

CVS and Amniocentesis



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This leaflet has been written to give you more information about CVS and amniocentesis. These tests are not offered routinely in pregnancy and it is your choice whether to have a test or not.

What is CVS?

‘CVS or ‘chorionic villus sampling’ is a diagnostic test that can be performed between 11–14 weeks of pregnancy. First the skin on your tummy is cleaned with antiseptic. Then, local anaesthetic is injected into your skin and the surface of the womb. This stings for a few seconds only and makes the way to the placenta numb. Using ultrasound scanning to guide the way through the numb area, a needle is inserted through your abdomen into the womb and a tiny piece of the placenta is removed, a process that usually takes a minute or so. Cells from the placenta usually have the same genetic make-up as the baby. These cells can be analysed to detect a chromosomal abnormality in the baby. Occasionally it is not possible to obtain a CVS because of the position of the placenta; in these cases it may be necessary to wait and have an amniocentesis later in the pregnancy.

What is an amniocentesis?

This is also a diagnostic test and performed in a similar way to a CVS. The skin on your tummy is cleaned and under ultrasound guidance, a needle inserted through the abdomen into the womb. A sample of the amniotic fluid that surrounds the baby is removed. The fluid contains some of the baby’s cells which can be analysed. Amniocentesis is performed later in the pregnancy, from around 14 or 15 weeks onwards. It takes about five to ten minutes, though the sampling itself usually only takes a minute or so and a local anaesthetic is not necessary.

Who might consider CVS or amniocentesis?

CVS and amniocentesis are the only way a chromosomal abnormality can be diagnosed with certainty during pregnancy.

These tests are offered:

- If you feel that you have had a high risk result following a screening test (such as after a combined screening or quadruple test)
- If you have previously had a pregnancy affected by a chromosome or genetic disorder that may recur
- If an ultrasound scan has detected features indicating an increased risk of a chromosome abnormality
- If you are concerned that your baby may have a chromosome abnormality and you wish to have a definite answer
- If there is a family history of a genetic disorder and specialist genetic counselling has advised that CVS or amniocentesis would be able to detect this condition in your unborn baby.

What information will a CVS or amniocentesis give you about your baby?

The test provides you with information about your baby's chromosomes.

Chromosomes are tiny structures within every cell in our body that carry our genes. Normally there are 22 matching numbered pairs of chromosomes and a 23rd pair labelled X or Y which determine the sex of your baby. Thus normally there are a total of 46 chromosomes in each cell. Problems may arise if there are too many or too few chromosomes or they are arranged in the wrong order.

The most common chromosome abnormality is Down's syndrome. People with Down's syndrome have 47 chromosomes in each cell instead of 46. This is caused by an extra copy of chromosome number 21 and is also known as trisomy 21.

Remember that CVS and amniocentesis will usually only tell you about the number of chromosomes in your baby. However in special cases, these tests can also be used to test for some inherited genetic conditions. Not all inherited conditions can be detected in pregnancy and it is NOT routine to look for these unless you have a family history of inherited genetic disease.

Your doctor or specialist midwife will discuss this with you in more detail if these extra tests are relevant in your case.

Likewise, CVS and amniocentesis do not detect spina bifida. An ultra-sound scan at 20 weeks is offered to look for this condition and other structural abnormalities in your baby.

Are CVS and amniocentesis safe for the mother and baby?

CVS and amniocentesis are not risk-free. Both tests carry a risk of causing a miscarriage. About one in every hundred (1%) tests will cause miscarriage, usually within a week of the procedure. The additional overall risk of miscarriage from amniocentesis is about 1% (1 in every 100 women who have an amniocentesis), while the additional overall risk of miscarriage from CVS is 1 to 2% (1 to 2 in every 100 women who have CVS). The miscarriage may be the loss of a baby with normal or abnormal chromosomes.

What can you expect after the test?

Many women chose to rest for the day after the procedure, but there is no evidence this affects the risk of miscarriage. It is common to experience some abdominal discomfort or mild cramping, often described as period type pain, during the next 24-48 hours. This is relatively common and it may be helpful to take some painkillers (it is safe to take paracetamol).

