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CHORIONIC VILLOUS SAMPLING AND AMNIOCENTESIS

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# **CHORIONIC VILLOUS SAMPLING AND AMNIOCENTESIS**

## **WHAT ARE CVS AND AMNIOCENTESIS?**

CVS and amniocentesis are tests carried out during pregnancy most commonly to check your baby for disorders such as Down syndrome and, where appropriate, rarer specific inherited disorders. Amniocentesis is also occasionally done to test for other disorders in pregnancy such as fetal infection.

In CVS a small sample of the placenta (afterbirth) is taken for testing, whereas in amniocentesis a small amount of amniotic fluid - the water around your baby inside your uterus (womb) - is taken for testing.

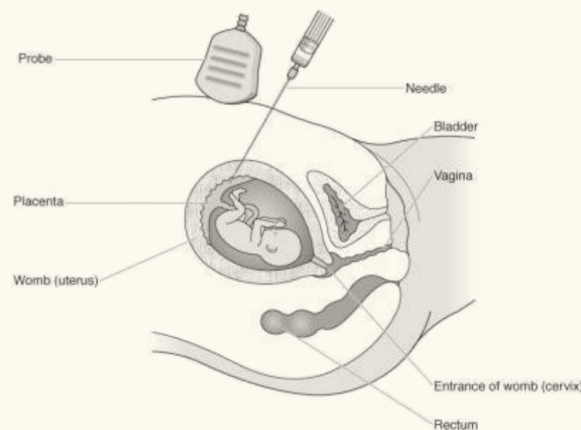
CVS is performed most commonly between 11 and 13 weeks. Amniocentesis is performed after 15 weeks.

## **HOW IS CVS PERFORMED?**

CVS involves taking a tiny amount of the developing placenta, where it is attached to the uterus. The placenta contains tissue that is genetically identical to your baby. CVS may be performed in two ways. Most CVS are performed through the abdomen, but CVS may also be performed through the cervix (neck of the womb). The Obstetrician doing the procedure will choose the method that he or she thinks is more appropriate for you. This will depend upon the position of the placenta. You will have a scan to check the positions of both your baby and placenta within the womb. You should be asked to sign a consent form before the procedure is carried out.

### **Through the Abdomen (Transabdominal)**

Your skin is then cleaned in the area where the needle will be inserted. The Obstetrician will use some local anaesthetic to numb the skin. Using an ultrasound probe to guide the direction, a fine needle is pushed through the abdomen and the wall of the womb into the placenta. A tiny amount of placental tissue is sucked up into a syringe. The needle is then taken out and your baby is observed for a short time on ultrasound.

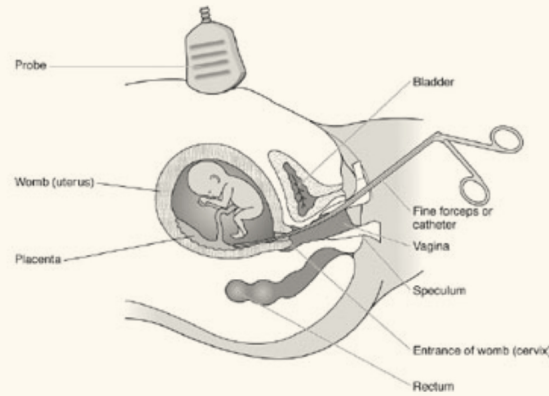


**Transabdominal CVS**



### Through the Cervix (Transcervical)

A speculum (a plastic or metal instrument used to separate the walls of the vagina) is inserted into your vagina to enable the doctor to see your cervix. The vagina and cervix are cleaned. Using ultrasound as a guide, fine forceps is passed through the cervix to the placenta. A tiny amount of placental tissue is removed. The baby is observed on ultrasound after this. After each procedure, the placental tissue, which contains some of your baby's cells, is sent to the laboratory for testing



