

Diagnostic Tests in Pregnancy

KEY	
	Hospital Entrance
	Disabled Entrance
<small>*The QPark Shuttle Bus is a free service and is available from various locations around the QE site to transport patients and visitors to the main QE hospital, BWH hospital and QEP hospital buildings.</small>	Disabled Parking
	Parking
	Railway Station

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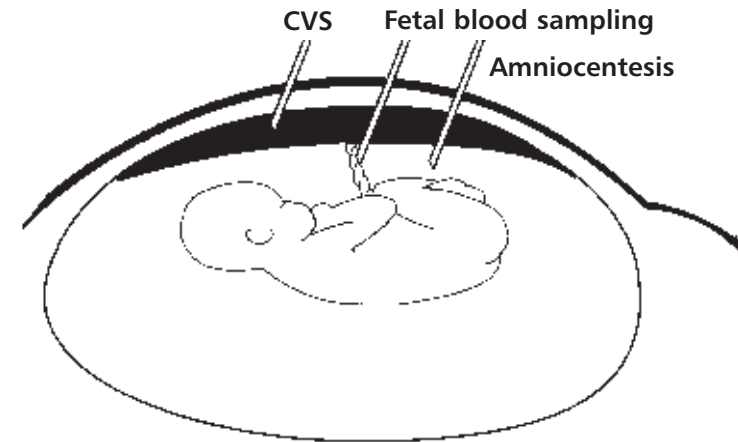
Birmingham Women's **NHS**
NHS Foundation Trust

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Information about
Amniocentesis (Amnio) and
Chorionic Villus Sampling (CVS)

Patient Information Leaflet

This diagram illustrates amniocentesis and CVS



Birmingham Women's Hospital contact numbers

Fetal Medicine Centre
0121 627 2683

Antenatal screening midwives
0121 623 6959

Main switchboard
0121 472 1377

Delivery Suite/enquiries
0121 472 3032 / 627 2665

Final results

A full check of all of the baby's chromosomes (called a full karyotype) will also be performed when you have an amniocentesis or CVS to check that every pair of chromosomes appears to be normal. This full result normally takes 2-3 weeks.

The baby's sex will be determined by looking at the chromosomes and we will ask you before the test if this is something you would want to know.

We will discuss how you will receive your results with you before you have the test.

After having a diagnostic test

You will not need to stay in hospital when you have an amniocentesis or CVS and you will be able to go home after the test if finished.

It is advisable to rest following the test and for the following day

- You should bring someone with you and not drive yourself home
- You should avoid heavy lifting and strenuous exercise for the following few days
- You may take a normal dose of paracetamol for discomfort or soreness following the test
- You can eat and drink as normal
- You can bath and shower as usual
- If you have Rhesus negative blood group then a 'anti-D' injection will be given

If you experience pain that is becoming worse or not going away with rest, fluid loss or bleeding vaginally, suddenly feel unwell or feverish or are worried about any other symptoms you should call 0121 627 2665, explaining the problem and the test you have had.

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Diagnostic (invasive) tests

Diagnostic tests are most often offered to parents when a screening test or an ultrasound scan has found there is an increased chance of their baby having a chromosomal or genetic abnormality.

If parents have had a baby with genetic abnormality before or have a condition that runs in their family an appointment with a genetic specialist will usually have been arranged before a diagnostic test is carried out.

Diagnostic tests look at a baby's chromosomes (genetic information in cells) and sometimes at specific genes when a particular condition is suspected.



In a normal human cell there should be 46 chromosomes, 23 pairs, including the sex chromosomes. Chromosomes are the structures that carry genes, and each chromosome carries hundreds or thousands of genes.

Diagnostic tests are not routinely offered to everyone in pregnancy because they carry a risk of miscarriage. The benefits, risks and possible complications will be discussed with you before a test is arranged.

This leaflet gives information about amniocentesis (amnio) and chorionic villus sampling (CVS).

Results for amniocentesis and chorionic villus sampling

Fast test results

A small amount of the fluid or placental tissue taken when you have a diagnostic test is used to obtain a fast result (called a qfPCR).

This is done for 3 particular chromosomes pairs - 13, 18 and 21 - because problems with these 3 pairs are more common than with other chromosomes. If the baby has 3 copies of one of these chromosomes, instead of the usual 2, this is called a trisomy and means they will have one of the most common syndromes:

- Down's syndrome (an extra chromosome 21)
- Edward's syndrome (an extra chromosome 18)
- Patau's syndrome (an extra chromosome 13)

In some cases the baby's sex chromosomes are also looked at on the fast test but this depends on why the test has been performed and is not done in every case.

