

Haemophilia A



Information for families

Great Ormond Street Hospital
for Children NHS Foundation Trust

Haemophilia A (also known as Classic Haemophilia or Factor VIII deficiency) is the most well-known type of clotting disorder. A specific protein is missing from the blood so that injured blood vessels cannot heal in the usual way. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Haemophilia A and where to get help.

What is a clotting disorder?

A clotting (or coagulation) disorder is a medical condition where a specific protein is missing from the blood.

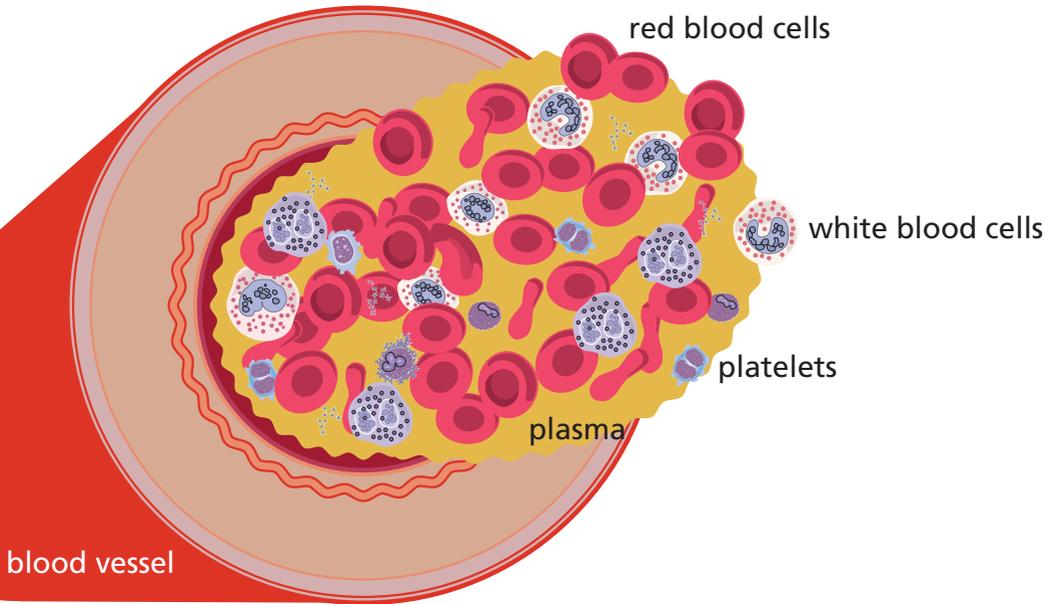
Blood is made up of different types of cells (red blood cells, white blood cells and platelets) all suspended in a straw-coloured liquid called plasma. Platelets are the cells responsible for making blood clot. When a blood vessel is injured, platelets clump together to block the injury site. They also start off a complicated chemical reaction to form a mesh made of a substance called fibrin. This complicated chemical reaction always follows a strict pattern – with each clotting protein (known as a coagulation

factor) turned on in order. When all of the factors are turned on, the blood forms a clot which stops the injury site bleeding any further.

There are a number of coagulation factors circulating in the blood, lying in wait to be turned on when an injury occurs. If any one of the factors is missing from the body, the complicated chemical reaction described above will not happen as it should. This can lead to blood loss, which can be severe and life-threatening. 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.

What is Haemophilia A?

Haemophilia A (also known as Classic Haemophilia or Factor VIII deficiency) is the most well-known type of clotting disorder. The specific coagulation factor that is missing or reduced in people with Haemophilia A is Factor VIII. The severity of symptoms ranges from mild to severe depending on the amount of Factor VIII present in the blood and its activity.



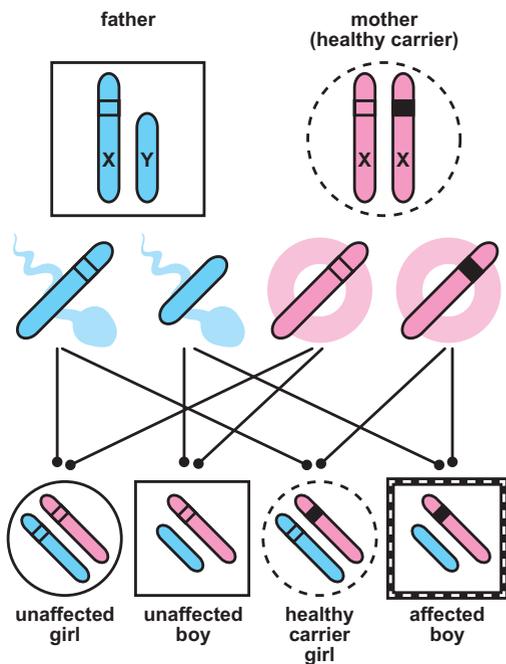
What causes Haemophilia A?

Haemophilia A is caused by a mutation (change) on the Factor VIII gene on the X-chromosome (specifically at location Xq28), which means that only boys are affected and the mother is a carrier of the disease. In most cases, this mutation is passed on from parent to child.

Human beings have about 30,000 to 40,000 different genes, each of which has a function in making an individual person. The genes are arranged in pairs (one of the pair from each parent) on 23 chromosomes – inevitably some of these genes are faulty.

The chromosome that determines the gender of the child will either contain 'XX' (female) or 'XY' (male).

Haemophilia A is inherited as an X-linked condition. As females have two 'X' chromosomes, the fault can be completely or partially overcome by the other healthy 'X' in the pair but in males, who only have one 'X', there is not another 'X' to provide a functioning gene. Unless there have been other affected boys in the family there may be no way of knowing whether the mother is a carrier, as most carriers remain healthy.



