specific meaning in haemophilia.

This can be different to how people usually talk about a bleed. In haemophilia, a 'bleed' often refers to internal bleeding into a joint, muscle (a bruise) or other areas inside the body and not just open bleeding from a cut.

Your haemophilia centre will be able to tell you how to recognise the different types of bleeds and what to do. The section 'How do I recognise and cope with bleeds?' on page 18 may help.

Unlike people with mild or moderate haemophilia, those with severe haemophilia have spontaneous bleeds into joints and muscles even when they have not been injured. Regular treatment is needed to stop these bleeds or to prevent them.[4]

How should I live my life day-to-day?

Even when haemophilia is mild to moderate, there are a few important, practical things you should do to help you, your child or the person you care for in everyday life.

Carry medical alert identification

It is important the right people, such as medical professionals, are made aware you have haemophilia, even if circumstances mean you are not able to tell them.

- **Your haemophilia centre will give you a 'green card' which you should keep with you at all times, just in case you have an accident**
- **Several companies make MedicAlert necklaces or bracelets that you can wear and are widely recognised by healthcare staff. Ask your centre for details**

Care in the home

A safe environment for young children may help reduce your concerns and limit the number of times you tell your child to be careful.

- **Watch out for sharp plants, furniture or belongings**
- **Make sure anyone who looks after your child knows what to watch for and who to contact in case of emergency**
- **As your child gets older, they will get to know their limits and may start to push them**
- **Teenage years may bring other challenges for parents as children face more complex social, psychological and physical situations**

School and work[1]

Haemophilia should not hold your child back and they should be encouraged to take part in most school activities.

- **When your child starts school, make sure the teachers and staff are informed, understand the condition and know who to contact if it becomes necessary**
- **School staff should feel confident your child should not be treated differently to other children or excluded from activities unnecessarily.**

  The haemophilia nurse at your centre will be able to provide information to give to the school and, if necessary, will talk to staff involved with your child on your behalf. Takeda have also developed a teacher's guide to haemophilia for the school environment. Ask your child's haemophilia treatment centre if this is available
- **If you or your child have mild or moderate haemophilia, it may help to inform your employer in case you need to take time off work**

Sports and activities[1,4]

Physical activities are beneficial to everyone and it is important for people with haemophilia to stay active to get the health benefits of regular exercise.

- **Those with mild or moderate haemophilia can take part in most activities, however, because there is a risk of bleeding following injury, it is best to avoid hard contact sports such as rugby and boxing**
- **Playing team sports helps build confidence, and swimming is excellent for low impact exercise**
- **Whatever sport your child chooses, the occasional injury will be difficult to avoid but can be managed**

Encourage them to tell you when they get hurt and how it happened. This will help you and the doctor to decide what treatment or adjustments they may need.

Care when away

With the right planning, haemophilia shouldn't limit your sense of adventure.

- **Check treatment availability at your destination**
- **Research what to do in an emergency and check you have appropriate medical cover on your travel insurance**
- **Always carry your medical alert card with you**

What general health issues should I be aware of?

Although people with mild or moderate haemophilia do not generally have bleeds in day-to-day life, vaccinations, dental procedures and operations need some care and planning.

Dental[1,4]

Regular dental check-ups are important so that potential problems can be spotted and dealt with early, such as bleeding gums. This is especially true for people with haemophilia.

Make sure your family dentist knows that you, your child or the person you care for has haemophilia, as there can be a risk of swelling and ongoing bleeding following dental procedures.

Bleeding may also occur when children lose their baby teeth. If so, check that the bleeding stops. If it continues for a long time, or stops and starts, ask your haemophilia centre for advice.

Contact your haemophilia centre if:

- You, your child or the person you care for is planning any type of dental work. They can advise you if treatment is required before or after a procedure and may be able to recommend a dentist

Bumps, bruises and minor accidents[1]

While minor accidents can happen at any age, as soon as a child starts to crawl and climb, bruises are unavoidable. Bruises can look worrying and while you should keep an eye on them, generally they should fade and disappear over time and you should not worry. Even for a child with haemophilia these are not usually painful.

