Rare bleeding disorders

The Haemophilia Society

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Introduction

The best-known and most common bleeding disorders are haemophilia A (factor VIII deficiency), haemophilia B (factor IX deficiency) and von Willebrand disorder. But there are many more rare bleeding disorders involving blood clotting factors and blood cells called platelets.

The outlook is now the best it has ever been for people with bleeding disorders. Medicine has made huge advances. There are still no cures, but with modern treatment children born now with a bleeding disorder can live a normal lifespan and enjoy the opportunities in life that anyone else has.

This booklet does not cover haemophilia or von Willebrand disorder (these are covered in separate booklets by The Haemophilia Society).

What is a clotting disorder?

A clotting disorder, also known as a coagulation disorder, is a medical condition caused by the absence of a certain protein in the blood. Blood is composed of various types of cells, such as red and white blood cells, as well as platelets, which are suspended in a yellowish liquid called plasma. Platelets are responsible for forming blood clots, which occur when they stick together and block an injury site in a blood vessel.

When there is an injury, the body activates a complex chemical reaction that forms a mesh made of fibrin. This reaction always follows a specific pattern, with each clotting protein, or coagulation factor, turned on in order. Once all the factors are activated, a clot stops the bleeding at the injury site.

Several coagulation factors are present in the blood, ready to activate in response to an injury. However, if any of these factors are absent, the complex chemical process will not occur as intended, resulting in potentially severe and life-threatening blood loss.

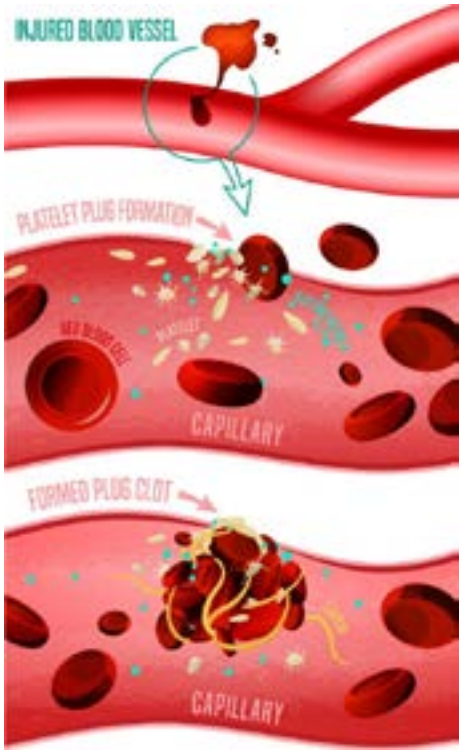
Each coagulation factor is given a number from I to XIII – they are always written as Roman numerals – and the effects of the missing factor will vary.

How does blood clot normally?

Blood is carried throughout the body in a network of blood vessels – arteries, veins and capillaries. When part of the body is injured, damage to blood vessels can cause holes in the vessel walls, where blood will leak out. The vessels can break near the surface, as in the case of a cut. Or they can break inside the body, causing a bruise or internal bleeding.

When a blood vessel is injured, the vessel walls contract to limit the flow of blood to the damaged area. Small blood cells called platelets are activated. They stick tothe site of injury and spread along the surface of the blood vessel to stop bleeding.

The activated platelets release chemical signals that attract more platelets to the area. These clump together to form what is called a platelet plug. On the surface of the platelets, many different clotting factors work together in a series of chemical reactions. This is known as the clotting (or coagulation) cascade – it's like a chain reaction. The result is a fibrin clot, which acts like a mesh to hold the platelets together and to stop the bleeding.



Normally, clotting factors circulate in the blood in an inactive form, because it can be dangerous for a clot to form in the body where it is not needed.

This picture shows the stages in clot formation in a way that makes it easier to understand.

How do you get a bleeding disorder?

Bleeding disorders are generally inherited, meaning they are passed from parent to child in their genes, so you would have the condition from birth. There are some bleeding disorders that you can develop later in life because of another illness or condition, but this is quite rare. These are known as acquired bleeding disorders.

When a baby is conceived, two sets of genes are brought together, one from each parent. Genes are small sections of DNA within the genome that code for proteins. Everything about us, from our eye colour to our height, is coded in our genes.

Different sets of genes carry information for different characteristics.

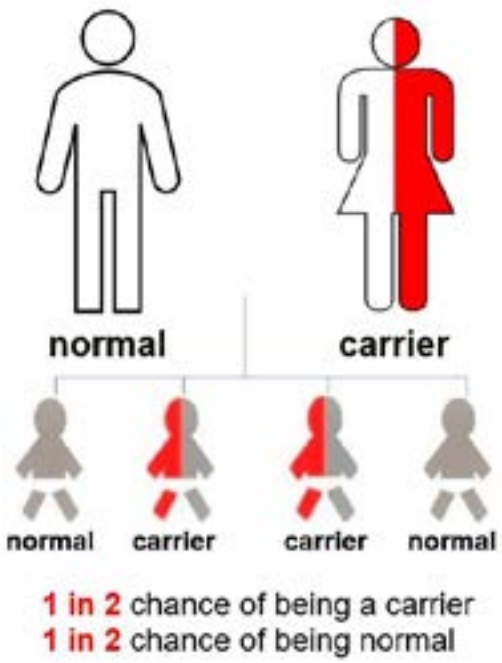
Sometimes genes carry faults that can be passed on to our children. For most bleeding disorders, it is necessary to inherit a specific gene fault from both parents before you develop the condition. People who inherit a gene fault from one parent are called carriers. In most bleeding disorders, they will not have the condition but could pass the gene fault on to their children. Carriers may have lower levels of the clotting factor than normal and may have mild symptoms (but also may have no symptoms at all).

The best-known bleeding disorder, haemophilia, is caused by a gene fault on the X chromosome, one of the two sex chromosomes. This means that it is far more common in boys, because they only have one X chromosome so only need to inherit a gene fault from one parent to have the disease. This is not the case with most bleeding disorders, which are not linked to the sex chromosomes and affect men and women equally.

There are five possible situations that can arise with parents passing on a clotting factor gene or platelet fault:

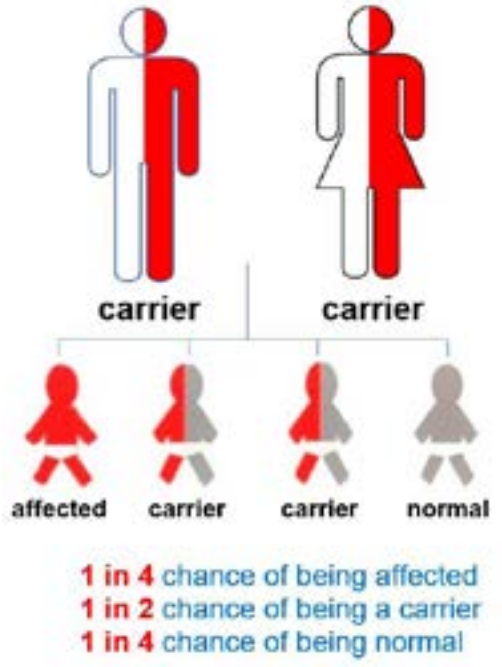
- **one parent is a carrier and the other has 'normal' genes**
- **both parents are carriers**
- **one parent has the condition and the other has 'normal' genes**
- **one parent has the condition and the other is a carrier**
- **both parents have the condition.**

The following show how children will be affected in each situation.



One parent is a carrier and the other has 'normal' genes

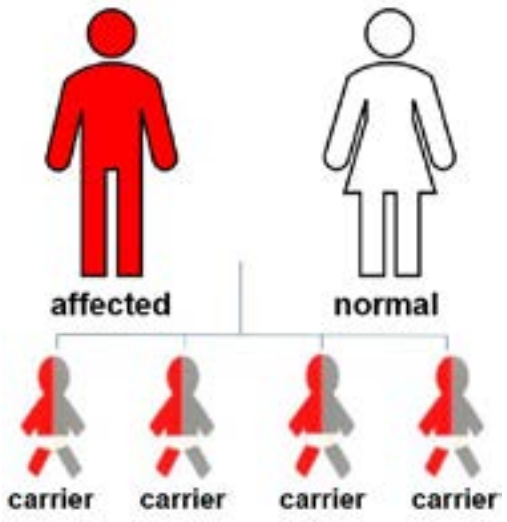
There is a one in two chance that each child will be a carrier and a one in two chance that each child will be a carrier and not have the condition.



Both parents are carriers

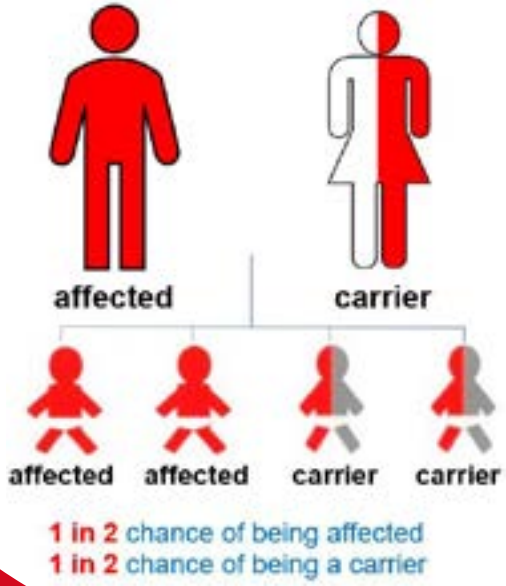
There is a one in four chance of each child being completely unaffected and a one in four chance of each child having the condition.

There is a two out of four chance of each child being a carrier.



One parent has the condition and the other has normal genes

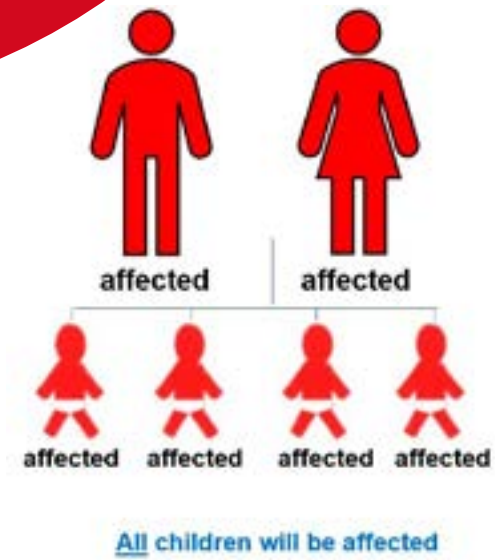
All the children will be carriers, but none will have the condition.



One parent has the condition and the other is a carrier

There is a one in two chance of each child having the condition and a one in two chance of each child being a carrier.

