

Free foetal DNA testing for haemophilia

A photograph of a woman with dark hair pulled back, smiling warmly at the camera. She is wearing a light-colored, textured cardigan. In the foreground, a baby with light brown hair and blue eyes is looking directly at the camera. The baby is wearing a white top. The background is a soft-focus indoor setting.

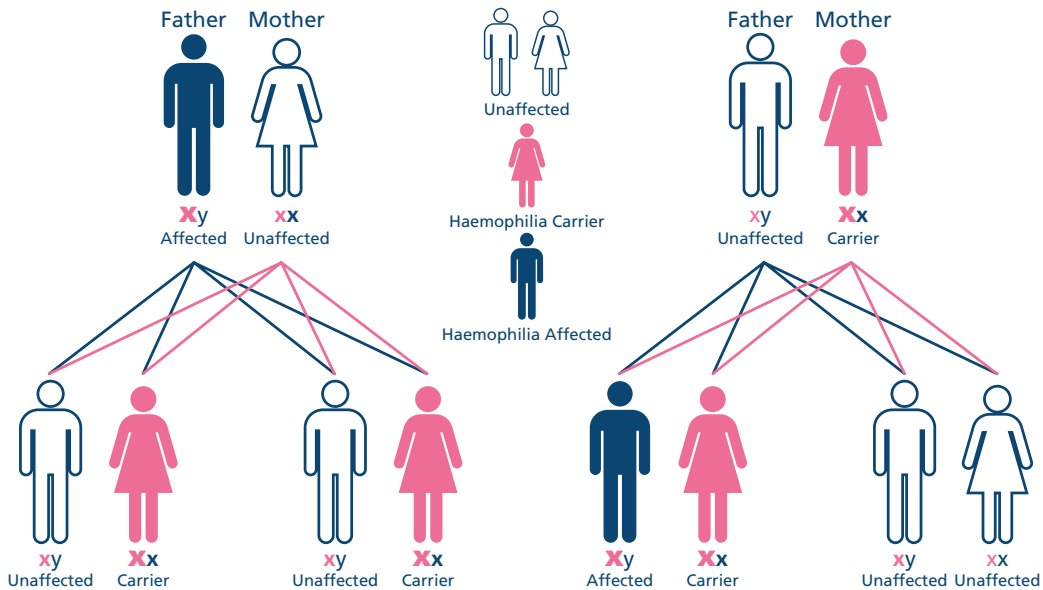
Information for families

Great Ormond Street Hospital
for Children NHS Foundation Trust

This information sheet explains about the free foetal DNA (ffDNA) test, which can be used to determine the sex of a baby in the womb early in pregnancy. It is only suggested when particular inherited disorders are suspected. Further details about the test and what to expect if you come to the Haemophilia Centre at Great Ormond Street Hospital (GOSH) follow.

Haemophilia is an X-linked inherited bleeding disorder caused by a deficiency of 'factors' in the blood. As haemophilia is an X-linked disorder, it only affects males. Carriers of haemophilia have a 1 in 4 chance of having an affected male with each pregnancy (as illustrated in the diagram to the right).

Recent advances in genetic testing allow us to screen your unborn baby to work out its sex early in the first trimester (first three months of pregnancy). This is known as free foetal DNA (ffDNA) testing and is done using a simple blood test. This test can help relieve any anxieties you may have about having another son with haemophilia, particularly if you are considering not continuing with the pregnancy of an affected male. It can also help with planning antenatal care and delivery if you decide to continue with the pregnancy.



What is ffDNA?

Cells from your baby (foetal cells) can be detected in your blood from as early as five weeks of pregnancy. It is these cells that are analysed to work out the sex of the foetus. We do not yet know how these cells enter the mother's bloodstream but we do know that they can be found very early in pregnancy.

How is the ffDNA test done?

The Haemophilia Centre at GOSH offers the ffDNA test from nine weeks' gestation, so first of all, we will need to confirm your pregnancy. We will then take a small blood sample – about 10mls or roughly one tablespoon – and send it to the laboratory for analysis. The scientists in the laboratory look for the presence of the Y chromosome. This is how we know whether the foetus is male or female, as females do not have a Y chromosome.

How long do the results take?

It generally takes about a week for the results to come back and we will contact you directly.

Is it reliable?

At the Haemophilia Centre at GOSH, we have been offering this service to mothers since 2006 and to date we have had 100 per cent accuracy. If there are any doubts about the result, it can usually be confirmed at your 20 week scan.

Is it safe?

As this test only involves a blood sample, rather than anything more invasive, there is no risk to you or your baby. If you are scared of needles, we can work with you to make the test as comfortable as possible.

Why should I have the test?

The ffdNA test allows us to identify the sex of the foetus early in pregnancy. This can reduce the need for further invasive testing later in pregnancy, which carries a risk of miscarriage. Knowing the results earlier than later may also relieve stress for you and your partner.

What happens when I get the result?

If you are having a boy then there are further screening options available to you and we can discuss these fully. If we know that you are having a boy, we can ensure that plans are put in place to manage a safe delivery. Good communication between the haemophilia team and your obstetric (pregnancy and delivery) team is essential. Additionally, if you want peace of mind that you can have a safe and uneventful delivery but would rather not know the sex of your baby, we can still carry out the test but only inform your obstetric team.

Notes

A close-up photograph of a woman with dark hair pulled back, smiling warmly at the camera. She is holding a baby whose face is partially visible in the foreground. The woman is wearing a light-colored, textured cardigan. The background is a soft-focus landscape with rolling hills under a bright sky.

**If you have any more questions, please contact us
at the Haemophilia Centre on 020 7829 8846**

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