Contact your haemophilia centre straight away:

- If a bruise grows larger over a few hours
- For a bruise on the head, neck, throat, joints or groin
- If there is blood in vomit, spit or saliva, wee or poo
- In case of any head injury, whether there is any outward sign of damage or not

COVID-19 vaccination[8]

There are no special precautions recommended for people with haemophilia other than those required for other vaccinations given into a muscle. Speak to your haemophilia centre if you have any concerns.

Vaccinations[1]

You or your child or the person you care for should be able to have all routine vaccinations, plus any others that your haemophilia centre may recommend.

This also applies to travel vaccinations at all ages. Ensure the person carrying out the vaccination is aware of your (or the patient's) haemophilia and that they may need to administer the vaccine in a different way. If needed, provide details of the haemophilia centre so they can call for advice.

Surgery[1,4]

Make sure the team looking after you, your child or the person you care for knows about your haemophilia and that they must contact your haemophilia centre for advice well in advance of the operation.

Contact your haemophilia centre:

- Well in advance of any surgical operation

IMPORTANT

A haemophilia doctor should check any suspected head injuries. If the injury occurs out-of-hours, follow the usual emergency procedure.

How do I recognise and cope with bleeds?

People with mild or moderate haemophilia are less likely to have bleeds due to minor accidents, but it is still important to learn how to recognise them and what to do. Here are some tips to help.[1]

Bleeds in muscles and joints

- The area feels tight, warm or swollen
- The limb is painful, stiff or difficult to extend

Seek immediate medical advice if there are reports of numbness or tingling

Bumps to the head

- There may be obvious swelling, lumps or bruising
- There may be no mark at all – but always seek help from the haemophilia centre

Bleeds in the mouth, stomach and intestines or urinary tract

- Visible bleeding from mouth or tongue
- Red or brown wee
- Bloody or black tar-like poo

Nose bleeds

- These can go on for longer than normal and can be more frequent

What to do?

Basic first aid procedures apply in haemophilia. The "**PRICE**" acronym is a good way to remember the steps:[1]

- **P** rotect the area
- **R** est the area
- **I** ce, wrapped in tea towel
- **C** ompression, add support and
- **E** levate

Don't delay, joint injuries or bleeds should be acted on quickly to avoid long-term joint damage.

Contact your haemophilia centre if you think an injury is complicated due to haemophilia

Never use any medicine that contains aspirin (acetylsalicylic acid) or other Non-Steroidal Anti-Inflammatory Drugs (NSAIDs) such as ibuprofen, unless advised to do so by your haemophilia centre. These medicines, as well as some herbal remedies, can interfere with blood clotting. If pain relief is needed, paracetamol may be used at the age-appropriate dose.[1]

How do I recognise and cope with bleeds in my child?

If your child has moderate haemophilia, it is possible that they may have bleeds without an apparent cause (as people with severe haemophilia do) or following a minor injury. If you are a parent of a small child with moderate haemophilia it is a good idea to get into the habit of observing them carefully.[1]

In general

- Is your child miserable or crying for no apparent reason (not because they are hungry, thirsty, need a nappy change or want a cuddle)?
- Do they avoid reaching for things with the nearest hand?
- Are they avoiding particular actions?
- Are they favouring one leg?

At changing or bath time

- Look at your child's skin. Are there any new bruises?
- Do their arms and legs look and move the same (no swelling or bruising)?
- Is there any bruising or swelling in the nappy area?

Contact your haemophilia centre if:

- You have any problems that you think may be related to haemophilia
- It is always better to seek advice than to let a potential problem go unnoticed

Understanding what treatment is suitable for you or your child

Your doctor or nurse at the haemophilia centre will tell you what type of haemophilia you have, or your child or the person you care for has, and what their clotting factor level is. They will then work out what treatment may be needed and how it should be used.

As people with mild or moderate haemophilia often have enough clotting factor to cope with minor injuries, treatment may only be needed following a serious accident or before and after an operation. However, individual lifestyles, activity levels, bleeding patterns and risks will all have a bearing on the treatment options your doctor will discuss with you.

As needed treatment: On-demand

When haemophilia is mild or moderate, clotting factor treatment may only be needed as and when a bleeding incident occurs.[9] This 'as needed' infusion regime to treat specific bleed instances is called 'on-demand' treatment.

