Chorionic villus sampling (QF-PCR and array CGH tests)

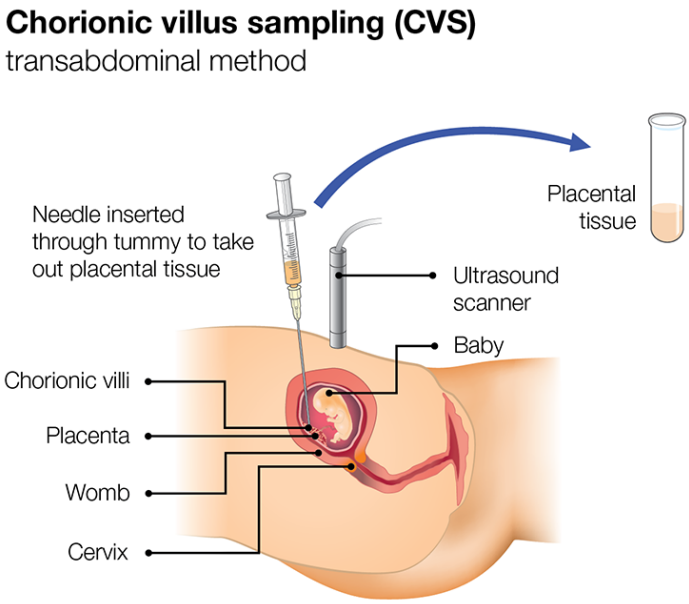
We have given you this factsheet because we would like to offer you a diagnostic test called chorionic villus sampling (CVS). It explains what CVS is, what the test involves, what the possible benefits and risks are for you and your baby, and what your options are if your baby is found to have a chromosomal or genetic condition.

This factsheet aims to support the discussions you will have with us and your midwife. It is important that you take time to consider your options and ask any questions you may have before you decide whether or not having CVS is the right choice for you.

What is chorionic villus sampling (CVS)?

CVS is a diagnostic test which involves us removing and testing a tiny sample of cells from your placenta. We usually perform this test between the 11th and 13th week of pregnancy.

We will also need to take a blood sample from each parent to assist with this analysis, as it will allow us to recognise naturally inherited variations. A member of our team will arrange this for you.



Why am I being offered CVS?

CVS is an optional test. We will only offer you CVS if:

- your screening test has shown that your baby has a higher chance of having a chromosomal or genetic condition
- you have had an ultrasound scan which suggests your baby may have physical differences
- you have a higher risk of passing on an inherited abnormality to your baby, such as sickle cell disease or thalassaemia major
- you have previously had a baby who was born with a chromosomal or genetic condition

This is because CVS is an invasive test and can increase your risk of a miscarriage.

Does CVS have any limitations?

Using both a QF-PCR and an array CGH test, CVS will detect whole extra chromosomes and small missing or extra fragments of chromosomes, but it will not detect all genetic or chromosome changes. It:

- may not pick up very subtle chromosome imbalances as some are too small to be detected
- does not provide information about tiny changes in individual genes so may not pick up all genetic conditions
- does not test for spina bifida or other physical differences
- cannot detect conditions such as autism or cerebral palsy
- may detect a chromosome change that we cannot fully interpret, and so it can be difficult to know how, or if, this change is linked to ultrasound findings
- can occasionally identify an unexpected chromosome change which is unrelated to the ultrasound findings but may have implications for the future health of your baby or the health of other family members

Are there any risks?

Miscarriage

The main risk associated with CVS is miscarriage (losing your baby in the first 23 weeks). Approximately one in every 200 women who have CVS will miscarry. The exact cause of miscarriage after CVS is unknown, but it may be caused by factors such as infection, bleeding or damage to the amniotic sac that surrounds your baby.

Due to the risk of miscarriage, you may wish to discuss the procedure with your partner, or a close friend or family member before making your decision. We will also be happy to discuss this with you and can recommend support groups available to you.

How should I prepare for the procedure?

Blood tests

Before the procedure, you must have had the following blood tests:

- Blood group – This test will ensure we give you (with your consent) an appropriate amount of anti-D if you are rhesus D negative. For more information about this, please speak to your midwife or obstetrician.
- Screening for HIV and hepatitis B infections – This test will ensure we take appropriate precautions to minimise the risk of transmission to your unborn baby, if the presence of these infections is known.

