Feedback

We appreciate and encourage feedback. If you need advice or are concerned about any aspect of care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From abroad: +44 115 924 9924 ext 85412 or 82301
Deaf and hard of hearing: text 07812 270003
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR

www.nuh.nhs.uk

You can also scan the QR code to leave patient, family or carer feedback. After scanning the QR code please go to 'How can I leave my feedback' and select the service that you have accessed and then follow the on screen prompts.

If you require a full list of references for this leaflet please email patientinformation@nuh.nhs.uk or phone 0115 924 9924 ext. 86623.

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NHS
Nottingham
University Hospitals
NHS Trust

Carriers of haemophilia

Information for patients

Nottingham Haemophilia Comprehensive Care Centre

This document can be provided in different languages and formats. For more information please contact:

Nottingham Haemophilia CCC
Queens Medical Centre
D Floor, East Block
Tel: 0115 970 9994

We Listen
We Care

What is haemophilia?

Haemophilia is a bleeding disorder. People with haemophilia bleed for longer than normal because their blood does not contain enough of a particular clotting factor.

There are two types of haemophilia: haemophilia A where the level of clotting factor 8 (VIII) is reduced and haemophilia B where the level of clotting factor 9 (IX) is reduced.

Haemophilia varies in its severity, depending on the level of the clotting factor in the blood. Haemophilia can be severe (less than 1% of normal clotting factor), moderate (1 - 5% of normal) or mild (5 - 40% of normal levels).

The amount of bleeding a person with haemophilia will have depends on the level of clotting factor in the blood. People with severe haemophilia may have frequent spontaneous bleeding episodes, usually affecting joints like knees, elbows and ankles, or large muscles. People with severe haemophilia will usually be diagnosed before they are a year old.

People with moderate haemophilia may have similar problems but these will occur much less frequently. People with moderate haemophilia may not be diagnosed until later in childhood.

People with mild haemophilia usually only bleed if they have an injury or a medical procedure. It is not unusual for people with mild haemophilia not to be diagnosed until they are older children, adolescents or well into their adult life.

At the moment we have no cure for haemophilia, but good treatment is available.

Notes

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What is the cause of haemophilia?

Haemophilia is an inherited disorder which means that it is passed from a parent to a child through the parent's genes.

Genes are responsible for carrying messages about the way the body develops, grows and functions. In people with haemophilia the gene responsible for the production of clotting factors 8 or 9 have been altered or changed so that not enough is made. The change in the gene is known as a variant or mutation

Haemophilia can occur in families both with and without a family history of haemophilia.

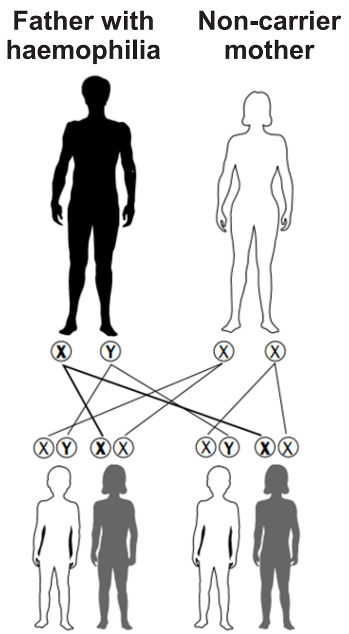
How is haemophilia inherited?

Genes are packaged within the body's cells on structures called chromosomes. The genes which cause haemophilia are located on the X chromosome. This is one of the chromosomes which determines whether a person is male or female.

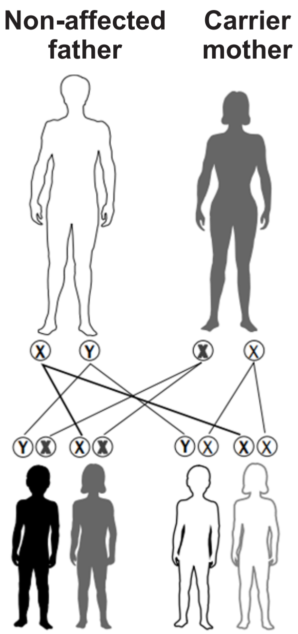
Men have one X chromosome, which they inherit from their mother and one Y chromosome which they inherit from their father. Women have two X chromosomes, one inherited from each parent.

