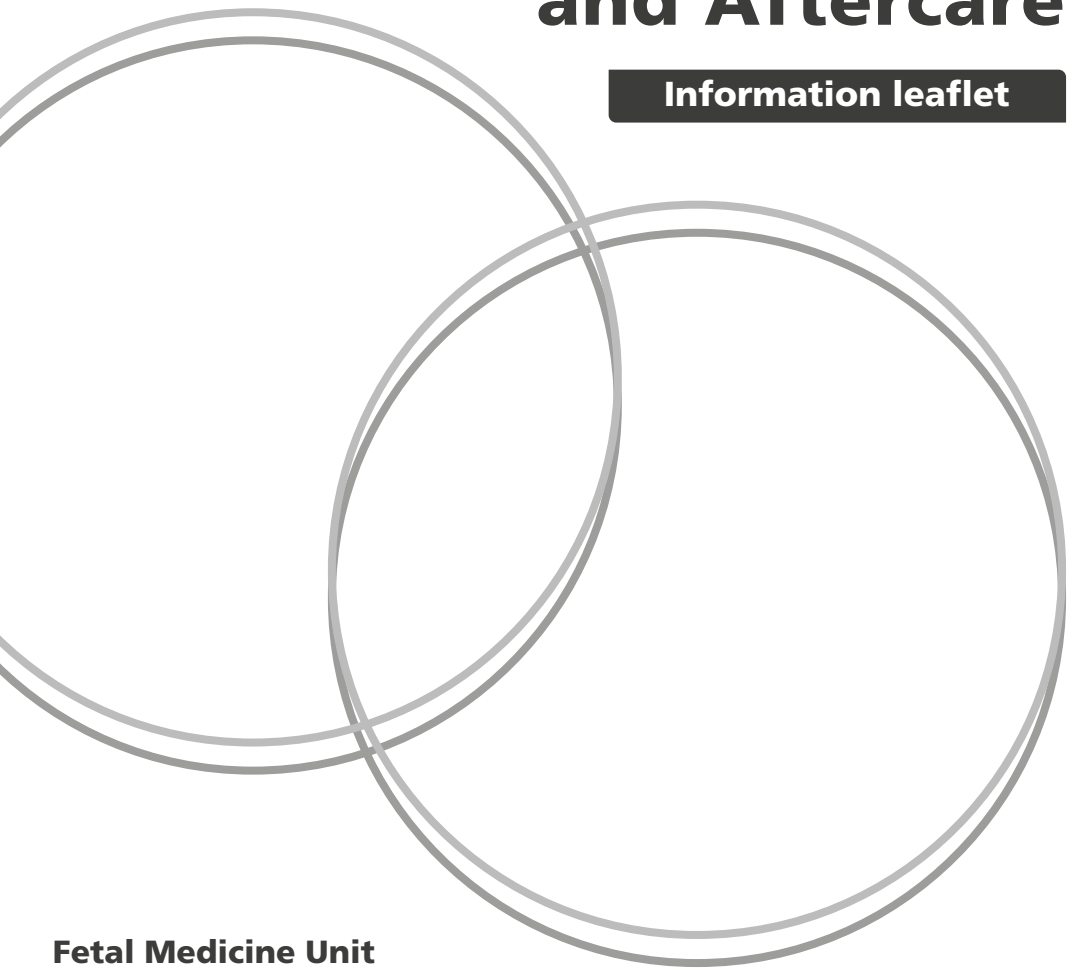
Oxford University Hospitals
NHS Foundation Trust

Chorionic Villus Sampling (CVS) or Amniocentesis Prenatal Diagnostic Tests and Aftercare

Information leaflet



Fetal Medicine Unit

Gender inclusive language in OUH Maternity and Perinatal Services:

- This leaflet uses the terms woman, women and mother throughout. These terms should be taken to include people who do not identify as women but who are pregnant. Similarly, where the term parent(s) is used, this should be taken to include anyone who has main responsibility for caring for a baby.
- The term partner refers to the woman's chosen supporter. This could be the baby's father, the woman's partner, a family member or friend, or anyone who the woman feels supported by and wishes to involve in their care.

You have been given this leaflet because we would like to offer you a CVS or amniocentesis. It explains what CVS and amniocentesis are, what each test involves and what the possible benefits and risks are for you and your baby.