No children will be unaffected.



Both parents have the condition

All children will have the condition.

General symptoms

The symptoms will vary, depending on the exact type of bleeding disorder you have. But there are some symptoms that are common to all of them. You may:

- bruise easily
- have heavy, painful periods lasting longer than a week.

There are other symptoms that don't happen in all bleeding disorders but can in most. You may have bleeding:

- into joints
- into muscles
- for longer than normal after minor or major surgery
- after dental work.

We have included information about preventing bleeding episodes later in this booklet.

Being diagnosed with a bleeding disorder

Depending on the type of bleeding disorder and your family history, you may know you have one from birth, or you may be diagnosed as a young child.

Some bleeding disorders can have mild symptoms, or no symptoms at all. So you may not find out until you are an adult, perhaps after having abnormally heavy bleeding when having a tooth out or after having an operation. It is usual to do blood tests before surgery these days, so it may be picked up during routine pre-surgical screening.

How bleeding disorders are diagnosed

You need to have blood tests to get a firm diagnosis of a bleeding disorder. These are not generally routine tests and may have to be carried out in a specialist centre. You may also have a genetic test to look for the exact mutation in your genes that is causing the condition.

Diagnosis before birth

If you know that a bleeding disorder runs in your family, you may wish to have a test during pregnancy to find out if your baby has the condition. It is not always possible – it depends on the exact type of bleeding disorder.

You may be able to have a test called chorionic villus sampling (CVS) if the gene mutation causing the bleeding disorder in your family is known. 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 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 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, between 15 and 20 weeks.

There is a risk of miscarriage with CVS and amniocentesis – 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.

Treating bleeding disorders

There are many different types of treatment for rare bleeding disorders. Which is appropriate for you will depend on the disorder you have. There is information in our factsheets on the exact treatment that doctors use for each one. This section contains a summary of all treatments that are used. Following that, there is information about how to manage a bleed if you have one.

Factor concentrates

This is the ideal treatment for a bleeding disorder as it replaces the missing or faulty factor. Unfortunately factor concentrates are not available for all bleeding disorders but more research is being carried out to try and manufacture other treatments. You have the treatment into a vein, usually as an injection but sometimes as a drip (an intravenous infusion). Depending on your condition and how severe it is, you may have factor concentrates to try to prevent bleeding (prophylactic treatment) or only after you have had a bleed.

Factors I, VII, VIII, IX, X, XI and XIII are available in replacement factors for bleeding disorders. They can all be made from human blood plasma – the straw-coloured liquid that carries the clotting factors. The plasma is treated during the process to make sure it cannot spread HIV or hepatitis.

For bleeding disorders involving factors VII, VIII, IX and XIII there are man-made factor ('recombinant') concentrates available. These are made in a laboratory. There is another factor replacement treatment called prothrombin complex concentrate (PCC). This is also made from human plasma and contains a mixture of factors II, VII, IX and X, although not all PCC products contain all of these. It is used to treat deficiencies of factors II and X, and inherited combined deficiency of vitamin K dependent factors (VKCFD).

Fresh frozen plasma

You may have treatment with human plasma if there is no factor concentrate available for your condition. You have the treatment through a drip into a vein. It is frequently used as treatment for factor V deficiency and is a straw-coloured liquid.

Platelet transfusions

You may have this if you have a platelet disorder. Platelets are small blood cells, found in human blood, that are important for clotting, they stick to each other

to form the platelet plug, which is important for the clotting factors to stick on to. These are given through a drip into a vein and looks very much like blood plasma.

Cryoprecipitate

This is made from blood plasma. It contains factor I, factor VIII, VWF and FXIII that are important for blood clotting. You have the treatment through a drip into a vein. It is more concentrated than FFP, so you have a smaller volume through the drip.

Desmopressin

Also known as DDAVP, this is a manufactured hormone that boosts factor VIII and VWF levels in people with mild haemophilia, some forms of VWD and will raise FVIII in combined factor V and VIII deficiency (F5F8).

Tranexamic acid (TXA)

This drug helps to stop clots breaking down. It is useful if you need to have a tooth out, before planned surgery or if you have nosebleeds or heavy periods. It can be used alongside factor replacement for other bleeding symptoms. It comes either as a tablet or liquid that you swallow or as an injection given into a vein.

Fibrin glue

You have this to treat an injury, rather than your condition. It is mostly used if you have 'open' bleeding such as a cut or wound. Your doctor or dentist will apply it directly to the bleeding site.

Vitamin K treatment

This is a treatment for some forms of inherited combined deficiency of vitamin K dependent factors (VKCFD). You may have it if you have a bleed or need surgery. It comes as an injection or in a tablet. If it doesn't help you will need factor replacement treatment.

Hormonal contraceptives

This means either the birth control pill or mirena coil. Any woman with a bleeding disorder can take this to help control heavy periods.

Treating a bleed

If you have a bleed, there are steps you can take to treat it. This is not a substitute for medical treatment, such as factor replacement, but can help. To help you remember what to do, think P.R.I.C.E.

Protection
Rest
Ice
Compression
Elevation

Protection – make sure you do not make the injury any worse. Contact your haemophilia team if you think you need a brace, splint or crutches.

Rest – walk as little as possible if you have an injury to a leg. You may need to use crutches or a wheelchair. Rest an affected arm in a sling.

Ice – apply ice to the injured area for about 15 minutes every two hours. Never put ice directly onto the skin – use an ice pack or bag of frozen peas wrapped in a damp towel. Ice lollies are good for mouth bleeds.

Compression – wrap an injured joint in an elastic bandage using a figure of eight pattern. Look out for signs that the bandage is too tight and cutting off the circulation. These are numbness, cold, a sharp pain or a change of colour in fingers or toes. If any of these happen, take the bandage off and rewrap using less tension.

Elevation – keep the affected limb above the level of your heart to limit swelling and improve the circulation.

Your haemophilia team will support you during any bleeding episodes. A major bleed into a muscle or joint can be very painful and cause permanent damage if not treated. So do contact your team if you are concerned that you have had a joint or muscle bleed.

Anaemia

You can become anaemic if you have frequent bleeds, such as nosebleeds or prolonged periods. It means you have a low haemoglobin level because of the regular blood loss. Common symptoms of anaemia are due to the reduced amount of oxygen in the body. These include tiredness, having little energy (lethargy), feeling faint and breathless. The treatment of anaemia depends on what is causing it. You may be asked to take regular iron supplements by your haemophilia centre team.

Preventing and recognising bleeds

Preventing a bleed

You can't always prevent bleeding but there are some things you can do to make having one less likely.

- Only take medicines that have been approved for you by your doctor and haemophilia team. Paracetamol (or Calpol for children) is the only over-the counter painkiller you should take.
- Never take aspirin unless your haemophilia team has agreed that you can take it for a heart condition, for example. This is particularly important if you have a platelet disorder because aspirin blocks platelet function.
- Don't take ibuprofen (e.g. Nurofen) or similar over-the-counter anti-inflammatory drugs. These can irritate the stomach lining and cause bleeding; they also affect platelet function. You may be advised to take this by your haemophilia team if you are on regular treatment.
- Always speak to your haemophilia team before taking any herbal medicines, vitamin supplements or alternative remedies.
- If you need to have surgery or dental treatment, contact your haemophilia team beforehand in case you need any treatment to prevent or minimise bleeding.

Being prepared

- If you are planning any trips, let your haemophilia team know. They will tell you if you need to take any precautions. There is a list of haemophilia centres worldwide on the World Federation of Hemophilia website: **www.wfh.org**

- Always stay in touch with your haemophilia team.

Recognising the signs of a bleed

Whether you have a severe or mild bleeding disorder, it is important that you know the signs of a bleed so you can spot it quickly, get appropriate treatment and minimise any complications. People with mild bleeding disorders may be more at risk of missing a bleed as they won't be so used to the signs. Some bleeds are more obvious than others, of course. Nosebleeds, bleeding from the gums, heavy periods or bright red blood in the urine are all easy to spot. But other types of bleeding may be more difficult.

Nose bleeds

Nose bleeds are very common in all children, they tend to get less as children get older. As a rule of thumb, normal first aid precautions should stop nose bleeds in people with bleeding disorders. If after 15-20 minutes this isn't the case you should contact your haemophilia centres.

Joint bleeds

Bleeds are most common in the elbows, knees and ankles, but they can happen anywhere so look out for signs if you've had a blow or sprain. The signs are pain, swelling, and stiffness. The joint may feel warm or hot. An early sign can be tingling or a bubbling feeling inside the joint. A bleed into a joint can cause permanent damage if not treated so do contact your haemophilia team.

Muscle bleeds

This may happen if you have had a blow or a sprain. The muscle may feel tight, hot or stiff. You may have trouble moving an arm or leg. More serious signs are pins and needles, tingling, a change of colour in the skin over the muscle or swollen veins. Contact your haemophilia team straightaway if you have any of these.

Gastrointestinal bleeding

This is not common but can be serious if it happens. You may have abdominal or stomach pain, feel faint, clammy or look pale. Signs of a gastrointestinal bleed include black, tarry bowel movements or passing fresh blood. Signs of a stomach bleed include vomiting blood or vomit that looks like coffee grounds. Contact your haemophilia team immediately if you see this.

Blood in the urine
Bright blood in the urine is easy to see. But a more minor bleed may cause the urine to look pinkish or dark brown. A kidney bleed may cause low back pain; contact your haemophilia team for advice.

Bleeds into the eye
This may be a result of a blow or injury near the eye. The area may swell, be painful, or change colour. You may have double vision, blurred vision or see spots. An ice pack may slow the bleeding, but you should still contact your haemophilia team.

Bleeding inside the skull
This can happen after a blow to the head or for no obvious reason. You may have a bad or worsening headache, feel or are sick, become confused, drowsy or sluggish, have slurred speech, stiffness in your neck or muscle weakness. You may also be unable to bear bright light or have double or blurred vision. This is a medical emergency – contact your haemophilia team immediately if you have a head injury.

Bleeding between periods
Women may have a small amount of internal bleeding when they release an egg (ovulate) between periods. A niggling pain on one side, low down in the abdomen is normal. But if pain becomes severe or you feel faint or dizzy, contact your haemophilia team. You may have a bleed into a cyst in your ovary.

Heavy periods
Girls often have heavy periods when they reach puberty. However, women with bleeding disorders can have periods that are heavier or last longer than 'normal'. Anything more than 7 days is NOT usual. You should keep a diary and discuss with your haemophilia team especially if this is affecting your lifestyle or you are very tired.

If you are experiencing any bleeding, it's important to contact your haemophilia team with any concerns you may have.