If the X chromosome that a male inherits from his mother has a variant (or mutated) factor 8 or 9 gene, he will have haemophilia.

If a female inherits the variant gene from either her mother or father, she is said to 'carry' the haemophilia gene and is known as a 'haemophilia carrier'. She has one normal gene and one variant gene or mutation.



All daughters of a man with haemophilia will inherit his variant gene and so will be carriers. His sons do not have haemophilia as they inherit their factor gene from their mother.



Carriers have one copy of the variant factor gene. They have a 50:50 chance of passing this gene to each child. Boys who inherit the gene will have haemophilia. Girls will be carriers.

What does it mean to be a carrier?

There are two main things to think about. The first is whether a woman who is a haemophilia carrier is herself at risk of getting problems with bleeding. Most women who carry a variant factor gene have normal levels of clotting factor and do not have problems with bleeding. However some women will have a low level of clotting factor - usually mild haemophilia. They may need treatment to prevent bleeding problems or symptoms.

The second reason why it is important to know whether a woman is a carrier of haemophilia is because of the implications for her in planning her family. If a woman knows that she carries

What happens if my baby has haemophilia?

As soon as we have confirmed the results of your baby's cord blood sample, a member of the haemophilia team will let you know the result.

If your baby has moderate or severe haemophilia, he will have a scan (cranial ultrasound) before he goes home. The haemophilia team will introduce you to the doctors and nurses who will be supporting you and your son and make sure you have all the information you need before you leave hospital.

Where can I find more information?

If you have any questions about the information in this leaflet, please contact your haemophilia team. Our contact details are on the cover of this leaflet.

We can arrange for you to meet a member of the team and can also arrange for you to meet other families affected by haemophilia if you would find this helpful.

We can also arrange pre-pregnancy counselling for carriers of haemophilia and their partners with the Obstetric Haematology Team.

What happens after the birth?

All male babies known to have haemophilia or to be at risk of having haemophilia will have a blood sample taken from the umbilical cord at the time of birth. In many cases we will be able to tell you soon afterwards whether your baby has haemophilia or not.

If the type of haemophilia in your family is mild, sometimes we cannot tell you straight away whether your baby has haemophilia or not. We will need to arrange an appointment for your baby to have another blood test when he is about 6 months old.

Until we know the baby's factor levels he will not receive any intra-muscular injections or blood tests. Vitamin K will be given by mouth.

Most female babies will not have a blood test at birth. This will be in your delivery plan if needed.

After delivery, a carrier's clotting factor level goes back down to her normal pre-pregnancy level quite quickly. This means that a carrier may be at risk of more bleeding than usual after the birth. If your levels are lower than normal, you may require treatment to boost these up as well as other treatment to help your blood to clot properly. Your obstetric team will also make sure that the placenta is delivered safely and you may receive medication to help your womb contract which helps to reduce bleeding.

a variant factor 8 or 9 gene, she can understand her risk of having a baby with haemophilia.

It is important that the birth of a male baby who may have haemophilia is managed safely. It is important to identify if a woman is a carrier in order to know if any precautions need to be taken when she is giving birth to keep her and her baby safe.

Many options are also available for women including reproductive technologies to reduce the chance of having a baby with haemophilia or pre-natal diagnosis to find out if their baby may be affected before birth. We will explain some of these later in this leaflet.

How do I know if I am a carrier?

All males who have haemophilia should have a genetic test to identify the change in the factor 8 or 9 gene that causes haemophilia in the family (called a variant or mutation). There are lots of different gene variants that can cause haemophilia, and the type of variant determines the severity of the haemophilia in that family.

Possible carriers

A woman whose son is the first person in the family to have haemophilia has about an 80% chance of carrying the gene herself. This can be confirmed by testing her genes to see whether she has the same change as her son. If she does not carry the variant gene, this means that the change in the factor gene occurred in the egg cell at the time when her son developed.