Each pregnancy carries a:

- 25 per cent chance of the child being an unaffected non-carrier girl
- 25 per cent chance of the child being an unaffected carrier girl
- 25 per cent chance of the child being an unaffected boy
- 25 per cent chance of the child being an affected boy.

Although Haemophilia A is an X-linked condition and therefore mainly affects boys, it can occur in females but this tends to occur when an affected father has children. Carrier females may have mild bleeding problems, such as heavy periods, but these are not usually severe enough to need treatment.

In some cases, the gene mutation occurs sporadically (out of the blue), with no family history of bleeding disorders.

What are the signs and symptoms of Haemophilia A?

The age at which symptoms appear varies, depending on the amount of Factor VIII in the blood. Children with little or no Factor VIII may start to show symptoms soon after birth, whereas those with some functioning Factor VIII may not show symptoms until later, often following surgery or injury.

Bleeding is the best-known symptom of Haemophilia A – people with the condition may bleed more severely following injury

or for a longer time. This is because the level of Factor VIII in the blood is too low to complete the clotting process as described earlier. Bleeding may occur inside the body as well as from the skin – this can include bleeds inside the joints. It can follow an injury or sometimes no trigger event occurs – this is called a 'spontaneous bleed'. Over time, each bleed can damage the joint making it swollen and harder to bend. Bruising is also common in people with Haemophilia A.

How is Haemophilia A diagnosed?

Haemophilia A can be diagnosed before birth (prenatally) if there is a family history of haemophilia. There are several options for this including chorionic villus sampling (CVS) early in pregnancy or amniocentesis around 20 weeks or so. An alternative is free-foetal DNA testing – this does not diagnose haemophilia but it can identify the sex of the baby in the womb, which is particularly helpful in X-linked disorders that mainly affect males.

After birth, Haemophilia A can be diagnosed using a sample of blood for testing in the laboratory. A test to measure how long a sample takes to clot may suggest a clotting disorder, which would then be investigated further. Doctors will try to identify the gene mutation as well, as this can be helpful for planning future brothers and sisters.

Imaging scans, such as MRI, CT or ultrasound scans may be used to identify any internal bleeds, for instance, inside a joint.

How is Haemophilia A treated?

There are two main methods of treatment – preventative (prophylactic) and on demand treatment to treat bleeds.

Preventative treatment aims to replace the missing or reduced Factor VIII with

a man-made substitute. This is given regularly as an injection, often into a central venous access device such as an implantable port. This means that injections can be given at home which is less disruptive to family life. People with mild Haemophilia A, that is they have some Factor VIII present in the blood, may not need preventative treatment.

On demand treatment is given following an injury or as part of planning surgery, for instance. This injection aims to boost the Factor VIII. It is used on a temporary basis to reduce the side effects of bleeding following an injury or during surgery. There are other measures that can help reduce the effect of a bleed – read our information sheet [Managing bleeds](#) for further information. Physiotherapy will usually be needed following a bleed – this may involve stretches or other exercises or splints to maintain the full range of joint movement. Development of ‘inhibitors’ that mean the body fights off Factor VIII injections can be a problem for people with Haemophilia A. This will be checked regularly at review appointments. If inhibitors develop, additional injections will be needed.

It is important that people with Haemophilia A should not use Non-Steroidal Anti-Inflammatory Drugs (NSAIDs such as ibuprofen) as this greatly increases the risk of bleeding. Other methods of pain relief should be used instead. Caution is needed for injections as

well – immunisations for instance, should be given subcutaneously (under the skin) rather than intramuscularly (into a muscle) to reduce the risk of a painful bruised swelling (haematoma) developing.

Children and young people with Haemophilia A will need regular reviews at their specialist centre to check that they are responding to treatment and not experiencing any side effects. This review will usually involve a check-up from the physiotherapist and dentist as well.



What happens next?

Children and young people with Haemophilia A have a normal lifespan – the introduction of a man-made replacement Factor has greatly reduced the numbers of people developing blood borne viruses following transfusion. Some activities may need to be avoided – such as contact sports that carry a high risk of head injury for instance – but most day to day activities will cause few problems.

Long term effects include the development of 'inhibitors', that is, the body no longer reacts to the Factor VIII injections. This will be checked regularly as part of the review process and can be treated with additional injections. Joint damage can occur in later life especially if there have been many bleeds, which might require surgery.

It is always advisable to wear a medical identity necklace or bracelet to alert health care professionals about having Haemophilia A – details are available via the support organisation. Any surgery will need careful planning in advance as well so it is important that all health care professionals involved are aware.

As Haemophilia A is a genetic condition that can be passed on from parent to child, it is usual to have genetic counselling before planning a family, both for affected individuals and unaffected carriers. Talk to us about a referral to our Clinical Genetics service.

Children and young people with Haemophilia A will need life-long monitoring and treatment, so as your child approaches their teenage years, we will start to talk to them about getting ready to move on to adult health services. This is a planned process so that they become more independent as they grow older and able to manage their own health.

Further information and support

Call the Haemophilia Comprehensive Care Centre at GOSH on 020 7829 8837.

The Haemophilia Society offers support and advice to anyone affected by haemophilia or any other clotting disorder. Call their helpline on 020 7939 0780 or visit their website at www.haemophilia.org.uk.

A wide range of resources is also available on the World Federation of Hemophilia website at www.wfh.org and UK-specific information regarding haemophilia, bleeding disorders and the National Haemophilia Database can be found via the United Kingdom Haemophilia Centre Doctors' Organisation website at www.ukhcd.org. Useful details of Haemophilia Centres when travelling in Europe can be found at www.euhanet.org/centrelocator.

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