Bruising and bleeding disorders

Bruises are discoloured patches on the skin that may have a raised middle. They occur when there is bleeding beneath the skin's surface and this results in pain, swelling and redness. The medical term for a bruise is haematoma. Bruises are a common occurrence for people with bleeding disorders. While a bruise may seem swollen and take time to heal, it is usually not a cause for concern unless it is large or on the head.

Signs of bruises:
A bruise is a dark area on the skin that may be raised in the centre. Typically, bruises are less painful than muscle bleeds, but if a bruise does cause discomfort, it may be a throbbing pain instead of a constant one.

How to treat bruises:
If you have a mild bruise, you typically do not need treatment.

If you experience a hard bump or hit to your skin, apply firm pressure to the affected area to reduce bruising. Applying an ice pack to the bruise as soon as possible can also help slow down bleeding from small blood vessels under the skin.

If you notice a lump in the bruise that keeps getting bigger or a lump appearing a day after the injury, you may need to speak to your haemophilia team for treatment.

After 24 hours, you can apply a warm cloth to the bruise as it may help it heal faster.

Important points to remember about using ice include:
- Be sure your child can tolerate the ice. Some young children may feel that the ice is more uncomfortable than the bleed.
- Do not leave the ice on longer than the recommended time. Leaving ice on too long (more than 15 minutes) can cause muscle weakness and can also cause an increase in blood flow.

Remember these things, too:
- As a bruise heals, it changes colour, indicating its age. A new bruise appears blue or burgundy but may turn green and yellow as it fades over time. Typically, it takes around two weeks for a bruise to completely disappear.
- Babies with bleeding disorders can easily bruise from simple handling, but this

should not discourage you from holding your baby. It is important to hold your baby regularly. To avoid causing bruises, scoop your baby up by placing your hands and arms underneath them instead of grabbing them around the chest.

- Toddlers often get bruises on their arms, legs, and faces as they explore and learn to walk and climb. It can be concerning when a child looks "harmed," but it is better to let them play and explore than to keep them sheltered out of worry about others' opinions.

Call your haemophilia centre if:

- You start bruising on your head or neck.
- You feel swelling, numbness, or a tingling feeling along with the bruise. This could mean that leaking blood is putting pressure on the nerves and blood vessels. This pressure can cause permanent damage.
- The bruise hurts.
- You have a hard time moving.
- The lump in the bruise gets larger or does not go away.
- The bruise was caused by a blow to your head or stomach. You could have damage inside your body.

Types of rare bleeding disorders

Bernard-Soulier syndrome

Bernard-Soulier syndrome is a very rare platelet function disorder. It is a bleeding disorder caused by an abnormality in your genes (part of the DNA you inherit from your parents). These genes affect a group of linked proteins (called a receptor) and are normally found on the surface of platelets, a type of blood cell. Because this receptor is missing or is not working correctly, platelets do not stick to the injured blood vessel wall as normal and a normal blood clot doesn't form properly.

What causes Bernard-Soulier syndrome?

This is an inherited genetic disorder that affects both males and females. It is known as recessive, meaning you inherit the gene defect from both parents (even though they do not have the disease). It affects men and women equally and can be found in all ethnic groups.

If you carry one copy of the gene fault, you are known as a carrier. You can only pass the condition on to your children if your partner also carries the gene fault.

You will not have the condition yourself, but if you have children that inherit the gene fault from you, they will also be carriers of the condition.

Symptoms of Bernard-Soulier syndrome

The commonest bleeding symptoms of Bernard–Soulier syndrome are:

- bleeding after surgery, dental care or circumcision
- bleeding from the umbilical cord stump at birth
- bleeding in the gut (gastrointestinal system)
- blood in your urine
- easy bruising
- heavier than usual bleeding after childbirth
- heavy periods that last longer than normal
- nosebleeds or bleeding into the mouth

If you think you or your child might be experiencing a bleed, contact your haemophilia centre for assistance. If left untreated, certain bleeds can lead to ongoing issues like muscle or joint bleeding.

Diagnosing Bernard-Soulier syndrome

Bernard-Soulier syndrome is diagnosed with specialised blood tests, which should be done at a haemophilia centre. A careful medical history will also be taken. They will look closely at:

- Whether they find the missing receptor needed to help your blood clot in the blood sample your supply (using a test called flow cytometry).
- Whether your platelets clump together normally when something called ristocetin is added to your blood (a substance that helps platelets to clump together).
- How long it takes for a platelet plug to form in your sample of blood (known as 'the closure time) and whether this takes longer than normal.

Doctors will also look for:

- platelets that appear larger than normal under a microscope
- fewer platelets than normal.

Treatment for Bernard-Soulier syndrome

Most people with platelet function disorders only need treatment during surgery or dental work and after injury or accidents.

When needed, Bernard-Soulier syndrome may be treated with:

- Tranexamic acid (TXA)
- fibrin sealants
- hormonal treatment (to control excessive menstrual bleeding)
- iron replacement (if necessary to treat anaemia caused by excessive or prolonged bleeding)
- platelet transfusions (only if bleeding is severe)
- recombinant factor VIIa.

If your child has difficult venous access, a central line or vascuport can be considered.

How much your bleeding disorder affects your daily life will depend on how severe it is. Contact your haemophilia team if you have any concerns.

Combined vitamin K-dependent clotting factors deficiency

This bleeding disorder caused by a lack of a protein that the body needs for several blood clotting factors to work properly. It is known as inherited vitamin K deficiency or VKDCFD.

What is inherited vitamin K deficiency?

Inherited combined deficiency of the vitamin K-dependent clotting factors (VKDCFD) is very rare. Doctors have diagnosed it in fewer than 30 families worldwide. VKDCFD is an autosomal recessive disorder and is found more frequently in areas of the world where marriage between close relatives is common.

VKDCFD is a bleeding disorder caused by the body producing less of one of the enzymes required to activate factors II, VII, IX and X. These are referred to as vitamin K-dependent proteins as that is also necessary to make them fully active. It causes problems because the clotting reaction that would normally control any bleeding is blocked too early, so your body doesn't make the blood clots it needs to stop bleeding.

What causes VKDCFD?

This is an inherited genetic disorder. It is what is known as recessive, meaning you have to inherit the gene defect from both parents. It affects men and women. If you carry one copy of the gene fault for VKDCFD, you are known as a carrier.

You can only pass the condition on to your children if your partner also carries the gene fault. You will not have the condition yourself, but any children that inherit the gene fault from you will also be carriers of the condition.

Symptoms of inherited vitamin K deficiency

Most people with inherited vitamin K deficiency are diagnosed early in life, at birth or in infancy or early childhood.
They may have:

- bleeding from the umbilical cord stump at birth
- bleeding in the gut (gastrointestinal tract)
- bleeding into muscles and joints
- bleeding into soft tissues, such as gums
- bleeding into the brain at birth
- easy bruising.

Girls and women usually have heavy periods that last for longer than normal (heavy menstual bleeding). Less often, people are diagnosed in early adulthood because of bleeding or routine blood tests during investigations for another condition. They may have:

- bleeding after surgery
- bleeding after an accident.

VKDCFD can be made worse at times because of other conditions. For example, after taking antibiotics that kill off normal bacteria in the gut which help the body to make vitamin K2. This will get better once the antibiotic course is finished and the normal bacteria recover.

Diagnosing VKDCFD

Inherited vitamin K deficiency is diagnosed with a series of blood tests. These are specialised tests and have to be done at a haemophilia treatment centre.

Most people are diagnosed fairly young in life, either at birth after bleeding from the umbilical cord stump, or because of a bleed in childhood. Or your baby may be tested at birth because VKDCFD runs in your family.

Treatment for VKDCFD

VKDCFD is relatively easy to treat in most cases. Babies and children are started on a daily dose of vitamin K and that usually controls bleeding. If it doesn't work as well as it should, your doctor may suggest a weekly injection of vitamin K instead.

While vitamin K tablets or injections control bleeding well from day to day, you need extra treatment to prevent bleeding if you are at higher risk for any reason. If you have minor bleeding, or are going to have minor surgery, your doctor may suggest that you take tablets called tranexamic acid (TXA) for a few days. These help to stop blood clots breaking down.

If you have a serious bleed or are going to have major surgery, your doctor may also want you to have treatment called prothrombin complex concentrate (PCC). This contains clotting factors II, VII, IX and X. If this isn't available, you may have fresh frozen plasma (FFP), which also contains these clotting factors. Both these treatments are made from donated human blood – from plasma, the straw-coloured fluid that the blood cells are carried in.

You have them through a drip into a vein (intravenously).

You may also need treatment with PCC if you are having a baby. You start the treatment once you've gone into labour and continue with it for three days after you've had your baby.

In general, VKDCFD will not affect your daily life too much, as long as you take your vitamin K as often as you should. You will only need extra treatment to prevent bleeding if you are due to have surgery or are having a baby. But you still have a higher risk of bleeding than normal and need to know the signs of a bleed so you can recognise when you need to seek medical help.

Factor II (prothrombin) deficiency

This bleeding disorder is related to problems with a blood-clotting factor called prothrombin or factor II (pronounced factor 2). Factor II is also called prothrombin, so you may hear this condition called prothrombin deficiency. It's a bleeding disorder that happens because of either:

- your body produces less prothrombin than it should, or
- the prothrombin your body makes does not work correctly.

This causes problems because the clotting reaction (how the body usually reacts to stop any bleeding) is blocked too early. So your body doesn't make the blood clots it needs to stop bleeding.

Factor II deficiency is very rare, and doctors estimate that it only affects about one in every two million people.

What causes factor II deficiency?

Factor II deficiency is an inherited genetic disorder. It is known as recessive, meaning you must inherit the gene defect from both parents. It affects men and women equally and can be found in all ethnic groups. It is frequently found in areas where marriage between close relatives is common.

You are known as a carrier if you carry one copy of the gene fault for factor II deficiency. You can only pass the condition on to your children if your partner also carries the gene fault. Children who inherit the gene fault from you will also be carriers of the condition. Carriers will not have the condition but could have slightly lower than normal prothrombin levels, which can cause more minor symptoms.

There are two main types of prothrombin deficiency:

- **Hypoprothrombinaemia** – where reduced amounts of prothrombin are produced by the body, which can cause mild, moderate or severe bleeding, depending on the amount of prothrombin present.
- **Dysprothrombinaemia** – this is where the amount of prothrombin produced is normal but it does not work properly.

The severity of symptoms ranges from mild to severe depending on the amount of prothrombin present in the blood and its activity.

