



Chorionic villus sampling (CVS)

This leaflet gives you information about a procedure that you have been offered, which is called **chorionic villus sampling (CVS) test**.

You may have been offered CVS because:

- you have a family history of an inherited disorder;
- you already have a child with a chromosome disorder or you had a previous pregnancy (which did not go to full term) and a chromosome disorder was found;
- you carry an inherited condition (for example, sicklecell disorders); or
- you have had an ultrasound scan which shows that your baby may have an increased chance of having abnormal chromosomes.

If you decide to have CVS, you can find more information about the procedure on page 6 of this leaflet.

This information may help you decide if you want to have this procedure.

You can also ask your midwife or obstetrician for more information.



Introduction

CVS is a procedure during which a doctor removes a small amount of tissue from your placenta (afterbirth) during pregnancy. The cells in this tissue are tested in the laboratory to look at your baby's chromosomes. You can usually have CVS from your 11th week of pregnancy to your 14th week, but in special circumstances you can have it done after 14 weeks.

If you have CVS, there is a 2% risk that you could have a miscarriage (that means one in every 50 women could lose their baby). A miscarriage is most likely to happen up to three weeks after the CVS. No one knows why this happens or who it will happen to. It can happen whether or not your baby has a chromosome problem. CVS is done early in pregnancy which is when miscarriages are slightly more common.

Because CVS is a specialised procedure, you may not be able to have it done in the maternity unit you usually go to. Instead, you may be offered an appointment at a different maternity unit.

Chromosome tests

In each cell of our bodies, we usually have 46 chromosomes (23 pairs). These are numbered from 1 to 22, with the 23rd pair being the sex chromosomes: XX for a girl, XY for a boy. Each chromosome carries thousands of genes, which are the instructions that tell our bodies how to grow, develop and function.

In Wales, two tests are done on the sample of tissue that a doctor takes during CVS. These are called the **PCR** test and the **karyotype** test.

PCR (polymerase chain reaction) test

The PCR test usually only looks for the following three chromosome problems in the baby:

- Down's syndrome (causes some degree of learning disability but is not life-threatening for the baby);
- Edwards's syndrome (a severe chromosome disorder that can be life-threatening for the baby); and
- Patau's syndrome (a rare and severe chromosome disorder that can be life-threatening for the baby).

If the ultrasound scan you had earlier in your pregnancy (to find out your due date) showed that your baby may

have a higher chance of Turner's syndrome, an extra test is carried out on the PCR test sample. Turner's syndrome only affects females.

It is possible that the karyotype test (see the next section) might detect other chromosome problems. This can happen even if your PCR result is normal.

Karyotype

This test takes longer than the PCR test but will detect most chromosome problems.

To do this test, the cells from your placenta are grown (or cultured) in the laboratory. The laboratory staff will examine the cells under a microscope to look for changes in the number and appearance of your baby's chromosomes.

But even a karyotype test will not detect **all** chromosome changes. Some changes are so small that they can't be seen, even under the microscope, and can only be detected using specialist tests. These tests are not done on a karyotype.

The karyotype test will not detect:

- changes in single genes, such as cystic fibrosis (each chromosome contains thousands of genes);

- microdeletions (loss of small segments of a chromosome); or
- other small changes in chromosomes.

Single-gene disorders

Single-gene disorders are caused by changes in a single gene. If you have a family history of an inherited disorder that is caused by an alteration in a single gene (such as cystic fibrosis), you may be offered a specific test, called a single-gene test, just for this. You may already have spoken to someone at the genetics service, or your midwife or obstetrician can refer you to the genetics service if you tell them you have a family history of an inherited disorder. If a single-gene test is done on CVS sample, you will usually be offered a karyotype test as well.

A karyotype test doesn't show how your baby is growing and developing. Only an ultrasound scan at between 18 and 20 weeks, when your baby is nearly fully formed, can show this.

If there is a problem

There will be some pregnancies in which problems develop. If your tests show there is a problem, you will be given information and support to help you make a decision about your pregnancy. There are experts who can give you information to help you decide what to do. These include fetal medicine consultants, geneticists and paediatricians, among others.

Some women will want to prepare themselves for the birth knowing that their baby has a problem. Some women may decide to end their pregnancy, while others may decide to continue with the pregnancy and give their baby up for adoption. These are difficult decisions and you will be given information to help you make the right decision for you.