This fast test result is normally ready within 2-4 working days of having the test.

Although it is a very reliable technique, sometimes it is not possible to get a result.

For example, if the sample also contains some of the woman's blood. In this case no results would be available until the full check of the baby's chromosomes was ready in 2-3 weeks (see *following page*).

The woman's stomach is cleaned with very cold antiseptic fluid and a local anaesthetic is usually given to numb the skin before a fine needle is passed through the skin, through the outside of the womb and into the placenta. Tissue is taken through the needle - this may take 30 seconds to a minute; depending how easy it is to reach the placenta and obtain enough tissue to perform the test.

The whole test normally takes around 30 minutes and may be uncomfortable.

What are the problems with CVS?

- It can cause miscarriage – this happens to around 1 in every 50 -100 women (1-2%) who have the test.
- The most common time for miscarriage to happen is in the first 3-4 days after the test but it can happen further along in the pregnancy.
- Some women feel sore or uncomfortable during or after the test and experience period type pain.
- Sometimes a CVS is not possible due to the position of the placenta – this may mean the test is delayed for a week or so or not possible at all (in which case we would discuss amniocentesis with you).
- It is not always certain how a particular chromosomal problem will affect your baby – this may mean we cannot tell how severe or complicated the problem will be.
- Sometimes a different chromosomal problem to the one suspected can be found.
- Sometimes the results of the test take longer than expected, the cells do not grow at all and sometimes CVS results are difficult to interpret. This is because the cells are taken from the placenta rather than the baby's own cells. We would discuss what this means for your results and if further test needed.

Amniocentesis (amnio)

What is amniocentesis?

This is a test that removes a small amount of the fluid, which surrounds the baby in the womb.

It is normally done between 15 and 22 weeks of pregnancy but may sometimes be done later.

Shed skin cells from the baby, which float in the fluid, can be grown (cultured) in a laboratory and examined to check the genetic make up (chromosomes) of the baby.

You may be offered an amniocentesis because:

- You have a higher chance of having a baby with Down's syndrome or other chromosomal condition.
- You have a family history of an inherited genetic / chromosomal condition, such as Sickle cell anaemia.
- You have had a baby with a genetic / chromosomal condition before.
- A problem has been found during an ultrasound scan and a genetic / chromosomal condition is suspected.

How is an amniocentesis performed?

An ultrasound scan is carried out to see where the baby is lying and how much fluid is around the baby. The scan is used throughout the procedure so that the needle can be seen and directed in to the right place, away from the baby.

The woman's stomach is cleaned with very cold antiseptic fluid before a fine needle is passed through the skin, through the outside womb and into the sack where the baby is lying. About 15 millilitres of fluid is taken and then the needle is removed.

The fluid taken away is replaced by the baby over the next few days. The whole procedure takes around 15 minutes – but the needle is only in the womb for around 30 seconds. The test is usually no more painful than having a blood test. No anaesthetic is needed.

What are the problems with amniocentesis?

- It can cause miscarriage - this happens to around 1 in every 200 women (0.5%) who have the test.
- The most common time for miscarriage to happen is in the first 3-4 days after the test but it can happen further along in the pregnancy.
- Some women feel sore or uncomfortable during or after the test.
- It is not always certain how a particular chromosomal problem will affect your baby - this may mean we cannot tell how severe or complicated the problem will be.
- Sometimes a different chromosomal problem to the one suspected can be found.
- Sometimes the results of the test take longer than expected and occasionally the cells do not grow at all – we would discuss what this means for your results and if the test needed to be repeated.
- If a chromosomal abnormality is found and you decide not to continue the pregnancy then labour would be induced to deliver the baby - a termination in theatre is not an option at this stage of pregnancy.

Chorionic Villus Sampling (CVS)

What is CVS?

This is a test that removes a small amount of tissue from the developing placenta (afterbirth).

It is normally done between 10½ and 13 weeks of pregnancy but may sometimes be done later.

The placenta originates from the same cells as the baby. When the cells are taken from the placental tissue and grown (cultured) in a laboratory, the genetic make up (chromosomes) should be the same as the baby.

You may be offered a CVS because:

- You have a higher chance of having a baby with Down's syndrome or other chromosomal condition.
- You have a family history of an inherited genetic / chromosomal condition, such as Sickle cell anaemia.
- You have had a baby with a genetic / chromosomal condition before.
- A problem has been found during an ultrasound scan and a genetic / chromosomal condition is suspected.

How is a CVS performed?

An ultrasound scan is carried out to see where the baby is lying and where the placenta is attached to the inside of the womb. The scan is used throughout the procedure so that the needle can be seen and directed in to the right place, away from the baby.