Getting factor II deficiency later in life

It is also possible to develop a factor II deficiency later in life. This is called acquired factor II deficiency. It may be temporary and can be caused by

- a lack of vitamin K
- some antibiotics or diseases can affect how well things are absorbed in the gut, or
- by liver disease

Symptoms of factor II deficiency

Factor II deficiency can cause varying symptoms depending on the amount of

prothrombin produced by the body. The lower prothrombin levels in the blood, the more frequent and severe the symptoms may be.

Common symptoms include:

- bleeding when the umbilical cord is separated
- bleeding after circumcision
- bleeding during or after surgery
- bleeding in the mouth, particularly after dental surgery
- bleeding into joints
- bleeding into muscles
- easy bruising
- heavy periods or periods that last longer than normal (also known as heavy menstual bleeding)
- nosebleeds.

You may also have these symptoms, but they are less likely:

- Heavier than usual bleeding during or after injury or childbirth
- bleeding in the gut (also known as gastrointestinal haemorrhage)
- bleeding into the brain or spinal cord (the central nervous system).

These symptoms are rare:

- bleeding in the central nervous system (the brain and spinal cord)
- blood in the urine (haematuria).

If your child has trouble with their veins, a central line or vascuport (implanted venous access device) may be considered by your haemophilia team.

You must contact your doctor or haemophilia centre if you think you or your child are having a bleed. Some bleeds can cause long-term problems if they aren't properly treated, such as bleeding into muscles or joints. And some can be life-threatening, such as a brain or spinal cord bleed.

If you are a carrier of the gene for factor II deficiency, you may have minor symptoms, such as bleeding for longer than normal after having a tooth out or having your tonsils out.

Diagnosing Factor II deficiency

Factor II deficiency is diagnosed with blood tests. If your baby had bleeding from the umbilical cord or when they were circumcised, your doctor may suggest these blood tests at birth. The blood tests are usually done at a haemophilia centre as the needed tests are very specialised. Once your blood has been taken, it is sent off to the lab for testing straight away. The lab will test your blood for many different things, including how quickly your blood clots and a specific test that measures the amount of prothrombin in your blood. Once they have your results, your haemophilia centre will contact you to discuss their findings.

During pregnancy, prothrombin deficiency can be diagnosed before birth (prenatally) if there is a family history. Several options include chorionic villus sampling (CVS) early in pregnancy or amniocentesis around 15 to 20 weeks or so.

You need to have DNA testing to find out if you or your children are carriers of the gene fault.

Milder forms of prothrombin deficiency (where the levels of prothrombin are reduced but less significantly) may only be discovered when bleeding continues after surgery or injury.

Treatment for factor II deficiency

How often you need treatment will depend on how severe your condition is.

You will probably only need treatment if you have a bleed. But very rarely, people with severe factor II deficiency may need regular treatment to prevent bleeds.

Your doctor may suggest that you take tranexamic acid (TXA) for minor bleeding or before minor surgery. These work by stopping the breakdown of blood clots.

If you have very heavy periods, your doctor may suggest you take hormone treatment to lighten them. Or you may take tranexamic acid tablets during your periods.

There are two treatments available for more serious bleeding in factor II deficiency:

- prothrombin complex concentrate (PCC) contains factor II along with two or three other clotting factors
- fresh frozen plasma (FFP)

Both are made from donated human blood from plasma, the straw-coloured fluid in which the blood cells are carried. You have these through a drip into a vein (intravenously).

You may also have treatment with PCC before major surgery or if you are having a baby when you go into labour and for a few days after your baby is born.

If you have very low prothrombin levels, you may need regular treatment with PCC to prevent bleeds. Your doctor may call this prophylaxis (pronounced proff-ill-axe-iss). It is usually possible to teach you to self-treat or treat your child at home.

The impact of your bleeding disorder on your daily life is determined by its severity. In some cases, it may not have a significant effect. However, it may become an issue when undergoing dental work, major surgery, having a baby, or experiencing an accident. Some individuals might require occasional treatment to manage minor or severe bleeding, while a minority may require regular treatment to prevent any bleeding.

Factor V deficiency

This bleeding disorder is related to problems with a blood clotting factor called factor V (five).

What is factor V deficiency?

It is a bleeding disorder caused by the body producing less of a certain clotting factor than it should. This causes problems because the clotting reaction normally controls any bleeding that is blocked too early. So your body does not make the blood clots it needs to stop bleeding.

Factor V deficiency is very rare, and doctors estimate that it affects about one in a million people worldwide.

What causes factor V deficiency?

This is an inherited genetic disorder. It is known as recessive, meaning you must inherit the gene defect from both parents to have the condition. It affects men and women equally.

You are known as a carrier if you carry one copy of the gene fault for factor V deficiency. You can only pass the condition on to your children if your partner also carries the gene fault. Children who inherit the gene fault from you will also be carriers of the condition. Carriers will not have the condition but could have slightly lower than normal factor V levels, which can cause more minor symptoms.

As Factor V is autosomal recessive, if you inherit the faulty gene from both parents, you will develop the severe form of the disease. Autosomal recessive disorders are more common in areas where marriage between close relatives is common.

In some cases, people can develop acquired factor V deficiency later in life. Although rare, this condition is often caused by anti-clotting drugs during heart or brain surgery. It has also been observed in a small number of individuals with rheumatological conditions, cancer or those who have undergone treatment with certain antibiotics.

Symptoms of factor V deficiency

Factor V deficiency symptoms can vary in severity, even among family members. The amount of factor V your body produces determines the severity of the symptoms.

The lower factor V in your blood, the more frequent and severe your symptoms will be. However, some symptoms may be too mild to cause any issues.

Common symptoms include:

- easy bruising
- nosebleeds
- bleeding gums
- heavy periods that last longer than normal (heavy menstual bleeding)
- bleeding after dental work, surgery or an accident.

You may also have these symptoms, but they are less likely:

- bleeding from the umbilical cord stump at birth
- bleeding into joints or muscles
- bleeding into the bladder or kidneys (genitourinary system)
- bleeding into the brain or spinal cord (central nervous system).

You must contact your doctor or haemophilia centre if you think you or your child are having a bleed. Some bleeds can cause long-term problems if they are not correctly treated, such as bleeding into muscles or joints. And some can be life-threatening, such as a brain or spinal cord bleed.

If you are a carrier of the gene for factor V deficiency, you may have minor symptoms, such as bleeding for longer than normal after having a tooth or your tonsils out.

Diagnosing factor V deficiency

Factor V deficiency is diagnosed with blood tests. These include tests to measure how quickly your blood clots. They are specialised tests, so you need to have them done at a haemophilia centre. The level of factor V in the blood and the level of factor VIII are checked, as these sometimes occur in combination, resulting in different bleeding. The gene mutation will also be identified to help plan pregnancies and test other family members.

Factor V deficiency can also be diagnosed before birth (prenatally) if there is a family history. Several options include chorionic villus sampling (CVS) early in pregnancy or amniocentesis at around 15 to 20 weeks.

Factor V deficiency is usually diagnosed at a young age, because of a bleed, with many people diagnosed in infancy or early childhood. If you have the condition mildly, though, it may not show up until you have bleeding, for example, after dental work or surgery.

Treatment for factor V deficiency

Generally, people with factor V deficiency only receive treatment if they bleed or before planned surgery. In rare cases with a history of severe bleeding or very low factor V levels, your doctor may suggest regular treatment to prevent bleeds. Your doctor may call this prophylaxis (pronounced proff-ill-axe-iss).

For a nosebleed or soft tissue bleed, you will most likely have treatment with tranexamic acid (TXA). These work by stopping the breakdown of blood clots. You may also need to take these if you have heavy periods. Or your doctor may suggest that you take the hormonal treatment or have an intra-uterine device (IUD) inserted to make your periods lighter.

At the moment, a replacement factor V concentrate is still in development and not yet available. Therefore, for a more severe bleed, treatment typically involves fresh frozen plasma (FFP), which has all the necessary clotting factors, including factor V. FFP is created using human blood and contains plasma. This straw-coloured liquid carries blood cells. This treatment is given through a drip into a vein (intravenously).

If your child has trouble with their veins, a central line or vascuport (implanted venous access device) may be considered by your haemophilia team.

How often you need treatment will depend on how severe your condition is. But if you have a severe bleed, you may need daily treatment for a while, as factor V only lasts in the body for a few hours. In rare situations where people need treatment to prevent bleeding, your doctor will likely suggest you have FFP, which you have through a drip. You have this treatment regularly enough to stabilise your blood levels, which may be as often as every two days.

If you have a baby, you may have FFP when you go into labour and for the next few days after your baby is delivered.

How much your bleeding disorder affects your daily life will depend on how severe it is. For many people, it won't have much effect at all. It may only be an issue if you have dental work, major surgery, a baby, or an accident. Others may need treatment occasionally to treat minor or more serious bleeding. A small number may need regular treatment to prevent bleeding.

If you have factor V deficiency you may need to avoid some activities, such as contact sports, with a high risk of head injury. Most day-to-day activities will be fine. Speak to your local haemophilia centre for advice.

Factor V and Factor VIII combined deficiency

This bleeding disorder is related to problems with two blood clotting factors: factor V (pronounced factor 5) and factor VIII (pronounced factor 8).

It is caused by the body producing less of two particular clotting factors than it should. This causes problems because the clotting reaction that normally controls any bleeding is blocked too early, so your body doesn't make the blood clots it needs to stop bleeding.

Factor V and factor VIII combined deficiency is extremely rare, affecting only one to two individuals out of a million. However, it may be more prevalent in specific populations such as Ashkenazi Jewish and Iranian, with a possible occurrence of one in 100,000. The condition may also be underdiagnosed due to its mild symptoms.

What causes factor V and factor VIII combined deficiency?

This is an inherited genetic disorder, and it affects both males and females. The condition is recessive, meaning you must inherit the gene defect from both parents to develop it. Only one gene fault is involved, as the affected gene is involved in the

production of both factor V and factor VIII.

You are known as a carrier if you carry one copy of the gene fault for factor V, factor VIII deficiency. You can only pass the condition on to your children if your partner also carries the gene fault. Any children that inherit the gene fault from you will also be carriers of the condition but will not have it themselves.

Symptoms of factor V and factor VIII combined deficiency
Factor V and factor VIII combined deficiency symptoms are generally quite mild. The commonest symptoms are:

- bleeding after circumcision
- bleeding gums
- easy bruising
- heavy periods or periods that last longer than normal (menorrhagia)
- nosebleeds
- skin bleeding.

You may also have bleeding:

- after dental treatment
- after giving birth
- after surgery
- into the soft tissues.

