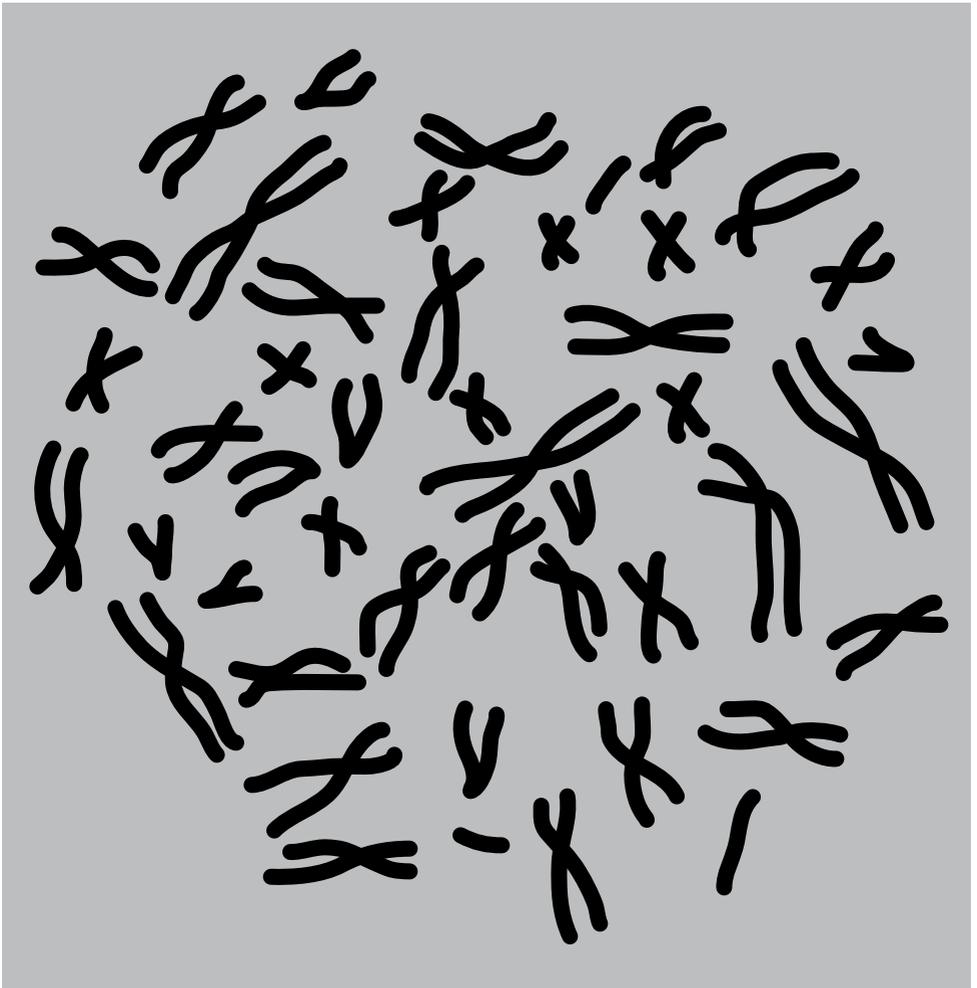


Fetal Medicine Unit

Chromosome and genetic testing in pregnancy

Information for parents



Introduction

This leaflet has been written to give you information about diagnostic tests in pregnancy called CVS and amniocentesis. Knowing this information may help you decide whether you wish to have either of the tests.

You may have been offered a diagnostic test because:

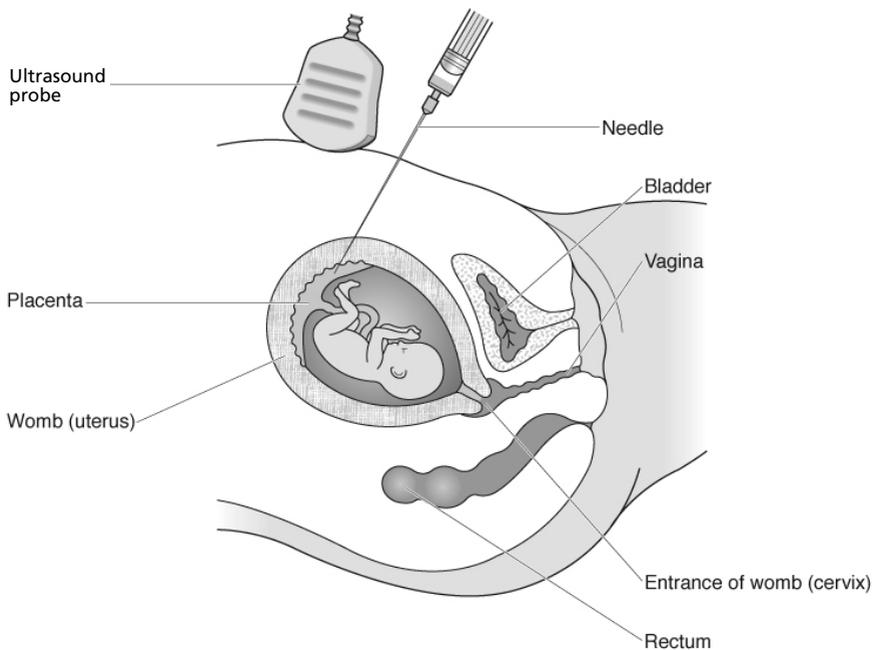
- you have a family history of an inherited (genetic) disorder
- you have had a child or previous pregnancy with a chromosome disorder
- you are a carrier of an inherited condition
- your screening test for Down's syndrome shows you are at increased risk of having a baby with Down's syndrome or other chromosome abnormalities
- you have had an ultrasound scan which shows that your baby may have an increased risk of having abnormal chromosomes.

It is your choice to accept or decline a test. This leaflet can help you to make that decision.

Chorionic Villus Sampling (CVS)

CVS can be performed after 11 weeks of pregnancy. Using an ultrasound for guidance, a fine needle is used to take a tiny sample of the developing placenta. The placenta contains tissue that is genetically identical to your baby. This tissue is examined in the laboratory to look at the chromosomes or to check for specific genes or conditions.

It is not always possible to perform CVS for technical reasons, such as when the placenta is in a position where it is difficult to take a sample.

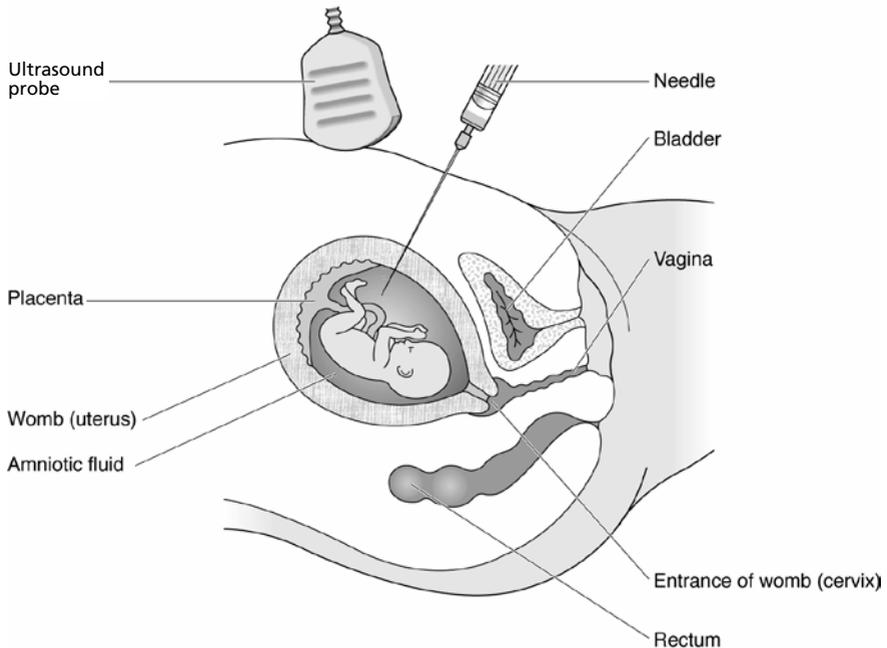


What are the risks of having CVS?

CVS has a chance of causing a miscarriage. We estimate this risk to be about 1 in 100. A miscarriage may occur up to three weeks after the CVS. We cannot predict which pregnancies will miscarry. CVS is performed early in pregnancy when miscarriages are slightly more common.

Amniocentesis

Amniocentesis can be performed after 15-16 weeks of pregnancy. Using an ultrasound for guidance, a fine needle is used to take a small amount of amniotic fluid from around your baby. The fluid contains cells from your baby. These are examined in the laboratory to look at the chromosomes or to check for specific genes or conditions.



What are the risks of amniocentesis?