Transcervical CVS

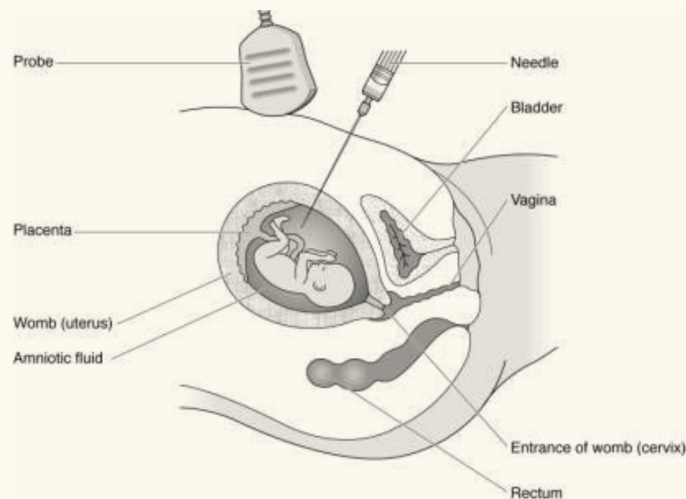
### IS CVS PAINFUL?

Most women say that having CVS, either transabdominal or transcervical, is uncomfortable rather than painful. Some women say the transcervical method is like having a cervical smear taken.

### HOW IS AMNIOCENTESIS PERFORMED?

Using an ultrasound probe for accurate guidance and to ensure a safe distance from your baby, a fine needle is pushed into your skin, through your abdomen and uterus. You should be asked to sign a consent form before the procedure is carried out.

A small sample of the fluid surrounding the baby is removed using a syringe. The needle is then taken out and your baby is observed on ultrasound.



Amniocentesis



The amniotic fluid, which contains some of the baby's cells, is sent to the laboratory for testing (see section on 'What are the laboratory tests?').

Occasionally, for fewer than 7 in every 100 women having amniocentesis, not enough fluid can be taken at the first attempt and the needle has to be re-inserted. This is usually due to the position of your baby. If a second attempt fails, you will be offered an appointment to have the procedure again on another day.

### **IS AMNIOCENTESIS PAINFUL?**

Women have said that having an amniocentesis gives a similar amount of discomfort as having a blood test taken. You will not usually need a local anaesthetic.

### **WHAT ARE THE RISKS OF CVS AND AMNIOCENTESIS?**

Every pregnancy carries a risk of miscarriage. As CVS and amniocentesis involve putting a needle through the wall of the womb, there is a small additional risk of a miscarriage due to injury or infection in the womb. The additional overall risk of miscarriage from amniocentesis is approximately 1:1000 (0.1%). The additional overall risk of miscarriage from CVS is 1:500 (0.5%).

Clinicians who do CVS or amniocentesis often, rather than occasionally, seem to be slightly better at getting enough placental tissue or amniotic fluid during the procedure, and may also have a lower risk of miscarriage.

You may notice some 'spotting' of blood and cramping for a few hours afterwards. This is normal and it is safe to take paracetamol. If you experience any unusual symptoms immediately after the test, such as feeling shivery (as if you have flu), fluid loss, bleeding or contractions, then you should seek advice immediately.

### **CAN I HAVE A CVS OR AMNIOCENTESIS IF I AM PREGNANT WITH TWINS?**

You can have a CVS or amniocentesis if you are pregnant with twins. It is very important to know which baby each sample of placenta or fluid has come from. If the results show one of the babies has a disorder, you and your medical team need to know which baby is affected. CVS or amniocentesis in multiple pregnancy is usually carried out by doctors with a high level of expertise in ultrasound scanning and this may mean you need to be referred to a specialist fetal medicine centre.

The doctor may need to insert the needle twice to get samples of placenta or fluid from each baby.

With CVS there is a small chance of getting two samples from the same baby, which could give misleading results.

The risk of miscarriage is slightly higher in amniocentesis with twins, and if this occurs it may lead to loss of both babies.



## **WHY MIGHT I CONSIDER CVS OR AMNIOCENTESIS?**

You might consider CVS or amniocentesis if you:

- Have had a high-risk screening test for Down syndrome
- Have had a previous pregnancy affected with a genetic disorder, or your partner have one or more relatives affected with a genetic disorder, which means you are at greater risk of having a child with a genetic disorder, such as cystic fibrosis, thalassaemia or sickle cell disease
- Have received a result from a scan, which shows certain abnormal ultrasound features, such as fluid collection at the back of the baby's neck (nuchal translucency), or a congenital heart defect, which indicates the baby may have a disorder such as Down syndrome. This may be suspected on a scan at 18–22 weeks
- Want to know for certain whether your baby has a genetic disorder or not.