A woman whose mother is a haemophilia carrier has a 50:50 chance of being a carrier herself. She can find this out by having a genetic test.

Obligate carriers

A woman is called an obligate carrier if it is known that she is a carrier based on her family history. A woman who has had more than one son with haemophilia is an obligate carrier.

All daughters of men with haemophilia are obligate carriers. Many women who are obligate carriers choose to have a genetic test to confirm that they are a carrier despite their family history. It is important to understand the implications of this testing.

Occasionally unexpected results about family relationships arise from these tests, which, if known, could cause embarrassment within a family. If it is found, for example, that an individual's parent is different from that assumed by the family, significant psychological problems can be caused and this may cause harm to the person being tested and other family members.

What tests can I have?

Two different tests are performed for possible carriers of haemophilia.

Factor levels (or assays) measure the amount of clotting factor in your blood. Some carriers have normal levels and so this test will only tell you whether you may be at risk of having some bleeding symptoms and cannot confirm whether a woman is a carrier or not. Levels can vary significantly among family members (mothers and daughters who are carriers can have very different levels). Factor levels must be done for each known or suspected carrier in a family.

A genetic test is the only way of knowing for sure if you are a carrier. If the gene variant that causes haemophilia in your family is known, then we can reliably tell you if you carry this variant or not. If the gene variant in the family is not known, a normal result in an at risk woman does not completely rule out that she is a carrier.

who carry haemophilia A will have normal levels by the time they are ready to deliver the baby. Factor 9 levels do not usually change in pregnancy. If your factor levels are low towards the end of your pregnancy, you may need treatment to prevent bleeding when you have the baby.

The team will make a delivery plan for you and your baby and you will be given a copy of this. If your baby is a boy with haemophilia, or at risk of having haemophilia, you will need to deliver at Queen's Medical Centre.

If your factor levels are normal, or if you have had treatment to raise them prior to birth, you should be able to have an epidural for pain relief if you choose this for labour.

There is an increased risk that a baby with haemophilia can bleed into his head during and immediately after birth. This risk is higher if the labour and delivery has been long or complicated. Your obstetric haematology team will discuss with you about how to deliver your baby if he is known to have haemophilia or may be at risk.

The best type of delivery depends on a lot of different factors and could be a normal vaginal delivery or a planned caesarean section. Your delivery plan will aim to avoid prolonged labour, and make sure that your baby is delivered in the least traumatic way possible. We will not use forceps to turn the baby, or vacuum extraction (Ventouse) and attaching scalp electrodes or taking blood from the baby's head in labour will be avoided.

If your baby is a girl, no specific precautions are required. Under some circumstances, if your baby is a girl you may be able to deliver at another hospital after discussion with the team.

Amniocentesis - This test is done after about 15 weeks of pregnancy. It is also performed using ultrasound guidance. A long thin needle is inserted into the womb through the abdomen and a sample of the amniotic fluid around the baby is taken. This test carries about a 1 in 100 chance of causing a miscarriage if done in the second trimester of pregnancy.

If you have chosen not to have any pre-natal diagnosis in early pregnancy, you will be offered this test in the last few weeks of pregnancy to guide the plans for delivery. At this time there is a small risk that the test will start labour a little early.

Both of these tests produce a small sample of the baby's cells. We will analyse the sample to see if the baby is a boy or a girl. If the baby is a girl, no further tests will be done. If the baby is a boy, we can look to see if he has the variant that causes haemophilia in the family and tell you whether he has haemophilia or not.

How will my pregnancy and labour be managed?

If you know that you are a carrier of haemophilia, or think that you might be, it is very important that you let us know as soon as you are pregnant. Your midwife or GP must refer you straight away to the Obstetric Haematology clinic at Queens Medical Centre. If you have any concerns, please contact your haemophilia team.

You will be seen in the clinic by a haematology doctor and an obstetrician as well as receiving midwife care. We can discuss all the pre-natal tests described in this leaflet with you and arrange for these to happen if you wish. We can also provide you with any additional information about haemophilia that you may find useful. You will be monitored during your pregnancy in the usual way.