Diagnostic tests like Chorionic Villus Sampling (CVS) and amniocentesis tell us if a baby is affected by a chromosomal condition by checking their chromosomes for genetic information.

The leaflet aims to support the discussions you will have with the midwife and doctor. It is important that you take time to consider your options and to ask any questions you may have before you decide whether having CVS or amniocentesis is the right choice for you.

This leaflet explains the invasive testing procedures we offer in this department. There are two invasive procedures that are possible as prenatal diagnostic testing, Chorionic villus sampling (CVS) and amniocentesis. Prenatal diagnostic tests take place in our Fetal Medicine Unit (FMU) and are performed by FMU consultants (senior doctors), alongside our FMU midwives and maternity support workers.

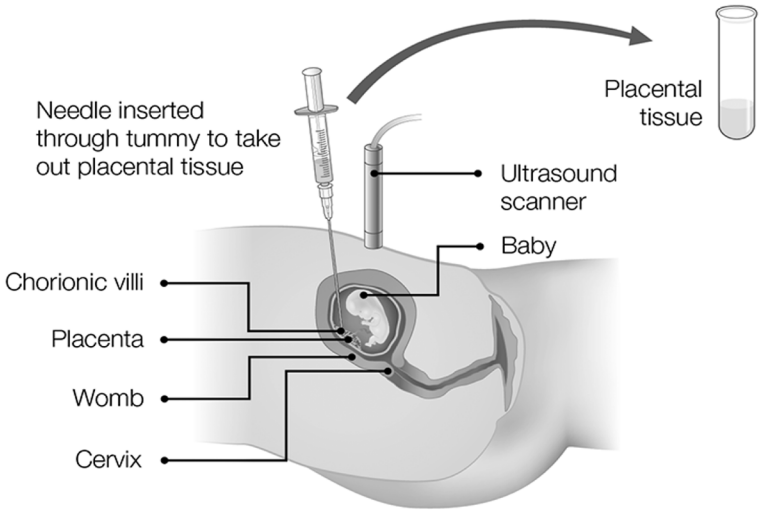
Contents

Transabdominal Chorionic Villus Sampling (CVS)

What is a chorionic villus sampling (CVS)?

A CVS is an in-utero (in the womb) diagnostic test which involves us removing and testing a tiny sample of cells from your placenta. We usually perform this test ideally between 11 and 14 weeks of pregnancy but this can be done slightly later in specific cases if needed.

What happens during the procedure?



Your abdomen (tummy) will be cleaned with antiseptic fluid and a local anaesthetic (pain relieving) injection may be used to numb a small area of skin. The aim is to take a tiny sample of tissue from the placenta for testing. A fine needle is inserted through your abdomen and into your uterus (womb) to take the sample. The sample contains some of the baby's cells which contain genetic information (DNA). An ultrasound probe is used throughout the procedure to scan and guide the direction of the needle. This tissue sample is then sent to a laboratory for analysis (examination). This is a diagnostic test and offers 99.9% accurate test results.

Preparation for the procedure

Food and fluids

You can eat and drink as normal before and after the procedure. Please come to the appointment with a comfortably full bladder as this will allow your uterus (womb) to rise which will make it easier for the position of the baby and placenta to be checked.

Ultrasound scan

Please prepare to discuss the procedure with an FMU midwife prior to the test. An ultrasound scan is performed before the CVS procedure to measure the baby and to check the position of the placenta.

Blood tests

Please prepare to have your blood taken following a successful procedure:

- **Blood group testing** – This test will ensure we give you an appropriate amount of anti-D if you have a rhesus negative blood group. (If this blood test has been performed earlier and is already available, it will not need to be repeated). For more information about this, please speak to your midwife or obstetrician (hospital doctor).
- **Maternal (mother of the baby) blood test** – This test may be required to reduce the chance of what is known as 'maternal cell contamination'. It is very likely that we will collect some maternal cells in addition to the baby's cells during a CVS. In order for the laboratory to ensure the baby's cells are tested correctly, we may need to have a sample of maternal blood too. The maternal blood will also be used as a comparison to the tested baby cells. Should a change in the baby's chromosomes be found, the maternal blood can be helpful in detailing the significance of this change, and we will explain how this may affect the baby and further care options. Sometimes we may need to talk with the genetics team, who may also offer to contact you for further discussion.