Finding out that your unborn child has a problem is distressing and deciding what to do is hard. Most women want and need some support. This might come from your partner, family or friends, or from the health professionals who are caring for you. Your partner or a friend can come to hospital appointments with you, if you want them to.

The karyotype can detect many types of chromosome disorders. This can mean that although your baby does not have Down's syndrome, Edwards's

syndrome, Patau's syndrome or Turner's syndrome, the test result could show a different problem.

If this happens, you would be able to discuss the result with a doctor and with a midwife who specialises in antenatal screening. You would also be able to discuss the result with someone who specialises in genetics.

We may need to ask you and the baby's father to provide blood samples for karyotyping as well to decide whether your baby has a problem.

More information

You can get more information from:

Antenatal Results and Choices (ARC)
73 Charlotte Street
London
W1T 4PN.

Helpline: 0207 631 0285
Website: www.arc-uk.org
E-mail: info@arc-uk.org

If you decide to have CVS

Preparing for CVS

- You can have breakfast or a light lunch before your appointment.
- You will need a full bladder for the scan you will have before the CVS.
- You may be asked to empty your bladder for the CVS procedure itself.
- You may be more comfortable if you wear loose clothing.
- You can bring your partner or a friend with you for support during and after the procedure, but please don't bring any children with you.
- If possible, arrange for someone to drive you home.

Having the CVS done

The procedure takes about 20 minutes and you will have it done as an outpatient, usually in the antenatal clinic. You will be awake for the procedure, and lying down.

You will have an ultrasound scan before the CVS. This is to check the baby's position and to find the best place to take the sample from your placenta.

The doctor will give you an injection of local anaesthetic to numb the skin of your abdomen. Your abdomen is then cleaned with an antiseptic solution to reduce the risk of infection. The doctor inserts a needle through your skin and the wall of your womb, and then takes a small sample from the placenta. The doctor will be watching the ultrasound scan to guide the needle and so avoid getting close to the baby. You may find the test a little uncomfortable.

The doctor or midwife will then examine the sample under the microscope to see if there is enough tissue to do the test. If not, the doctor will need to take another sample.

You will also be asked to give a blood sample so that the laboratory can be sure that the results they get from the CVS sample are for your baby rather than for you.

What happens after the procedure

After the procedure, you should stay in the clinic for up to 30 minutes to rest. You may have tummy cramps afterwards, rather like period pains.

If your blood group is Rhesus negative, you will be offered an injection of anti-D after the procedure. This is to reduce the chance of antibodies developing in your blood, which could happen if your baby's blood group is Rhesus

positive.

Some doctors may advise you to take things easy for a couple of days after the procedure, and to avoid having sex, or doing any heavy lifting or strenuous exercise. You won't need complete bed rest.

If you have any pain or discomfort, you can take a normal dose of paracetamol.

Most women are back to normal after two days.

What you should look out for

- Severe pain, which you can't control by taking mild painkillers (like paracetamol).
- Any bleeding or unpleasant discharge from your vagina.
- Any fluid leaking from your vagina.
- If you suddenly feel unwell, with a high temperature or flu-like symptoms.

These symptoms do not always mean there is a problem but you may need further care and attention. For advice, please contact:

- the clinic where you had the CVS;
- your midwife; or
- your GP.

Getting the results of the test

Your PCR test result is usually available within three days, and the karyotype test result is usually ready within two weeks. If the PCR test result shows that your baby has a problem, we may ask you to wait for the result of the karyotype before making any decisions about your pregnancy.

The midwife or doctor will discuss with you how you want to get your result. You may want to come into the hospital or you may want to be given your result by phone. If you choose to have the result by phone, please remember that if the test has shown there is a problem with your pregnancy, you might prefer to get this information in person so that you can speak to someone who can answer your questions. Results of tests for single-gene disorders take different lengths of time. You will need to make your own arrangements for getting your results with the genetics team before you have the test done.

Sometimes it takes a bit longer to grow the cells in the laboratory and you will have to wait a few extra days for the karyotype result. If the cells grow more slowly, this does not mean there is a problem with your baby.

Occasionally, in fewer than one in 100 tests, the cells do not grow in the

laboratory so you will not get a result from the test. You may be offered a repeat CVS procedure.

The karyotype test will show the sex of your baby. If you don't want to know whether it's a boy or a girl, please tell your midwife or doctor beforehand.

You can get more information about CVS results from your midwife or from the clinic where you had the CVS done.

Local contact details