

Patient information factsheet

Chorionic villus sampling (QF-PCR test)

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 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 you 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.



Chorionic villus sampling (CVS)

Why am I being offered CVS?

CVS is an optional test. We will offer you this test if an earlier screening test has suggested that you have a higher chance of having a baby with chromosome differences, such as Down's syndrome (Trisomy 21), Edward's syndrome (Trisomy 18) or Patau's syndrome (Trisomy 13).

Does CVS have any limitations?

Using a QF-PCR test, CVS will only detect changes due to whole extra chromosomes. 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.

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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

loss of vaginal fluid

bleeding

• flu-like symptoms

What will happen to the placental tissue sample?

We will send the sample of placental tissue to the laboratory for analysis, where a QF-PCR test will be performed. The QF-PCR test is able to identify some extra chromosomes, such as those found in Down's syndrome (Trisomy 21), Edward's syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13).

Results

The QF-PCR result will usually be available two to three working days after the laboratory has received the sample.

We will contact you by telephone with the results. If an anomaly is identified, 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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