- **Paternal (father of the baby) blood test** - In some instances, we would also recommend taking the blood from the father of the baby. The father of the baby's blood will be used as a comparison to the tested baby cells. If a change in the baby's chromosomes is found, paternal blood can be helpful in detailing the significance of this change, and we will explain how this may affect the baby and further care options. Sometimes we may need to talk with the genetics team, who may also offer to contact you for further discussion.

CVS results

QFPCR

- The first test, called a QFPCR or RAPID test, usually takes 5 days in total, this does not include the day the sample was taken.
- Occasionally, the laboratory cannot provide a result for Down's Syndrome (chromosome 21), Edward's Syndrome (chromosome 18) or Patau's Syndrome (chromosome 13) within the expected timeframe. This is because the cells may need time to grow before the DNA can be extracted and a result can be obtained. If this happens, you will be informed of the need to wait for the full results.
- We will telephone you with the results as soon as we receive them.
- When we call you to tell you the results (whether they are normal or abnormal) we will always ensure you feel the time and place of the discussions feels safe to you before we continue.
- If you have specific instructions about how you wish to receive your results, please let us know and we will try to accommodate any preferences.

Array-CGH analysis

- Array-CGH is not always performed, as only certain structural abnormalities qualify for this test.
- The array CGH analysis which checks for smaller changes, deletions or rearrangements in the chromosomes, can take 2 to 3 weeks for the result. This does not include the day the sample was taken.
- We will telephone you with the results as soon as we receive them.
- When we call you to tell you the results (whether they are normal or abnormal) we will always ensure you feel the time and place of the discussions feels safe to you before we continue.
- If you have specific instructions about how you wish to receive your results, please let us know and we will try to accommodate any preferences.

General results advice

- If the QFPCR result or the array-CGH analysis shows a change in the baby's chromosomes, we will discuss this with you. We will explain how this may affect the baby and further care options. Sometimes we may need to talk with the genetics team, who may also offer to contact you for further discussion.

Does the CVS have any limitations?

- In rare instances, less than 1 in 100 cases, an unusual result is obtained due to a rare condition called confined placental mosaicism (CPM). This is where the sampled DNA from the placenta is not the same as the baby's DNA. In these circumstances, we may offer you an amniocentesis to verify the result and baby's DNA and would offer further detailed explanation from our genetics team.
- It does not test for spina bifida (a condition that affects the spine) or other physical differences.

- It cannot detect conditions such as autism or cerebral palsy.
- It can occasionally find an unexpected chromosome change which is unrelated to the ultrasound findings but that may have implications on the future health of the baby or the health of other family members.

The FMU midwives are there to support you and to offer explanation of the results as needed. If further counselling by the specialist obstetric and genetic teams is required, this will also be arranged for you.

Are there any risks?

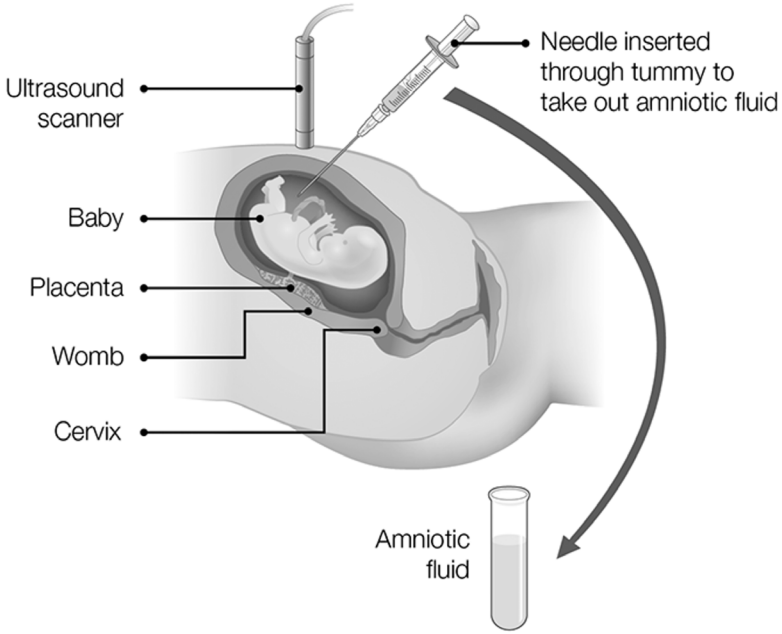
- Less than 1 in 200 women will suffer a miscarriage (loss of the baby) after having a CVS.
- We do not know why some women miscarry after having a CVS but causes of miscarriage may include infection and suspected fetal abnormalities. Most miscarriages happen within 3 days of the procedure, but they can happen up to 2 weeks afterwards.
- It is not always possible to get a result from the first CVS or amniocentesis. Up to 6 in 100 women will be offered a second procedure if the first procedure is unsuccessful.