People with severe haemophilia may require both on-demand and preventative ("prophylactic") treatment, e.g. following an injury.[9]

Added clotting factor when your body needs it:

Injury › Bleed › On-demand treatment › Clot (prevents further bleeding)

Remember to keep a record of your bleeds so you and your healthcare professional can develop an ongoing treatment plan that fits your individual needs.[4] The completion of Haemtrack (which can be accessed via the website haemtrack.mdsas.com or app) is also an important way in which to record and monitor your or your child's bleeds.[10] This easy-to-use system helps you to report all bleed and treatment information to your haemophilia treatment centre as quickly as possible, in order for them to provide you or your child with the best possible care.[10]

Preventative treatment: Prophylaxis

Because repeated bleeds can also lead to long-term damage, particularly to the joints, some people with moderate haemophilia who experience regular bleeds may need regular infusions of factor, similar to patients with severe haemophilia.[5]

In general, this happens between two and three times a week, individualised to the child and often at specific times, such as mornings before school, to prevent bleeds occurring in the first place.[9]

These types of treatment, called prophylaxis, are usually effective in preventing bleeding, including that which occurs for no obvious reason (these are called spontaneous bleeds).[1]

Maintain higher levels of clotting factor:

Preventative treatment › Bleed › Clot › Preventative ("prophylactic") treatment

What are the treatment options?

There are several alternative treatments for bleeding. Here are the most common types:[1,4]

Desmopressin (DDAVP)

- On-demand treatment
- Synthetic hormone that stimulates the body to release stored Factor VIII
- Can be given as an infusion into a vein, injection under the skin of the tummy, or nasal spray
- As desmopressin releases stored Factor VIII, it does not work for haemophilia B

Antifibrinolytics (tranexamic acid)*

- On-demand treatment
- Stops blood clots from breaking down after they have been formed
- Can be taken as tablets, mouthwash or injection
- Often used to treat short-term bleeding, e.g. during dental treatment, nosebleeds or heavy periods

These treatment alternatives may or may not apply to you. Your haemophilia centre will tell you what's suitable. You can also ask about other advanced treatments, such as gene therapy that may become available in the future.

Factor concentrates

- Preventative or on-demand treatment
- Replacement for missing clotting factor
- Are given as an injection directly into a vein
- There are different types available and your doctor will advise which is suitable for you:
  - Standard half-life (SHL): Stays in the blood for a relatively short amount of time, so they need to be injected every 2 to 3 days
  - Extended half-life (EHL): Stays in the blood for longer than an SHL and may reduce the number of injections needed

Some people with haemophilia will develop a type of antibody (inhibitor) which makes it more difficult to stop a bleeding episode because they prevent the treatment from working. Your doctor will monitor you for the development of inhibitors and if this does occur, then other treatment options will be considered.

*Antifibrinolytics are not indicated in haemophilia A or B, except for tranexamic acid in the management of dental extractions.[4]

Special advice for girls and women

Female haemophilia carriers can also have low levels of clotting factor and may face many of the same bleeding issues as boys or men with mild or moderate haemophilia.[1] For girls or women, haemophilia can also bring other challenges.

Menstruation

If menstruating there may be times when you need to manage heavy periods or choose a female contraceptive. You can get advice from your haemophilia centre, where a doctor will be able to talk you through your contraceptive options or may recommend treatment to help control heavy periods.

Pregnancy[1,4]

If you are planning to get pregnant, it is important to let your haemophilia centre know as early as possible so they can arrange the appropriate specialist care.

Problems with bleeding are rare in pregnancy, however, you may need treatment to control bleeding before, during and after giving birth.

Prenatal genetic diagnosis is recommended to help with planning and risk assessment to protect you and the health of your newborn baby. You can request a test during pregnancy to determine the sex of your child. If appropriate, further genetic tests can be carried out to determine the likelihood of your child having haemophilia.[4] You should discuss your options with your haemophilia and obstetric teams.

Final thoughts

You are not alone

Being diagnosed with haemophilia can seem overwhelming at first. There is a lot to learn, but there is also a lot of support. You will soon adapt your lifestyle, and managing your haemophilia, or that of your child or the person you are a carer for, will become more routine.

Get to know your haemophilia centre. While you should not need to contact them too often, it is useful to have a familiar person to talk to if you are ever concerned. They are always there to help if you need them and can provide you with a wide range of information, contacts and reassurance to help you live your life to the full.