Amniocentesis has a chance of causing a miscarriage. The risk is slightly lower than in CVS. We estimate the risk to be between 1 in 100 and 1 in 200. A miscarriage may occur up to three weeks after the amniocentesis but we cannot predict which pregnancies will miscarry.

Chromosome tests

Karyotype

This is the standard diagnostic test to check all your baby's chromosomes.

The laboratory grows the cells from the sample taken. This test will detect many types of chromosome abnormalities, not just Down's syndrome, and will give you a definitive result. It can also tell you the sex of your baby if you wish to know. The results take 2-3 weeks.

Occasionally it is not possible to get a full result, or the result is not clear and a further test is required. This is more likely to happen after CVS than amniocentesis.

Rapid results

Rapid testing (called PCR or FISH) can be performed in addition to the karyotype test. Rapid testing will be done if there is very high suspicion of a problem or can be done as a private test by request (and charged for). This test is to specifically examine for three chromosome abnormalities:

- Down's syndrome (trisomy 21)
- Edward's syndrome (trisomy 18)
- Patau's syndrome (trisomy 13)

The results take about 3-4 working days.

Single-gene disorders/inherited conditions

If you have been offered a diagnostic test for a specific genetic disorder or condition, the results will come separately from the chromosome test. These results are usually given to you by your specialist.

Accuracy of test results

We aim to offer tests that are proven to be safe and accurate, however there are limitations with the standard karyotype test which means very small alterations within chromosomes may not be detected.

If you decide to have a diagnostic test

Preparing for the test:

- please bring your maternity notes with you
- you can eat and drink normally before the test
- you can bring your partner or a friend with you, but please do not bring children
- you will need someone to drive you home afterwards.

Your appointment

Please be prepared to spend up to a couple of hours at the hospital. You will spend some time with one of the Specialist Midwives and the procedure will then take about fifteen to twenty minutes.

The procedure is performed by a doctor in one of the scan rooms. You will have an ultrasound scan first so the doctor can see the position of the placenta or identify a pool of amniotic fluid. Using the ultrasound for guidance, a fine needle is used to take the sample. The procedure may be uncomfortable. Sometimes the sample cannot be taken and the doctor may suggest you return on another day.

If you have a Rhesus Negative blood group you will be given an injection of anti-D after the procedure. This is to reduce the possibility of antibodies developing in your blood.

After the procedure

You should stay in the hospital for at least half an hour. If you feel well after this time, you may go home and rest. Some women have mild cramping pains afterwards, but this should settle with rest and a normal dose of paracetamol.

We advise you take it easy for a couple of days, depending on your lifestyle or occupation. You will also need to avoid sexual intercourse, heavy lifting or strenuous exercise. Most women are back to normal after a couple of days.

Signs to look out for

Please contact Fetal Medicine during clinic hours, your local maternity unit or your GP if you experience any of the following:

- severe abdominal pain which does not settle with rest and a normal dose of paracetamol
- vaginal bleeding
- fluid leaking from your vagina
- suddenly feeling unwell, with a high temperature or flu-like symptoms.

Getting the results

The Specialist Midwives will talk to you about how you want to receive your result.

Rapid results (whether Private or NHS) take 3-4 working days and are received late in the afternoon. These results are usually given to you by telephone.

Karyotype results are posted to your home address by the Cytogenetics laboratory, if the results are normal. If the results are abnormal the Specialist Midwives will contact you. The analysis takes 2-3 weeks to be completed. Sometimes the results are delayed when the cells take longer than expected to grow in the laboratory. You will be contacted if this happens.

Very occasionally the cells fail to grow and a result will not be possible. Occasionally, a CVS sample is too small to grow and there may only be a limited result available. If this happens you will be informed and offered the opportunity to discuss a repeat procedure.

When a problem is found

If a problem is found, we will give you information and support to help you make a decision about your pregnancy. Some parents will want to prepare themselves for the birth knowing their baby has a problem. Some parents may decide not to continue with the pregnancy. These are difficult decisions and we will give you time and information to help you make the decision that is right for you.

How to contact us

Fetal Medicine Unit

Tel: **01865 221 716**

Monday to Friday, 9am - 5pm

Level 6, Women's Centre

John Radcliffe Hospital

Oxford OX3 9DU

Further information

Further information can be found on the following websites:

www.healthtalkonline.org

www.screening.nhs.uk/screeninginfo

www.arc-uk.org

www.rcog.org.uk

If you have a specific requirement, need an interpreter,
a document in Easy Read, another language, large print,
Braille or audio version, please call **01865 221 473**
or email **PALSJR@ouh.nhs.uk**

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