You may choose to discuss the procedure with a partner or close friend or family member before deciding about whether you want to have this test or not. We are here to discuss this with you and can provide you with contacts of groups that can offer additional help and support.

Transabdominal Amniocentesis

What is amniocentesis?

Amniocentesis is an in-utero (in the womb) diagnostic test which involves removing and testing a small volume of amniotic fluid (the fluid surrounding the baby). This test is usually performed from 15 weeks of pregnancy, but it can be done later in pregnancy from approximately 32 weeks also if needed.



Your abdomen (tummy) is cleaned with an antiseptic fluid and a fine needle is passed through your abdomen and into your uterus (womb) to take the sample. The sample contains some of your baby's cells which contain genetic information (DNA). An ultrasound probe is used throughout the procedure to scan and to guide the direction of the needle. The fluid sample is then sent to a laboratory for analysis (examination). This is a diagnostic test and offers 99.9% accurate test results.

Preparation for the procedure

Food and fluids

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

Ultrasound scan

An FMU midwife will discuss the procedure with you prior to the test. An ultrasound scan will also be performed before the amniocentesis procedure to measure the baby and check the position of the placenta.

Blood tests

Please prepare to have your blood taken following a successful procedure:

- **Blood group testing** – This test will ensure we give you an appropriate amount of anti-D if you have a rhesus negative blood group. (If this blood test has been performed earlier and is already available, it will not need to be repeated). For more information about this, please speak to your midwife or obstetrician (hospital doctor).
- **Maternal (mother of the baby) blood test** – This test may be required to reduce the chance of what is known as 'maternal cell contamination'. It is very likely that we will collect some maternal cells in addition to the baby's cells during an amniocentesis. In order for the laboratory to ensure the baby's cells are tested correctly, we may need to have a sample of maternal blood too. The maternal blood will also be used as a comparison to the tested baby cells. Should a change in the baby's chromosomes be found, maternal blood can be helpful in detailing the significance of this change, and we will explain how this may affect the baby and further care options. Sometimes we may need to talk to with the genetics team, who may also offer to contact you for further discussion.

- **Paternal (father of the baby) blood test** – In some instances, we would also recommend taking the blood from the father of the baby. The paternal blood will be used as a comparison to the tested baby cells. If a change in the baby's chromosomes is found, paternal blood can be helpful in detailing the significance of this change, and we will explain how this may affect the baby and further care options. Sometimes we may need to talk with the genetics team, who may also offer to contact you for further discussion.

Amniocentesis results

QFPCR

- The results of the first test, called a QFPCR or RAPID test, usually take 5 days to process.
- Occasionally, the laboratory cannot provide a result for Down's Syndrome (chromosome 21), Edward's Syndrome (chromosome 18) or Patau's Syndrome (chromosome 13) within the expected timeframe. This is because the cells may need time to grow before the DNA can be extracted and a result can be obtained. If this happens, you will be informed of the need to wait for the full results.
- We will telephone you with the results as soon as we receive them.
- When we call you to tell you the results (whether they are normal or abnormal) we will always ensure the time and place of the discussions feels safe to you before we continue.
- If you have specific instructions about how you wish to receive your results, please let us know and we will try to accommodate any preferences you may have.

Array-CGH analysis

- This test is not always required, as not all women will qualify for this additional testing.
- The result of the array CGH analysis which checks for smaller changes, deletions or rearrangements in the chromosomes, can take about 2 to 3 weeks to process. This does not include the day the sample was taken.
- We will telephone you with the results as soon as we receive them.
- When we call you to tell you the results (whether they are normal or abnormal) we will always ensure the time and place of the discussions feels safe to you before we continue.
- If you have specific instructions about how you wish to receive your results, please let us know and we will try to accommodate any preferences.

General results advice

If the QFPCR result or the array-CGH analysis shows a change in the baby's chromosomes, we will discuss this with you. We will explain how this may affect the baby and the further care options that are available. Sometimes we may need to talk with the genetics team, who may also offer to contact you for further discussion.