Useful resources

The Haemophilia Society
Get support, news and information about haemophilia
url: haemophilia.org.uk*

We bleed differently
Practical support tools for people with bleeding disorders and their care teams
url: www.webleeddifferently.co.uk

I am number 17
Get support and hear from people who have rare diseases
url: iamnumber17.geneticalliance.org.uk

*Takeda UK makes no representation as to the accuracy of the information contained on sites we do not own or control. Takeda UK does not recommend and does not endorse the content on any third party websites such as haemophilia.org.uk. Your use of third party websites is at your own risk and subject to the terms and conditions of use for such sites.

Glossary

Carrier

A woman who has the gene that causes haemophilia on one of her X chromosomes. Women rarely suffer with the condition, as their other X chromosome is normal. So they still produce either Factor VIII or Factor IX, just at lower amounts than those who don't carry the gene

Chromosome

DNA-containing structures that contain all or most of our genes

Clot

A coagulated mass produced by clotting of blood

Clotting factors or factor

Proteins in the blood that act in sequence to stop bleeding and form a clot. Medicines used to prevent or treat bleeds in persons with haemophilia may also be referred to as replacement clotting factors, or factor

Gene

A specific DNA sequence that defines the structure of the body's proteins and related functions

Haemophilia A or B

A hereditary bleeding disorder in which a clotting factor protein, such as factor VIII (haemophilia A) or IX (haemophilia B), is completely or partially lacking

- Mild haemophilia – a factor VIII or IX level from 6% to 40% of normal levels
- Moderate haemophilia – a factor VIII or IX level from 1% to 5% of normal levels
- Severe haemophilia – a factor VIII or IX level below 1% of normal levels

Haemophilia comprehensive care centre (CCC) or treatment centre (HTC)

A treatment facility that provides a range of services for patients with bleeding disorders and their families including information about haemophilia and assistance in learning to live with the condition

Haemophilia multidisciplinary team

The medical team and staff that care for and educate people who live with haemophilia, parents or carers

The Haemophilia Society

A UK-wide charity for people affected by a genetic bleeding disorder

Half-life

The time taken for half the amount of a drug to be eliminated or disintegrated by natural processes

Prophylaxis

A treatment regimen to prevent symptoms of a disease developing. Prophylaxis in haemophilia involves regular injections of clotting factor or monoclonal antibody to prevent bleeding

Protein

Complex molecules coded for in DNA that include many essential biological compounds (such as clotting factors)

References

1. Haemophilia.org Understanding Haemophilia. Available at: https://haemophilia.org.uk/resources/publications/booklets/understanding-haemophilia/. Accessed August 2022.
2. UKHCDO Annual Report 2021 & Bleeding Disorder Statistics for 2020/2021. Available at: http://www.ukhcdo.org/wp-content/uploads/2021/12/2021-UKHCDO-Annual-Report-2020-21-Data.pdf. Accessed August 2022.
3. Office for National Statistics. UK population estimate. Available at: https://www.ons.gov.uk/peoplepopulationandcommunity/populationandmigration/populationestimates. Accessed August 2022.
4. Srivastava A, *et al.* WFH Guidelines for the Management of Hemophilia, 3rd ed. Haemophilia. 2020;26(Suppl 6):1–158. DOI: 10.1111/hae.14046.
5. Cleveland Clinic. Hemophilia. Available at: https://my.clevelandclinic.org/health/diseases/14083-hemophilia. Accessed August 2022.
6. World Federation of Hemophilia. Hemophilia in Pictures. 2008. Available at: http://www1.wfh.org/publications/files/pdf-1318.pdf. Accessed August 2022.
7. Gooding R, *et al*. *J Blood Med* 2021;12:209–20.
8. Haemophilia.org Coronavirus And Bleeding Disorders FAQs. Available at: https://haemophilia.org.uk/bleeding-disorders/faqs/coronavirus-and-bleeding-disorders/. Accessed August 2022.
9. Risal A, Carpenter SL. Hematology/Oncology 2017;12(2):15–31.
10. Haemtrack. Leaflet 2015. Available at: https://apps.mdsas.nhs.uk/Haemtrack/Home/DownloadOpenpdf?file=HaemtrackLeaflet2015.pdf. Accessed August 2022.

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C-ANPROM/GB/HG/0063 Date of preparation: August 2022