More serious bleeds, such as into the gut (gastrointestinal system) or brain and spinal cord (central nervous system), have been known to happen but are rare. Serious bleeds are rare with factor V and factor VIII combined deficiency.

Diagnosing factor V and factor VIII combined deficiency
Factor V and factor VIII combined deficiency is diagnosed with blood tests. These include a series of tests to measure how quickly your blood clots. Blood tests measure the levels at between 5% and 30% of normal. These are specialised tests, so you need to have them done at a haemophilia treatment centre.

Your doctor may suggest these blood tests at birth because your baby had bleeding after the umbilical cord was cut or when a circumcision was done. It may also be diagnosed later in life due to other bleeding symptoms, such as those after dental work or surgery.

Treatment for factor V and factor VIII combined deficiency
Generally, people with factor V and factor VIII combined deficiency only have treatment if they bleed or before planned surgery.

Replacement factor V concentrate is not yet available. So treatment for a bleed usually involves fresh frozen plasma (FFP), which contains all the clotting factors, including factor V. It only has low levels of factor VIII, so you have factor VIII concentrate along with the FFP if you need treatment.

FFP is made from plasma, the straw-coloured fluid in which the blood cells are carried, and factor VIII is made in a laboratory. You have all these treatments through a drip into a vein (intravenously).

Factor VII deficiency

This bleeding disorder is related to problems with a blood clotting factor called factor VII (pronounced factor 7).

What is factor VII deficiency?
Factor VII deficiency is a bleeding disorder caused by the body producing less of a particular clotting factor than it should. This causes problems because the clotting reaction that would normally control any bleeding is blocked too early, so your body doesn't make the blood clots it needs to stop bleeding.

Factor VII deficiency is rare but is the commonest of the rare bleeding disorders. The severity of symptoms ranges from mild to severe depending on the amount of factor VII present in the blood and its activity.

What causes factor VII deficiency?
This is an inherited genetic disorder. It is what is known as recessive, meaning you must inherit the gene defect from both parents. It affects men and women equally, but women tend to be more likely to have symptoms.

If you carry one copy of the gene fault for factor VII deficiency, you are known as a carrier. You can only pass the condition on to your children if your partner also carries the gene fault. Any children that inherit the gene fault from you will also be carriers. Carriers will not actually have the condition, though they may have lower than normal factor VII levels. About one in five carriers will have some symptoms.

It is also possible to develop a factor VII deficiency later in life. This is called acquired factor VII deficiency. This is rare and is most often caused by liver disease. Even more rarely, it can be caused by other conditions such as cancer, infection or after a bone marrow transplant.

Symptoms of factor VII deficiency

Symptoms of factor VII deficiency are very variable. Unlike other bleeding conditions, the severity of your symptoms does not seem to be entirely linked to the level of factor VII in your blood. Some people have no symptoms at all, others have minor symptoms, and some have very severe symptoms.

People with minor symptoms may have:

- bleeding after dental work
- bleeding at circumcision
- bleeding gums
- easy bruising
- heavy periods or periods that last longer than usual (heavy menstual bleeding)
- nosebleeds
- signs of blood in the urine.

People with severe symptoms may also have bleeding:

- from the umbilical cord stump at birth
- into joints
- into muscles
- into the gut (gastrointestinal system)
- into the brain or spinal cord (central nervous system)
- after childbirth (post-partum haemorrhage)
- bleeding in the head (newborns).

The age at which symptoms appear varies depending on the amount of Factor VII in the blood and how well it is working. Children with little or no factor VII may start to show symptoms soon after birth.

Some bleeds can cause long-term problems if they are not properly treated, such as bleeds into muscles or joints. Occasionally some can be life threatening, such as a bleed into the brain or spinal cord. A bleed into the brain may show as a headache that keeps getting worse, sickness, confusion and increasing drowsiness.

It is very important that you contact your doctor or haemophilia centre if you think you or your child are having a bleed. If you have frequent bleeds, such as nosebleeds, you can become anaemic. This means you have low haemoglobin levels because of the frequent loss of blood. Anaemia can make you feel tired and breathless.

Diagnosing factor VII deficiency

Factor VII deficiency is diagnosed with blood tests. These include tests to measure how quickly your blood clots and tests to measure your factor VII levels. These are specialised tests, so you need to have them done at a haemophilia treatment centre.

People with factor VII deficiency that causes severe symptoms are generally diagnosed soon after birth, from cord bleeding or bleeding after circumcision. If you have the condition with milder symptoms, it may be diagnosed in young childhood. If you have the condition with no symptoms, it may only be picked up during a family study because someone else also has it, or during routine blood tests, for example before an operation.

Treatment for factor VII deficiency

The main treatment is factor VII concentrate. Generally, you only have this after a bleed. But some children and adults with severe symptoms have treatment three to four times a week to prevent serious bleeds. Your doctor may call this prophylaxis (pronounced proff-ill-axe-iss).

If you have mild symptoms, your doctor may suggest you take tablets before minor surgery or dental work. This is usually a drug called tranexamic acid (TXA), which helps to stop clots breaking down.

If you have a more serious bleed, you will need treatment with factor VII concentrate. This can now be made in the laboratory and is called recombinant factor replacement. As it doesn't have to be purified from donated human blood, it means there is no risk of infection. You have this treatment as an injection into a vein (intravenously). As an alternative to recombinant factor replacement, you might be given plasma derived factor VII which stays in the blood for slightly longer than recombinant factor VII.

You may also have factor VII concentrate to prevent bleeding if you are going to have major surgery. You are most likely to have one dose before your operation and at least two afterwards.

If your child has trouble with their veins, a central line or vascuport (implanted venous access device) may be considered by your haemophilia team.

How much your bleeding disorder affects your daily life will depend on how severe it is. For many people, it will not have much effect at all. It may only be an issue if you are having dental work, major surgery, are having a baby or have an accident. Others may need treatment from time to time to treat minor or more serious bleeding. A small number may need regular treatment to prevent bleeding.

Factor X deficiency

This bleeding disorder is related to problems with a blood clotting factor called factor X (pronounced factor 10).

What is factor X deficiency?

Factor X deficiency is a bleeding disorder caused by the body producing less of the clotting factor than it should. This causes problems because the clotting reaction that would normally control any bleeding is blocked too early. So, your body doesn't make the blood clots it needs to stop bleeding. Factor X needs vitamin K from the liver to be activated. Factor X deficiency is rare. Doctors estimate that it affects about one in a million people.

What causes factor X deficiency?

This is an inherited genetic disorder. It is what is known as recessive, meaning you must inherit the gene defect from both parents. It affects men and women equally. Inherited factor X deficiency cannot be cured. If you carry one copy of the gene fault for factor X deficiency, you are known as a carrier.

You can only pass the condition on to your children if your partner also carries the gene fault. You will not have the condition yourself, but any children that inherit the gene fault from you will also be carriers of the condition. It is also possible to develop a factor X deficiency later in life. This is called acquired factor X deficiency. Acquired factor X deficiency is not inherited and occurs in individuals with no family history of the disorder. This is rare, but may be caused by other diseases, including severe liver disease, amyloidosis, cancer and infections.

Symptoms of factor X deficiency

Symptoms of factor X deficiency can be mild, moderate or severe, depending on the amount of factor X your body is able to make. People with mild symptoms may have:

- nosebleeds
- easy bruising
- bleeding gums
- heavy periods or periods lasting for longer than normal (heavy menstrual bleeding).

People with moderate to severe symptoms may also have bleeding:

- from the umbilical cord stump after birth
- after circumcision
- into joints
- into muscles
- into the gut (gastrointestinal bleeds)
- into the brain or spinal cord (central nervous system).

It is very important that you contact your doctor or haemophilia centre if you think you or your child are having a bleed. If you have frequent bleeds, such as nosebleeds, you can become anaemic. This means you have low haemoglobin levels because of the frequent loss of blood. Anaemia can make you feel tired and breathless.

Diagnosing factor X

If you have mild factor X deficiency, it may only be diagnosed because of family history or after routine blood tests, before surgery for example. People with moderate factor X deficiency may be diagnosed because they've had bleeding after surgery or an accident. For girls, it may be when their periods start and are heavy or longer than normal.

People who carry one copy of the faulty gene are known as carriers. Most carriers are healthy, but sometimes carriers may show mild symptoms of factor X deficiency, which may or may not need treatment. Severe factor X deficiency is usually diagnosed soon after birth, because of severe bruising, bleeding from the umbilical cord stump or after circumcision. Other people are diagnosed in childhood because of easy bruising or bleeds into muscles or joints.

Treatment for factor X

For minor bleeding such as nosebleeds, bleeding gums or heavy periods, your doctor may suggest you take tranexamic acid (TXA). These work by stopping the breakdown of blood clots. If you have very heavy periods, your doctor may suggest that you take hormonal treatment to make them lighter. You will probably only need other treatment if you have a more serious bleed or before planned surgery.

Treatments include:

- factor X concentrate (FX) – which contains only factor X
- prothrombin complex concentrate (PCC), which contains factor X along with two or three other clotting factor
- fresh frozen plasma (FFP). All are made from donated human blood – from plasma, the straw-coloured fluid that the blood cells are carried in.

You have these treatments into a vein (intravenously). If you need treatment regularly, it is usually possible to teach you to give it yourself at home. In the UK, everyone with severe factor X deficiency and unmeasurable levels of the protein will be offered regular treatment to prevent bleeds. Your doctor may call this prophylaxis (pronounced proffill-axe-iss). You have treatment with FX, PCC or FFP concentrate two or three times a week.

If your child has trouble with their veins, a central line or vascuport (implanted venous access device) may be considered by your haemophilia team.

Women with factor X deficiency should plan their pregnancy carefully as there is an increased risk of early miscarriage or extended bleeding after giving birth. Treatment for bleeding during the last three months of pregnancy, during labour and for a few days afterwards also can be given.

How much your bleeding disorder affects your daily life will depend on how severe it is. For many people, it won't have much effect at all. It may only be an issue if you are having dental work, major surgery, are having a baby or have an accident. Others may need treatment from time to time to treat minor or more serious bleeding. A small number may need regular treatment to prevent bleeding.

Any surgery (including dental) will need careful planning in advance so it is important that all health care professionals involved are aware of the condition.

Factor XI deficiency

This bleeding disorder is related to problems with a blood clotting factor called factor XI (pronounced factor 11).

What is factor XI deficiency?

Factor XI deficiency was also sometimes called haemophilia C (pronounced heem-oh-fill-ee-ah) but has a very different bleeding pattern and should not be confused with haemophilia A and B.