Does the Amniocentesis have any limitations?

- It does not test for spina bifida (a condition that affects the spine) or other physical differences.
- It cannot detect conditions such as autism or cerebral palsy.
- It can occasionally find an unexpected chromosome change which is unrelated to the ultrasound findings but that may have implications on the future health of the baby, or the health of other family members.

The FMU midwives are there to support you and to offer explanation of the results as needed. If further counselling by the specialist obstetric and genetic teams is required, this will also be arranged for you.

Are there any risks?

- Less than 1 in 200 women will suffer a miscarriage (loss of the baby) after having an amniocentesis.
- 1 in 20 women who have a late amniocentesis will go into premature (early) labour.
- We do not know why some women miscarry after having an amniocentesis but causes of miscarriage may include infection and suspected fetal abnormalities. Most miscarriages happen within 3 days of the procedure, but they can happen up to 2 weeks afterwards.
- It is not always possible to get a result from the first CVS or amniocentesis. Up to 6 in 100 women will need to be offered a second CVS or amniocentesis.

You may choose to discuss the procedure with a partner or close friend or family member before deciding whether you want to have the test or not. We are here to discuss this with you and can provide you with contacts of groups that can offer additional help and support.

Multiple pregnancies and invasive procedures

CVS and amniocentesis testing is possible in a twin or multiple pregnancy, but it is a much more complex procedure to perform in this situation. There are multiple factors to consider and therefore a consultation with a fetal medicine consultant and midwifery staff would be recommended prior to CVS or amniocentesis being offered to you.

Aftercare advice and ongoing care

After the test today

- You may wish to relax and avoid doing anything strenuous for the rest of the day.
- You may experience some mild cramping, like period type pain, that may last for 1 or 2 days.
- You can take paracetamol to ease any mild discomfort you experience. (Please follow the dosage instructions on the packet).
- Do not take ibuprofen for pain relief following a CVS or amniocentesis (we do not recommend taking Ibuprofen at all during pregnancy).
- You may want to try using a hot water bottle for pain relief (avoiding any areas where local anaesthetic was given).

When to seek medical advice

If you experience any of the following symptoms:

- Severe abdominal (tummy) pain
- Contraction-like abdominal pain
- Vaginal bleeding
- Vaginal fluid loss (if you think your waters might have broken/ fluid is leaking from your vagina)
- Signs of an infection:
  - Feeling hot/cold or shivery
  - Temperature above 38 degrees celsius or a temperature above 37.5 degrees celsius on 2 occasions more than 1 hour apart

Please use the contact numbers below and explain you have had a CVS/amniocentesis:

If you live out of area:

– Your local screening team:

– Your local Maternity Assessment Unit or Assessment Team:

If you live in the Oxford area:

– Gynaecology Department: **01865 222 001 (24hr)**

– Maternity Assessment Unit: **01865 220 221 (24hr)**

– Fetal Medicine Unit Midwives: **01865 221 716** (Monday to Friday: 8.30am to 5.30pm)

– Email: oxfordfetalmedicine@ouh.nhs.uk

We appreciate waiting for results is often challenging and stressful. If you have any questions or concerns following your procedure, please do not hesitate to contact us. We have included some websites and support groups that you may find helpful.

Support groups and useful websites

ARC: Antenatal results and choices

www.arc-uk.org/tests-explained

Soft UK: Support Organisation for Trisomies

www.soft.org.uk/understanding-chromosomes

FASP: Fetal abnormality screening programme

www.gov.uk/government/publications/cvs-and-amniocentesis-diagnostic-tests-description-in-brief

Further information

If you would like an interpreter, please speak to the department where you are being seen.

Please also tell them if you would like this information in another format, such as:

- Easy Read
- large print
- braille
- audio
- electronic
- another language.

We have tried to make the information in this leaflet meet your needs. If it does not meet your individual needs or situation, please speak to your healthcare team. They are happy to help.

We would like to thank the Oxfordshire Maternity Voices Partnership for their contribution in the development of this leaflet.

Author: Fetal Medicine Unit (FMU) Lead Midwife
Edited by: Quality Assurance and Improvement Midwife
August 2024
Review: August 2027
Oxford University Hospitals NHS Foundation Trust
www.ouh.nhs.uk/information

Leaflet reference number: OMI 106471