Food and fluids

You can eat and drink as normal before and after the procedure. Please come to your appointment with a comfortably full bladder, as this will lift your womb out of your pelvis, which will make it easier for us to check the position of your baby and placenta.

Ultrasound scan
We will carry out an ultrasound scan before the procedure to measure your baby and check the position of your placenta.

What will happen during the procedure?

Assuming your placenta is accessible (we are able to get to it easily), we will begin the procedure by cleaning your abdomen. We will then inject a local anaesthetic into the skin of your abdomen to numb it. We will then insert a fine needle through your abdomen and into your placenta. We will continuously check the position of your baby and placenta using an ultrasound scan while we do this. Using the needle, we will remove a small sample of the placental tissue. We will send this tissue to a laboratory for analysis.

What will happen after the procedure?

We recommend that someone drives you home after the procedure.

You will be able to return to a normal level of daily activity as soon as you feel able to do so. There is no evidence to support the need to rest in bed or avoid strenuous activity.

After the procedure, you may experience some mild discomfort for which you can take paracetamol. Contact the fetal medicine team or your local maternity hospital immediately if you experience any of the symptoms below:

- severe pain
- bleeding
- loss of vaginal fluid
- flu-like symptoms

What will happen to the placental tissue sample?

We will send the sample of placental tissue to the laboratory where two different types of laboratory test can be performed: a QF-PCR and an array CGH. The tests we perform will depend on the reason why we have offered you CVS. We may also carry out some additional tests that look for changes to specific genes, depending on your medical history. We will discuss this with you in more detail.

What are the laboratory tests (QF-PCR and array CGH)?

The laboratory tests will examine the chromosomes in your baby's cells. Chromosomes are structures within each of your baby's cells which carry genes. Genes are instructions which tell the body how to develop and function. Having too much or too little chromosomal material can cause significant problems with your baby's physical and intellectual development.

The QF-PCR test can detect:

- **extra chromosomes** such as those found in Downs's syndrome (Trisomy 21), Edward's syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13).

The array CGH test can detect:

- **missing chromosomes or chromosome rearrangements**. If the QF-PCR test result is normal, but you have had an ultrasound scan which suggests your baby has physical differences, we will perform an additional laboratory test known as an array CGH. This test looks for small deletions (tiny pieces of chromosome that may be missing) or duplications (tiny pieces of extra chromosome). If deletions or duplications have occurred, this may explain the ultrasound findings and allow you to receive more precise information about the implications for your baby.
- **sex chromosome abnormalities** such as Turner's syndrome (Monosomy X).

Additional specific gene tests, if required, can detect:

- **genetic conditions** such as sickle cell disease, if they are known about in your family.

Results

The QF-PCR result will usually be available two to three working days after the laboratory has received the sample. The array CGH test result takes longer and is usually available within 14 days. Additional specific gene tests may take longer if testing for rare conditions.

We will contact you by telephone with the results. If an anomaly is identified or suspected, you will have the opportunity to discuss your options with us and your midwife. You will also be offered information and support from support groups outside the health service.

You may wish to continue with your pregnancy. Alternatively, you may wish to consider adoption or termination of pregnancy. No one can make this difficult decision for you. It is important to think through your options very carefully and come to a decision that is right for you.

Occasionally, the QF-PCR test result may be unclear. This is due to a rare condition called confined placental mosaicism (CPM), in which the DNA in your placenta is not the same as your baby's DNA. We may offer you a second invasive test called an amniocentesis if this happens.

Contact us

If you have any further questions or would like to discuss having CVS in more detail, please do not hesitate to contact us.

Fetal medicine team
Telephone: **023 8120 6025**

Useful links

Antenatal Results and Choices (ARC)
Website: **www.arc-uk.org**

The Down's Syndrome Association
Website: **www.downs-syndrome.org.uk**

Support Organisation for Trisomy 13/18
Website: **www.soft.org.uk**

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For help preparing for your visit, arranging an interpreter or accessing the hospital, please visit **www.uhs.nhs.uk/additionalsupport**

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