It is a bleeding disorder caused by the body producing less of a clotting factor than it should. It causes problems because the clotting reaction normally controls bleeding and is blocked too early. So your body doesn't make the blood clots it needs to stop bleeding. Factor XI is important for producing thrombin protein that converts fibrinogen to fibrin during clotting.

Factor XI deficiency is rare; doctors estimate it affects about one in a million people. But it is much more common in some populations, including the Ashkenazi Jewish community, affecting up to one in 450 people.

What causes factor XI deficiency?

It is an inherited genetic disorder affecting men and women equally. It is generally known as recessive, meaning you must inherit the gene fault from both parents to have the disorder in a severe form.

If you carry one copy of the gene fault for factor XI deficiency, you do not have the disorder yourself but are a carrier. You can only pass on a severe form of the condition to your children if your partner also carries the gene fault. Children who inherit the gene fault from you will also be carriers of the condition. Although carriers will not have a severe form of the condition, they can have slightly lower than normal factor XI levels. It means they may be at risk of bleeding after surgery, following dental extractions or having heavy periods.

It is also possible to develop a factor XI deficiency later in life. This is called acquired factor XI deficiency and is generally caused by liver disease.

Symptoms of Factor XI deficiency

Most people with factor XI deficiency don't have any symptoms. In those who do, the commonest symptoms are bleeding after surgery or an accident. Spontaneous bleeding is uncommon, but some people have nosebleeds or bruises more easily than normal.

A bleed is most likely after dental surgery or an operation on your mouth, throat, bladder, kidneys or prostate.

Carriers of factor XI deficiency are also at risk of bleeding after surgery.

Heavy periods are common in women with factor XI deficiency. They are also more at risk of bleeding after having a baby (a postpartum haemorrhage).

Diagnosing Factor XI deficiency
Factor XI deficiency is diagnosed with blood tests, including tests to measure how quickly your blood clots. These are specialised tests, so you need to have them done at a haemophilia centre.

Your doctor may suggest these blood tests at birth because your baby bled after circumcision or because you have a family history of factor XI deficiency. It is also common for factor XI deficiency to be diagnosed later in life because of heavy periods or other bleeding symptoms – after dental work, surgery or an accident.

Treatment for Factor XI deficiency
Most people with factor XI deficiency never need treatment. You are only likely to need it if you have an accident, prevent bleeding before planned surgery, or have a baby.

For minor operations, you may have tranexamic acid (TXA) to take beforehand and a few days afterwards. These help to stop the breakdown of blood clots. If you have very heavy periods, you may have tranexamic acid tablets during your periods. Or your doctor may suggest that you take the contraceptive pill to make your periods lighter.

For more serious operations, you may have treatment with fresh frozen plasma (FFP), which is often used to treat rare bleeding disorders. It contains all the clotting factors, including factor XI. It is made from human blood – plasma is the straw-coloured fluid in which the blood cells are carried. You have it through a drip into a vein (intravenously).

Factor XI concentrate is available and is another possible treatment, but many doctors prefer to use FFP as factor XI concentrate can cause problems with blood clots.

If you are having a baby, whether you have treatment to prevent bleeding will depend on the usual factor XI level in your blood. Most women will not need any treatment. If you have a low factor XI level, you may have tranexamic acid tablets to take when you go into labour. Or you may have factor XI concentrate.

How much your bleeding disorder affects your daily life will depend on how severe it is. For most people with factor XI deficiency, it won't have much effect at all. It may only be an issue if you have dental work, major surgery, a baby, or an accident.

Factor XIII deficiency

It is a bleeding disorder related to problems with a blood clotting factor called factor XIII (pronounced factor 13).

What is factor XIII deficiency?
Factor XIII deficiency is a bleeding disorder caused by the body producing less of a clotting factor than it should. It causes problems because the clotting reaction normally controls bleeding and is blocked too early. So your body doesn't make the blood clots it needs to stop bleeding.

Factor XIII deficiency is one of the rarest types of clotting disorder. Doctors estimate that it affects about one in every two million people. Factor XIII plays an important role in wound healing, pregnancy and forming new blood vessels. However, more research is needed to understand this better.

What causes factor XIII deficiency?
It is an inherited genetic disorder known as recessive, meaning you must inherit the gene defect from both parents. It affects men and women equally.

You are known as a carrier if you carry one copy of the gene fault for factor XIII deficiency. You can only pass the condition on to your children if your partner also carries the gene fault. You will not have the condition yourself, but any children that inherit the gene fault from you will also be carriers of the condition.

It is also possible to develop factor XIII deficiency later in life, called acquired factor XIII deficiency. It can be caused by liver disease, some types of leukaemia, inflammatory bowel disease and an auto-immune disease called systemic lupus erythematosus.

Symptoms of Factor XIII deficiency
The first clinical sign of inherited factor XIII deficiency is often a few days after birth or when the umbilical cord separates. The severity of symptoms ranges from mild to severe depending on the amount of factor XIII in the blood and its activity. Most people with factor XIII deficiency have some bleeding.

The commonest symptoms are:

- bleeding after surgery
- bleeding from the umbilical cord stump at birth
- bleeding into muscles or joints
- bleeding into the brain or spinal cord (central nervous system).
- easy bruising
- heavy periods that last longer than normal
- poor wound healing.

Less common symptoms are:

- bleeding in the gut (gastrointestinal system)
- bleeding in the urinary system
- nosebleeds or bleeding into the mouth

You must contact your doctor or haemophilia centre if you think you or your child are having a bleed.

Diagnosing Factor XIII deficiency

Factor XIII deficiency is diagnosed with a specialised blood test to measure factor XIII activity. The usual clotting tests come out as normal with this particular disorder, so you need to have the specialised test, which must be done at a haemophilia centre.

Factor XIII deficiency is usually diagnosed early in life, either at birth after bleeding from the umbilical cord stump or due to a bleed during childhood. If the condition runs in your family, your newborn may be tested at birth.

Treatment for factor XIII deficiency

Because bleeding can be common with factor XIII deficiency, many people have regular factor replacement to prevent bleeding from birth or as soon as they are diagnosed. Your doctor may call this prophylaxis (pronounced proff-ill-axe-is). You will need preventative treatment if:

- you have had bleeding episodes
- members of your family with the condition have a history or bleeds
- you have very low factor XIII activity.

It is given regularly as an injection, sometimes into a central venous access device such as an implantable port. Injections can be given at home, meaning less disruption to family life.

Development of 'inhibitors' means the body fights off factor XIII injections can be a problem for people with factor XIII deficiency, though it's extremely rare. It will be checked regularly at review appointments. If inhibitors develop, additional injections will be needed.

If you have mild bleeding, such as after dental surgery or a minor operation, your doctor may ask you to take tranexamic acid (TXA) and your monthly preventative treatment. If you have more serious bleeding or major surgery, you may need extra doses of factor XIII replacement.

It is very important to keep factor XIII levels up during pregnancy, so your doctor may suggest you increase your preventative treatment to fortnightly or three-weekly as soon as you know you are pregnant. You may also need extra doses when you go into labour.

Any surgery (including dental) will need careful planning, so all healthcare professionals involved must be aware of the condition.

How much your bleeding disorder affects your daily life will depend on how severe it is. Many people with factor XIII deficiency need regular treatment to prevent bleeding. You may need extra treatment for minor or more serious bleeds or if you have surgery.

Women with severe factor XIII deficiency should receive regular factor replacement throughout pregnancy to reduce the risk of early miscarriage.

Fibrinogen (Factor 1) deficiency

This bleeding disorder is related to problems with a blood-clotting factor called fibrinogen or factor I (one). Fibrinogen (pronounced fie-brin-oh-jen) abnormalities are bleeding disorders caused by the body producing less fibrinogen than it should or because your body's fibrinogen doesn't work correctly. They cause problems because the clotting reaction normally controls any bleeding that is blocked too early. So your body doesn't make the blood clots it needs to stop bleeding.

Factor I deficiency is an umbrella term for several related disorders known as congenital fibrinogen defects. There are several different types of factor I bleeding disorders, all affecting males and females equally.

Afibrinogenaemia is pronounced ay-fie-brin-oh-jen-ee-mee-ah. It means a complete lack of fibrinogen. It is rare; doctors estimate it affects about one in a million people.

Dysfibrinogenaemia is pronounced diss-fie-brin-oh-jen-ee-mee-ah. It means that your body makes fibrinogen, but it doesn't work as it should. The inherited form is rare. But it can also develop as a result of other diseases and this is more common.

Hypofibrinogenaemia is pronounced high-po-fie-brin-oh-jen-ee-mee-ah. It means having lower than normal levels of fibrinogen. This is also a rare condition.

Hypodysfibrinogenaemia is pronounced high-po-diss-fie-brin-oh-jen-ee-mee-ah. It means that you have low levels of fibrinogen and that the fibrinogen you do have doesn't work as it should. It is very rare.

Because some people with hypofibrinogenaemia and hypodysfibrinogenaemia do not have any symptoms, we don't know exactly how rare these conditions are.

What causes fibrinogen deficiencies?

Afibrinogenaemia is an inherited genetic disorder. It is known as recessive, meaning you must inherit the gene defect from both parents.

Hypofibrinogenaemia, dysfibrinogenaemia, and hypodysfibrinogenaemia are usually inherited genetic conditions. They can be recessive, meaning both parents must carry the gene defect to pass it on to a child. Or they can be dominant, meaning that only one parent must carry the gene fault to pass the condition on to a child.

People can also develop dysfibrinogenaemia later in life. This is called acquired dysfibrinogenaemia. It is much more common than the inherited form and is usually caused by liver disease.

Symptoms of fibrinogen deficiencies

The severity of symptoms ranges from mild to severe depending on the amount of fibrinogen present in the blood and its activity.

Symptoms of afibrinogenaemia and hypofibrinogenaemia are similar. But your symptoms with hypofibrinogenaemia will depend on how much fibrinogen you have in your blood. The less you have, the more symptoms you are likely to have and the more severe they will be.

Common symptoms include:

- abnormal bleeding after circumcision
- abnormal bleeding after injury or surgery
- bleeding after childbirth (post-partum haemorrhage)
- bleeding from the umbilical cord stump at birth
- bleeding in the mouth, particularly after dental surgery
- bleeding into joints
- bleeding into muscles
- easy bruising
- heavy periods or periods that last longer than normal (menorrhagia)
- nosebleeds
- problems during pregnancy, including miscarriage.

You may also have these symptoms, but they are less likely:

- bleeding in the gut (gastrointestinal haemorrhage)
- bleeding into the brain or spinal cord (the central nervous system)
- blood clots (thrombosis)
- problems with slow wound healing.