These symptoms usually settle within a couple of days, however if you are concerned please contact us.

You should also contact us if:

- you experience a lot of pain which persists
- you have vaginal bleeding or lose fluid
- you develop a high temperature
- you feel unwell.

Remember – in the majority of women, the pregnancy continues without further problems.

How will I receive the result?

You can choose how to receive your results. In most cases we will phone you as this is likely to be the quickest way. However we can arrange for you to come to the hospital to be given the results in person although we cannot be sure exactly when the results will be back. Your doctor or midwife will discuss which method you would prefer.

How long do the results take to come back?

Usually the results will take two to three working days. In a few cases further tests are carried out on the sample and this will take a further two to three weeks. Your doctor will advise you as to which tests are indicated and when results can be expected:

Rapid test (QF - PCR)

All samples have a rapid test, also called the QF-PCR, which usually takes 2 to 3 working days. This test checks for the three most common chromosome conditions.

It gives a result for:

- Down's syndrome – Trisomy 21 – an extra chromosome 21;
- Edward's syndrome – Trisomy 18 – an extra chromosome 18;
- Patau's syndrome – Trisomy 13 – an extra chromosome 13.

The QF-PCR test does not detect other chromosome rearrangements, low level mosaicisms (where most cells are normal but a few cells have an extra chromosome) or abnormalities of the sex chromosomes. However, we know that most women with an increased risk after screening for Down's, Edward's or Patau's syndromes are only at increased risk for one of these three conditions. Therefore in most cases it is not necessary to perform any other tests and this is the only result you will receive.

Further tests: Full Karyotype and Array CGH

Further tests are *only* performed if there are concerns that your baby is at an increased risk of having an abnormality involving a chromosome that can't be checked for on the rapid (QF-PCR) test. The specialist genetic laboratory will decide which test will give the most useful information about your baby according to your clinical history or scan findings. Your doctor or specialist midwife will explain why one of these further tests has been offered.

Here are some common reasons why further tests might offered:

- If there is a family history of a chromosome re-arrangement (e.g. translocation)
- If problems in a previous pregnancy have indicated an increased risk of recurrence of a chromosome abnormality
- If it was not possible to get a rapid (QF-PCR) result for technical reasons
- The rapid (QF-PCR) result was abnormal
- If ultrasound scanning has detected an abnormality that could be related to a chromosome problem.

Full karyotype (Full Culture)

This test examines all 46 of the baby's chromosomes and detects all major chromosomal abnormalities, trisomies and most other major chromosome rearrangements. However it cannot detect all chromosomal abnormalities. It takes around two weeks for the result to come back.

Array CGH (microarray based comparative genomic hybridisation)

This test can examine each chromosome in much more detail than is possible on a full karyotype, so it is possible to detect very small chromosome imbalances. Such imbalances are known as micro-deletions and micro-duplications:

- **Micro-deletions:** here a tiny part of a chromosome is missing, so the baby might have some genes missing which may cause problems.
- **Micro-duplications:** here an extra tiny part of a chromosome is found, so the baby might have some extra genes which may cause problems.

If we find a micro-deletion or a micro-duplication we would always refer you to the specialist genetics team to help you understand the result. Such imbalances do not always cause a problem in the baby.

Although array CGH can be performed on all the chromosomes, sometimes it is only necessary to examine just one or two chromosomes. This is known as targeted array CGH.

Array CGH cannot detect balanced translocations or low level mosaicisms however. Results take around two weeks.

Occasionally a problem will be detected with one of the chromosomes on the full karyotype or array CGH that could not be tested for in the rapid PCR result. It is very rare to find such a problem and is nearly always unexpected. It can be very shocking to be told that there is such a problem, especially after receiving a normal result for the rapid test. These cases often require input from a counsellor or doctor who is a specialist in genetics and we will arrange this as required.

Will the test tell me the sex of my baby?