Carriers of haemophilia will also have additional monitoring of their clotting factor levels if these were low outside of pregnancy. The levels of Factor 8 usually rise in pregnancy, so most women

Why is it important to know if I am a carrier?

Women who carry a haemophilia gene and have low factor levels can have bleeding symptoms such as bruising, nose bleeds and heavy periods. They may also bleed more during surgery and dental treatment. Some women may have bleeding after delivering a baby. Treatment is available for bleeding symptoms and to prevent bleeding during surgery and childbirth.

If you are a carrier of haemophilia, there are lots of options available when you are planning your family. Tests are available to find out if a baby is affected with haemophilia before being born. This is called pre-natal diagnosis. There are also other technologies available before a baby is conceived.

It is very important that the birth of a baby who is known to have, or could possibly have haemophilia, is managed safely to protect from bleeding problems. This is why it is important to identify women who are carriers so that we know what precautions to take to make sure her baby is delivered safely.

What options do I have when I am planning my family?

Before getting pregnant

If you know you are a haemophilia carrier or think you might be at risk, it is very important that you have a consultation with a doctor or nurse at your haemophilia centre.

They can arrange tests for you to confirm whether you are a carrier or not. They will explain your risk of having a child with haemophilia. They can also explain all about modern treatment of haemophilia and the options available to you for pre-natal diagnosis, and how a pregnancy would be managed.

Conception options

Some people choose to conceive naturally. They will then be offered the option of pre-natal diagnosis if they wish.

Pre-implantation genetic diagnosis (PGD)

PGD is a technique used in reproductive medicine to identify genetic variants in embryos created through in vitro fertilisation (IVF). It can be offered to a woman who carries haemophilia, providing the genetic cause in the family is known.

PGD can determine whether the embryos produced from her eggs and her partner's sperm have haemophilia or not. Unaffected embryos can then be implanted into the mother's womb. This procedure is available on the NHS, but eligibility needs to be checked for any individual couple requesting the procedure. The success rate for pregnancy with IVF is much lower than natural conception.

What tests can I have when I am pregnant?

Couples who have conceived naturally may wish to know whether their baby is affected by haemophilia before they are born. This is called pre-natal diagnosis. Some people want this information because they may be planning to terminate an affected pregnancy. This decision is a very difficult one and may not be considered for religious, ethical or cultural reasons. Your haemophilia centre team can provide support for you if you need this.

Some people want to find out whether their baby has haemophilia in order to prepare themselves for the birth and afterwards. All the tests that can reliably tell you whether your baby has haemophilia or not involve an invasive medical procedure.

The team managing the pregnancy of a woman at risk of having a baby with haemophilia will always suggest finding out the sex of the baby before they are born to help with managing the birth safely. This is advised whether or not you want to have an invasive procedure.

What types of pre-natal diagnosis can I have?

Non-invasive pre-natal diagnosis

You will be offered tests to give you some information about whether your baby might be at risk of having haemophilia. These tests are non-invasive and just give information about whether the baby is male or female.

You can have a blood test at 8 - 9 weeks of pregnancy to look for the baby's DNA (genetic material) in your bloodstream, this is called a Free Foetal DNA test. If the test shows male DNA in the mother's blood, then it shows the baby is a boy. You can then decide whether to have an invasive test to find out whether he has haemophilia or not. Otherwise the birth will be managed to ensure that it is safe for a baby who could have haemophilia.

You will routinely have a scan at around 20 weeks of pregnancy to see if the baby is developing normally. We will find out at the scan whether the baby is male or female which helps in planning a safe delivery.

Invasive pre-natal diagnosis

There are 2 invasive tests that can be offered which will tell you whether your unborn baby has haemophilia or not.

Chorionic villous sampling - This is usually done between 11 and 13 weeks of pregnancy. This test is done under local anaesthetic using ultrasound guidance. A sample is taken from the placenta, either by inserting a fine needle through the lower part of the abdomen, or by inserting a thin catheter into the vagina.

Haemophilia carriers who have low factor levels may need some treatment to ensure that they do not have any bleeding problems from the procedure. This test carries about a 1 in 100 (1%) chance of causing a miscarriage.