If you suspect that you or your child are experiencing a bleed, it is crucial to contact your doctor or haemophilia centre immediately. Inadequate treatment of some types of bleeds, such as those affecting muscles or joints, can result in long-term complications. Additionally, bleeds in the brain or spinal cord can be life-threatening.

The symptoms of dysfibrinogenaemia depend on how well your fibrinogen works. If you have symptoms, they will be similar to those listed above. About 1 in 2 people (50%) have no symptoms, and about 1 in 4 people (25%) have problems with bleeding.

And up to 1 in 5 people (20%) have problems with abnormal blood clots forming.

The symptoms of hypodysfibrinogenaemia are similar to those above, but how many you have and how severe they are will depend on how much fibrinogen your body produces and how well it works.

Diagnosing fibrinogen deficiencies
Fibrinogen bleeding disorders are diagnosed with blood tests. These include tests to measure how quickly your blood clots and a specific test that measures the amount of fibrinogen in your blood. These are specialised tests, so you need to have them done at a haemophilia centre. You need DNA testing to get a definite diagnosis of an inherited Factor I deficiency. But this may not be necessary if your family has a clear history of the disorder.

Your doctor may suggest these blood tests at birth because your baby had bleeding from the umbilical cord or when a circumcision was performed. Afibrinogenaemia is usually diagnosed in this way.

Or the bleeding disorder may show up later in life because of other bleeding symptoms – after dental work or other surgery, for example. This is more usual with hypofibrinogenaemia and dysfibrinogenaemia, which can be diagnosed in adulthood.

Treatment for fibrinogen deficiencies
How often you need treatment will depend on how severe your condition is. Many people with hypofibrinogenaemia and dysfibrinogenaemia do not need any treatment at all.

If you have very heavy periods, your doctor may suggest you take the hormonal treatment to lighten them. Or you may have a tablet to take during your periods to stop the breakdown of blood clots, such as tranexamic acid (TXA).

Your doctor may also suggest you take tranexamic acid tablets for minor bleeding, such as nosebleeds or before minor surgery.

There are three main treatments for more serious bleeding in fibrinogen deficiency. All are made from donated human blood from plasma, the straw-coloured fluid in which the blood cells are carried. They are:

- cryoprecipitate
- fresh frozen plasma (FFP)
- fibrinogen concentrate.

Depending on your condition's severity, you may need regular treatment once or twice a week to keep your fibrinogen levels up. Your doctor may call this prophylaxis (pronounced proff-ill-axe-iss). Or you may need treatment if you have a serious bleed before surgery or dental work. If you have very low fibrinogen levels and become pregnant, you may have fibrinogen concentrate twice a week until you deliver your baby.

You may also need treatment to prevent abnormal blood clots from forming, as this can be a complication of these conditions. If you have had problems with clots, your doctor may suggest you take a low dose of an anti-clotting drug such as heparin (enoxaparin), particularly during pregnancy.

The impact of your bleeding disorder on your daily life varies based on its severity. Some people may not experience much effect, except for dental work, major surgery, having a baby, or an accident. However, others may require regular treatment to prevent or manage bleeding episodes.

Glanzmann's Thrombasthenia

Glanzmann thrombasthenia is a rare disorder of blood platelets

What is Glanzmann's Thrombasthenia?
Glanzmann's Thrombasthenia (or GT for short) is a bleeding disorder. It is caused by blood cells called platelets not working correctly. It is because of an abnormality in the glycoproteins IIb/IIIa genes. The glycoprotein IIb/IIIa genes are carried on chromosome 17 of your DNA. Platelets are small blood cells that are very important for blood clotting. When your body needs the blood to clot to stop bleeding, the platelets normally clump together to form the clot. You have enough platelets in GT, but they cannot bind to a protein called fibrinogen, which normally binds them together in the clumping process. Because they cannot stick together, they cannot form a stable clot to stop bleeding.

Both men and women are equally affected by this condition. Infants are typically diagnosed early on, often before their first birthday, due to experiencing prolonged bleeding episodes or skin rash caused by bleeding underneath the skin.

What causes Glanzmann's Thrombasthenia?
GT is an inherited genetic disorder. It is known as recessive, meaning you must inherit a gene fault from both parents.

Very rarely people can develop GT later in life. It is called acquired GT and is caused by the body developing antibodies to the IIb/IIIa receptors that normally bind to fibrinogen.

It is a type of autoimmune disorder that can occur in association with other autoimmune disorders, such as systemic lupus erythematosus (SLE). Or it sometimes happens because the person has another medical condition such as Hodgkin or non-Hodgkin lymphoma or another platelet disorder called immune thrombocytopenic purpura (ITP). It can also occur for no obvious reason.

Symptoms of Glanzmann's Thrombasthenia
The symptoms of GT vary in severity, but most people can experience bleeding symptoms with little or no injury. In all cases, treatment is needed to prevent bleeding around surgery and dental extractions. Common symptoms include:

- bleeding from the nose or gums (sometimes after losing baby teeth, during teething or even after vigorous brushing)
- easy bruising
- excessive bleeding after childbirth
- excessive bleeding after surgery or injury
- prolonged bleeding after dental or other surgery
- very heavy periods.

The most common symptom in children is frequent and heavy nosebleeds, which can lead to iron deficiency anaemia.

Less common symptoms include:

- blood in the urine
- bleeding into the head and brain (headache, neck ache, drowsiness, loss of
- consciousness)
- Bleeding into the gastrointestinal tract (vomiting blood, bloody or black bowel movements).

GT can also cause bleeding into joints and muscles, but this is rare compared to other types of bleeding disorders if there is no direct trauma.

You must contact your doctor or haemophilia centre if you think you or your child are having a bleed.

Diagnosing Glanzmann's Thrombasthenia
Like other bleeding disorders, GT is diagnosed with blood tests. Your platelet count will be measured, which is usually normal in GT. Blood samples in the laboratory will be tested to see if the platelets aggregate (clump together) when stimulated with certain chemicals. These are specialised tests, so you need to have them done at a haemophilia treatment centre. If the aggregation test is abnormal or only a small blood sample can be obtained, then a flow cytometry test can be used to look directly for the IIb/IIIa protein on platelets. Flow cytometry will be the diagnostic test for a baby or child.

Your doctor may suggest these blood tests in a baby or young child because they show bleeding symptoms, such as bruising, rashes, bleeding gums or prolonged bleeding after circumcision. Inherited GT is usually diagnosed very young, before the age of one.

In the rare cases of acquired GT, you would have these tests because your doctor is concerned about unexplained bleeding.

Treatment for Glanzmann's Thrombasthenia
For most of the time, you may not need any treatment at all. Generally, you only need treatment if you have a bleed that cannot be controlled or are about to have surgery. Bruising may also need treatment, especially if extensive, raised haematomas or painful. Contact your Haemophilia Centre if you have a large, raised, painful bruise, especially on the face.

GT may be treated with the following:

- antifibrinolytic drugs (tranexamic acid)
- recombinant factor VIIa
- platelet transfusions (only if bleeding is severe)
- fibrin sealants

- hormonal contraceptives (to control excessive menstrual bleeding)
- some patients with extremely severe bleeding may have a stem cell transplant to replace their bone marrow with one that produces normal platelets.

Antifibrinolytics: drugs that stop clots from breaking down. The most commonly used drug is called tranexamic acid (TXA). Your doctor may suggest you take tranexamic acid tablets three or four times a day if you bleed or before and after procedures likely to cause bleeding, such as minor dental work or surgery.

If you are having a tooth out, your doctor will ask you to take TXA the day before and for up to a week afterwards. Women should also take it during their periods and at least two weeks after having a baby.

If you have a mouth bleed, TXA comes in a mouthwash, and if needed, you can make your mouthwash by dissolving a TA tablet in 10 ml of water. If you have a nosebleed, your doctor may put gel or gauze up your nose that is soaked in TA or another treatment called topical thrombin (another clotting factor).

Recombinant factor VIIa

It is a blood clotting factor called factor VII (seven), sometimes written as rFVIIa. 'Recombinant' means that it can now be made in the laboratory rather than getting it from blood donors. As it doesn't have to be purified from donated human blood, there is no risk of infection. You have this treatment as an injection into a vein (intravenously).

rFVIIa is often chosen for minor bleeds to avoid the development of antibodies against platelets or because patients have already developed antibodies to them. You may have it if tranexamic acid can't control a minor bleed. You may also have it before and after planned surgery or during childbirth.

When you have treatment with rFVIIa for a bleed or after surgery, you are likely to have several infusions about 90 minutes to two hours apart. You may have platelets at the same time. rFVIIa doesn't work for everyone but can be very helpful for many people with GT. It is less likely to work in severe bleeding, mouth and nose bleeding, or if treatment has been delayed. If it doesn't work, your doctor will also give you platelets.

Platelet transfusion

Platelet transfusions are often used to stop bleeding in GT. The platelets are given through a drip into a vein. It can work very well, but some people develop antibodies to platelet transfusions. This means the platelets get removed from the blood quicker and are less effective at stopping bleeding. To try and reduce the risk of this, your doctor may prescribe 'HLA matched platelets' when the transfusion is planned (for example, to cover surgery). These platelets have been matched to your blood proteins, so you are less likely to develop antibodies. It takes around six hours to get hold of the right HLA-matched platelets, so you will have to have non-matched platelets in an emergency.

You may also develop antibodies against the IIb/IIIa protein. It can reduce the effectiveness of platelet transfusions and mean that additional treatments need to be used.

Stem cell transplant

It is an intensive treatment, and you are only likely to have this if you have very severe GT, with frequent heavy bleeds that are difficult to control. Most transplants that have been done so far are in children.

To have a stem cell transplant, you usually have a stem cell donor who is a family member (almost always a full brother or sister) with a blood type similar to yours (an HLA matched).

The patient must have drug treatment that kills off their bone marrow and stops the body from rejecting the donor stem cells. You have this treatment through a drip into a vein over several days. After that, the donor stem cells are given through a drip.

Due to the loss of your bone marrow, you are more susceptible to infections and will need to stay in an isolated room at the hospital. The medical team will closely monitor you for several weeks until your stem cells have regenerated successfully, allowing you to fight off infections once again.

Treatment complications

When you receive a platelet transfusion, your body can develop antibodies to HLA markers that are not identical to yours. HLA-matched platelets can be supplied to reduce the risk of developing antibodies. There is still a risk that you may develop antibodies to the IIb/IIIa protein.

Pregnancy in women affected by GT carries multiple risks. You must contact your haemophilia centre for pregnancy advice.

How much your bleeding disorder affects your daily life will depend on how severe it is. For many people, it will not have much effect at all. It may only be an issue if you have dental work, major surgery, a baby, or an accident. Others may need treatment occasionally to treat minor or more serious bleeding. A small number may need regular treatment to prevent bleeding.