In most cases the result will NOT tell you the sex of your baby. However, in some situations it may be important to know the sex of the baby in order to offer specialist advice. For example:

- if there is a family history of a sex-linked inherited condition;
- if ultrasound scanning detects an abnormality that may be related to an abnormality of the sex chromosomes.

In these rare situations it is possible to determine the sex by rapid QF-PCR test which will take two – three working days.

In addition, samples undergoing full karyotyping always report the baby's sex as part of the result. Samples undergoing array CGH may report the baby's sex but not in all cases.

Where the baby's sex has been reported, we would only inform you of the sex if you ask specifically.

Will the test need to be repeated?

In about one in every hundred (1%) cases, the test will need to be repeated. This is slightly more likely following a CVS. This is because cells may not grow in the laboratory or the results are inconclusive.

What happens after I receive the results?

Normal results

The majority of women who have a CVS or amniocentesis will be given a normal result, their pregnancy will continue with no problems and they will give birth to a normal baby. However, a normal result does not exclude the possibility of other abnormalities. In some cases we may ask to see you again for further scans and investigations.

Abnormal results

Unfortunately, in a few cases, the results of the CVS or amniocentesis will be abnormal. You will be told what the abnormality is and how this could affect your baby. This information can be given to you over the phone but we will also arrange for you to be seen by a specialist midwife or doctor for further counselling within two to three days. After discussion, you and your partner may consider whether or not to continue with the pregnancy.

In the case of CVS, where abnormal results are reported from the rapid (QF-PCR] test and there are no ultrasound abnormalities, it is often recommended that women wait for confirmation of the result from the full karyotype before deciding on further management. Your doctor would discuss this with you in more detail if it applies to you.

If you continue with the pregnancy we will put you in touch with various support groups to improve your understanding of your baby's condition. We can also arrange for you to see our paediatric team to discuss the care your child may require at birth and beyond. We may also ask to see you again for further scans to monitor your pregnancy closely and advise on the best time, method and place for your delivery.

If you decide to have a termination of pregnancy, we will help make the necessary arrangements for the procedure. The decision whether or not to continue with the pregnancy always rests with the parents.

A word about Anti-D

If your blood group is rhesus-negative (one in eight women have this blood group), you will be given an injection of Anti-D after the procedure. Its purpose is to prevent the mother developing antibodies against the fetal blood cells, which could cause problems in this or future pregnancies. If you need an injection, your doctor or midwife will discuss this further.

IMPORTANT:

You should read the following before making a decision about CVS or amniocentesis...

Before a CVS or amniocentesis is carried out, it is important for you to consider what difference an abnormal result would make to your wish to continue with the pregnancy. Having information about your baby's chromosomes has to be balanced against the small but real risk of losing the pregnancy.

Remember: It is always your choice whether to have this procedure or not. Please discuss any concerns you may have with your doctor or midwife.

For more information

If you have further questions about amniocentesis or CVS, you can speak to your doctor or midwife. You might also want to contact the following:

Antenatal Screening Co-ordinator:

Local midwife specialist for antenatal screening and testing.

For further discussion of tests and results please call **07876 357 423**

ARC – Antenatal Results and Choices:

National organisation providing support and information about screening and diagnosis of fetal abnormality.

www.arc-uk.org Helpline 0845 077 2290

Down's Syndrome Association:

Provides information and support for people with Down's syndrome, their families and carers.

www.downs-syndrome.org.uk Helpline 0208 614 5100

S.O.F.T. UK:

Provides information support for families affected by Patau's syndrome (Trisomy 13) and Edward's syndrome (trisomy 18).

www.soft.org.uk Helpline 07923 056 132

If you have vision, mobility or access issues please contact the specialist screening midwife team on 07876 357 423 for further information. You can also call the midwives at the hospital in the antenatal clinic on:

Royal Sussex County Hospital 01273 696955 (ask for antenatal clinic)

Princess Royal Hospital 01444 441881 (ask for antenatal clinic)

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The information in this leaflet is for guidance purposes only and is in no way intended to replace professional clinical advice by a qualified practitioner.

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