PREDICTING YOUR BABY'S SEX BY TESTING FETAL DNA IN YOUR BLOOD

NHS Lothian

A GUIDE FOR PREGNANT WOMEN WHO ARE CARRIERS OF X-LINKED CONDITIONS

This leaflet gives you information about the fetal sex prediction test now available during pregnancy to women who are carriers of X-linked conditions.

This test is non-invasive and can help identify the sex of the baby (fetus) early in pregnancy without the need for an invasive test which poses a risk of miscarriage.

INTRODUCTION

Genes contain the DNA that determines many of our characteristics, including whether we are male or female. We now know that some of the fetal DNA circulates in the mother's blood during pregnancy. We call this cell-free fetal DNA (cffDNA).

This cffDNA can be tested and the fetal sex predicted by taking a sample of the mother's blood in pregnancy. At the moment the test can only tell us the baby's sex, not whether the baby is affected by a particular condition.

HOW IS THE TEST DONE?

You will be offered the test by your obstetric or genetic doctor. The timing is important as the test works best from nine weeks of pregnancy.

First, we will perform an ultrasound scan to find out how many weeks pregnant you are and to see if there is more than one baby in the womb. We can only offer the test if there is one baby.

The test is performed on a sample of the mother's blood. About 20mls of blood is drawn into a special tube. Only males have a Y chromosome, so if the test identifies DNA from a Y chromosome in the mother's blood, the fetus is likely to be male. Absence of a Y chromosome but presence of fetal DNA predicts a female pregnancy and further confirmation is needed.

The cffDNA is lost from the maternal blood stream within a few hours of delivery and so testing is specific to the baby in any given pregnancy.

HOW ACCURATE IS THE TEST & WHAT WILL THE RESULTS SHOW?

The accuracy of the test is currently around 98% and there are three possible outcomes:

1: Y chromosome DNA is detected: the pregnancy is likely to be male.

2: No Y chromosome DNA detected: the pregnancy is likely to be female. Fetal sex will be confirmed at your 20 week anomaly scan.

3: Test Failure: e.g. insufficient DNA. In a few cases the lab may not be able to demonstrate the presence of fetal DNA. In this situation the test could be repeated or you may wish to discuss the option of an invasive test (chorionic villus sampling or amniocentesis).

4: The Birmingham laboratory asks for repeat samples if an <u>inconclusive</u> result is generated or for apparently female result on an early gestation (i.e. before 9 weeks' gestation) sample.

HOW SAFE IS THE TEST?

As this is a blood test taken from your arm, the test carries no significant risk to you or your baby.

HOW LONG DOES IT TAKE TO GET THE TEST RESULTS AND HOW WILL I GET THEM?

It usually takes around five working days to get the results. Your doctor or fetal medicine midwife will discuss this with you, including how you will get your test results.
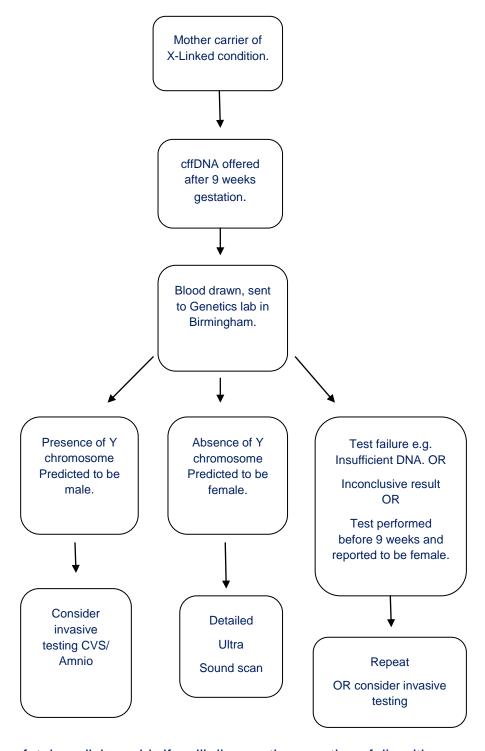
WHAT HAPPENS NEXT?

What happens next depends on the results of the fetal sex prediction test.

If your baby is predicted to be male, you might decide to have an invasive test (Chorionic Villus Sampling (CVS) or an amniocentesis) to determine if the baby has the condition you are concerned about.

Separate leaflets describing these tests are available

Alternatively you might decide to continue with the pregnancy without any further testing.

If your baby is predicted to be female, the fetal sex would be confirmed at your detailed anomaly scan at 20 weeks of pregnancy.

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        │  Mother carrier of  │
        │  X-Linked condition.  │
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        ┌─────────────────┐
        │   cffDNA offered    │
        │   after 9 weeks     │
        │     gestation.      │
        └─────────────────┘
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        ┌─────────────────┐
        │  Blood drawn, sent  │
        │  to Genetics lab in │
        │     Birmingham.     │
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Presence of Y chromosome Predicted to be male.	Absence of Y chromosome Predicted to be female.	Test failure e.g. Insufficient DNA. OR Inconclusive result OR Test performed before 9 weeks and reported to be female.
Consider invasive testing CVS/ Amnio	Detailed Ultra Sound scan	Repeat OR consider invasive testing

Your doctor or fetal medicine midwife will discuss these options fully with you.

Leaflet adapted from RAPID leaflet by EF, CD and MP, August 2013.

RAPID website

Information for patients about Reliable Accurate Prenatal non-Invasive Diagnosis.
www.rapid.nhs.uk

CONTACT DETAILS / FURTHER INFORMATION

Your local genetic centre

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