Self-advocacy for your / your child's bleeding disorder

As you navigate your or your child's medical condition, you may become the expert on the diagnosis. You will meet many medical professionals that perhaps have never heard of your diagnosis, let alone understand what it means.

If you feel your or your child's needs are not being met, don't hesitate to speak up and challenge the medical professionals.

Shared decision making (SDM) is a collaborative process between you and your healthcare team (HCT), where you are supported in making decisions that align with your personal circumstances, goals, values, and beliefs.

Your HCT provide their expertise, including information about treatment options, evidence, risks, and benefits, while also considering your unique perspective as the patient. Together, you can decide on the treatment that is best for you.

For this conversation, it's important for both you and your HCT to understand what matters most when deciding on a treatment plan. Your healthcare team will provide information about all available treatment options for your specific health concern, including one that may be more appropriate based on your medical history and test results.

When you talk to your HCT, you share details about your life and experiences with illness and treatment. You may have preferences about which treatment option would work better for you. Sometimes, these preferences differ from what your HCT thinks is the best medical treatment.

To make the best treatment decision, you and your HCT need to have a conversation where you try to understand each other's perspective. You should agree on the best treatment option, which could be based on medical reasons or what works best for your situation.

Living with a bleeding disorder

How much your bleeding disorder affects your daily life will depend on the type you or your child have and how severely you have it. Bleeding disorders are very variable. You can have a mild form of some types and if so, you may not have any symptoms at all. It may only be an issue if you are having major surgery, having a baby or have an accident. But you need to be able to spot the signs of a bleed and know how to deal with it if it happens.

Coping with your diagnosis

Finding out that you or your child have a bleeding disorder can be upsetting and bring on a range of different emotions. Some people are frightened and anxious. Others may be relieved that they finally have a diagnosis for symptoms they've had for some time. Parents may feel guilty that their child has been born with an inherited disorder and feel that they have caused it.

All this is perfectly normal. You have had a bit of a shock. Talking to other people with a similar condition, as well as your haemophilia team, can be a real help.

Finding out as much as you can about your condition can help you to deal with it, so do ask as many questions as you need to. Your haemophilia team will be more than happy to answer your questions no matter how many times you ask.

You may also get support from talking to your friends and family. Passing on information to them about your condition can help them to support you. Get in touch with your treatment centre, or call The Haemophilia Society on **020 7939 0780** or email **info@haemophilia.org.uk**

Dental care

It is very important to look after your teeth and pay attention to mouth hygiene. This will help to lower your risk of gum disease and tooth decay and minimise the need for dental treatment later in life. Dental procedures can cause prolonged or excessive bleeding if you have a bleeding disorder.

- Brush your teeth at least twice a day.
- Floss your teeth daily.
- Use toothpaste that contains fluoride.
- Have regular dental check-ups.

Having a tooth out or root canal work can cause bleeding. Your dentist will need to contact your haemophilia team before you have any dental work done. They can advise on any risks or precautions that need to be taken. They may ask you to take Tranexamic Acid (Cyklokapron) or other treatment beforehand to minimise bleeding.

Carrying medical information with you

In an emergency, it is important that anyone giving you medical treatment knows that you have a bleeding disorder. You should carry with you:

- information about your disorder
- information about any treatment you are on
- the name and phone number of your doctor and haemophilia centre.

If you are registered with the National Haemophilia Database, you will have a 'Bleeding Disorder Information Card' with the name of your disorder and contact details for your haemophilia centre. It is very useful to have this with you in case you need to go to another hospital. If you're going away, find out where the nearest haemophilia centre is and take the address and phone number with you.

If you prefer, you can get a MedicAlert disc or piece of jewellery. You must buy these but as you wear them, they are a good way of making sure your medical details are always to hand. Visit **medicalert.org.uk** to find out more.

Special issues for girls and women

Women have added issues to deal with if they have a bleeding disorder, because they can affect periods, pregnancy and childbirth.

Girls just starting their periods may have heavy bleeding. Women of any age may have heavier periods that last longer than normal. This can make you anaemic – a lower than normal level of red blood cells that can cause tiredness and breathlessness.

If you have an inherited bleeding disorder, you may want to see a genetic counsellor before you become pregnant. They can talk through the risk of having an affected child and tell you whether prenatal testing is available. You will need to see an obstetrician earlier than most women – as soon as you know you are pregnant. The obstetrician will need to be in contact with your haemophilia centre throughout your pregnancy.

If you have factor XIII (13) deficiency, or the factor I (1) disorder afibrinogenaemia, you have a greater risk of miscarriage and a complication of pregnancy called placental abruption. This is where the placenta comes away from the wall of the womb, meaning that the baby gets less blood flow and oxygen. You will need treatment throughout your pregnancy to lower the risk of miscarriage and placental abruption. Contact your haemophilia centre before planning a pregnancy and as soon as you think you might be pregnant.

All bleeding disorders carry a greater risk of bleeding for you during delivery and after the baby has been born. Treatment can lower the risk of bleeding and minimise it if it does happen. Treatment is different for each woman and will depend on your own and your family's history of bleeding symptoms, how severe your bleeding disorder is and how you deliver your baby. Some women may need factor replacement treatment. You need to discuss your pregnancy and delivery with your haemophilia team even if you are delivering in another hospital.

Frequently asked questions

Can I have genetic counselling?
It is possible to have genetic counselling before planning a family, both for affected individuals and unaffected carriers. You can discuss this with your haemophilia centre.

Can I have over the counter medication
You should not take aspirin or other non-steroidal anti-inflammatory drugs, as they can worsen bleeding symptoms.

Other medicines that interfere with the way platelets work. This includes some anti-depressants and high doses of omega 3 fish oils.

It's always recommended to seek advice from your haemophilia centre team if you're uncertain about taking a specific medication.

How should immunisations be given?
You should have immunisations or other injections subcutaneously (under the skin) rather than intramuscularly (into a muscle) to reduce the risk of a painful, bruised swelling (haematoma) developing.

What activities can I do with my bleeding disorder?
Contact sports, which carry a high risk of head injury, need to be avoided, but most day-to-day activities will cause few problems. Your haemophilia centre can help you choose activities that are right for you or your child.

General guidance

If you have bleeding disorder, it is a good idea to:

- Tell your surgeon or dentist about your bleeding disorder if you are due to have a procedure or operation – you may need to take medication to reduce the risk of bleeding before and afterwards. Your surgeon or dentist may also need to contact your doctor to discuss the procedure.
- Tell your doctor or nurse about your bleeding disorder if you need a vaccination. They can give the injection just under your skin to avoid painful bleeding in your muscles.
- Avoid aspirin and non-steroidal anti-inflammatory drugs (NSAIDs) like ibuprofen unless your specialist advises you it is safe to use them, as these can make bleeding worse. Use other medicines such as paracetamol instead.
- Ask your haemophilia doctor or nurse if there are any activities you need to avoid – you should be able to take part in most sports and activities, but it is best to check first.
- Ask your haemophilia centre to provide you with a Bleeding States Information Card – you always need to keep this card with you so if you are involved in an emergency you can show the card and give attending staff the relevant information and phone numbers to call for advice.
- Boys at risk of a bleeding disorder should not be circumcised without first speaking to your haemophilia centre, due to the risk of excessive bleeding.

If you prefer, you can buy a MedicAlert disc or piece of jewellery. As you wear them, they are a good way of making sure your medical details are always to hand. Visit **medicalert.org.uk** to find out more.

Glossary

Carrier
People who inherit a gene fault causing a recessive disorder from one parent only.

Chromosome
A threadlike structure of nucleic acids and protein found in the nucleus of most living cells, carrying genetic information in the form of genes. Humans have 22 chromosome pairs and two sex chromosomes. Females have two X chromosomes; males have an X chromosome and a Y chromosome.

Factor concentrate
A factor protein that has been made into a powder.

Fibrin
Strands of protein that weave around and through a platelet plug to form a blood clot.

Gene
The basic unit of heredity. Each gene has a certain position on a chromosome.

Genetics
The type of science that studies heredity.

Haemophilia
A lifelong, hereditary blood disorder in which bleeding lasts longer than normal. It is caused by a defect in either factor VIII or IX proteins needed for blood clotting.

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

Menstrual period
Also called menstruation or just a 'period'. The shedding of the lining of the uterus through the vagina. On average, it occurs every 28 days and lasts from three to five days.

Placenta
An organ attached to the lining of the womb during pregnancy. It keeps the unborn baby's blood supply separate from the mothers.

Platelet
A small disk-shaped particle in the blood that is used in the clotting process. Also called a thrombocyte.

Platelet function tests
Blood tests to determine how well the platelets work.

Platelet plug
A weak fix of a leaking blood vessel. It is made when platelets begin sticking to each other at the site. Platelet plug formation is part of the blood clotting process.

Tranexamic acid (TXA)
An antifibrinolytic agent available as tablets, a mouthwash or an injection.

About The Haemophilia Society

We are the only UK-wide charity for all those affected by a genetic bleeding disorder; a community of individuals and families, healthcare professionals and supporters.

For 70 years we have campaigned for better treatment, been a source of information and support, and raised the awareness of bleeding disorders.

We want to ensure that everyone affected by a bleeding disorder:

- Has equality of opportunity
- Has the opportunity to connect with others in the community
- Has the knowledge to feel empowered

We do this by:

- Raising awareness about bleeding disorders
- Providing information and support throughout our members lives
- Influencing and advocating on health and social care policy and access to treatment

More than 36,000 men, women and children in the UK have a diagnosed bleeding disorder, and the number rises every year. Membership of The Haemophilia Society is free and open to all. Our peer support through local groups around the UK, global family network, and online community, offers friendship and a listening ear when needed, as well as enabling people to share their views and experiences. By bringing people together for information and support at events tailored to all life stages, we amplify their voices to reduce isolation and influence government, welfare and health care policy.

Our community are at the heart of everything we do – we work collaboratively with members and health professionals to ensure we make decisions influenced by their valued input and direction.

As bleeding disorders are rare, many people will never encounter The Haemophilia Society; we are largely invisible beyond the communities we serve. So, we have to work doubly hard to raise both awareness and understanding of bleeding disorders and vital funds needed to give those affected the services they deserve and need to live the best life they can.

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.**

With special thanks to Dr Mary Mathias, Haemophilia Consultant, Great Ormond Street Hospital and Natalie Lawson, Haemophilia Nurse, Birmingham Children's Hospital for reviewing this booklet

The Haemophilia Society makes every effort to make sure that its services provide up-to-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

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