Your age on its own is not a reason for having either test. You may decide not to have CVS or amniocentesis - it's your choice.

## **WHAT ARE THE LABORATORY TESTS?**

There are two types of laboratory tests that can be used to look at the baby's chromosomes. These are:

- A full karyotype which checks all the baby's chromosomes. Results from this test are usually ready in two weeks
- A rapid test which checks for specific chromosomes.
  - The disorders that can be detected by the rapid test include
    - Down syndrome (known as 'Trisomy 21' caused by an additional chromosome 21,
    - Edwards syndrome (known as 'Trisomy 18' caused by an additional chromosome 18,
    - Patau syndrome (known as 'Trisomy 13' caused by an additional chromosome 13)
    - And if requested, sex chromosome disorders.
  - Results from this test are usually ready within two working days. There may be a charge for this test.

In rare cases the short-term result may differ to the final result and further counselling regarding this discrepancy will be provided.



The sex of the baby can be confirmed by the chromosome result, however this information will only be provided at the woman's request.

Occasionally more complex genetic tests are performed. The unit where you had your CVS or amniocentesis will arrange with you how you will receive your results.

### **HOW RELIABLE ARE THE LABORATORY TESTS?**

For most women the laboratory test will give a definite 'yes' or 'no' answer. The result will let you know, one way or the other, whether the baby has the disorder the test was looking for.

With the full karyotype test there is small chance (about 1 in 100) that it will not give a clear result. This could be because it was not possible to analyse the sample in the laboratory (culture failure) or that the sample was analysed and the test gave an uncertain result.

If the test gives an uncertain result, you may be offered another diagnostic procedure, usually an amniocentesis.

### **WHAT WILL THE RESULTS TELL ME?**

The laboratory tests that look at the baby's chromosomes are able to detect a range of disorders, but not all.

- Most women who have CVS or amniocentesis will have a 'normal' result. In other words, their baby will be born without the disorder(s) that the test was looking for.
- Some women will be informed that the baby has the disorder the test was looking for.
- Very occasionally women have CVS or amniocentesis to detect Down syndrome and a different disorder is detected by the test.
- In a few cases women will have a 'normal' result and yet in spite of this they will have a baby born with another chromosomal or genetic condition. A normal result does not exclude every disorder.

### **WHAT ARE MY CHOICES IF THE RESULTS ARE ABNORMAL?**

If the results are abnormal, these will be discussed fully with you. For the majority of disorders, there is no treatment or cure. You will need to consider what is best for you and your baby.

If you decide to continue with your pregnancy, you will have appropriate support from healthcare professionals to help you and your family prepare for the birth and aftercare of your baby.

If you decide to end your pregnancy, you will be given information about what this involves. It will depend upon how many weeks pregnant you are when you make the decision.

### **MAKING A DECISION ABOUT CVS/AMNIOCENTESIS**

Making a decision about having a diagnostic procedure during pregnancy, such as CVS or amniocentesis, can be difficult. You may be making this decision alone or with your partner. It may



be helpful to talk through all the options with your doctor before you make your decision. You may also want to know more about what is involved in ending a pregnancy and how you may feel afterwards. Your obstetrician or midwife should discuss the following information with you:

- The disorders that can be detected by CVS or amniocentesis
- Which test (CVS or amniocentesis) would be recommended in your situation
- The types of laboratory tests available and what the results will tell you
- The reliability of the laboratory test(s)
- The risk of having an uncertain result and being offered a repeat procedure
- The risk of miscarriage from CVS (both transabdominal and transcervical) or amniocentesis
- How long the results will take
- How you will get the results
- Your options if the baby is found to have a genetic disorder.

### **WHAT IS THE DIFFERENCE BETWEEN CVS AND AMNIOCENTESIS?**

In making a decision about having a diagnostic test such as CVS or amniocentesis, it is important that you have enough time and that you feel supported in your final decision. You should be given time to talk through your options and be able to request any further information.

The final decision is yours. Only you can weigh up how much you want early information about a disorder against the slight risk that the procedure may lead to miscarrying a baby who may, or may not, have a genetic disorder.

### **CARE FOLLOWING CVS OR AMNIOCENTESIS**

Some women have mild cramping and a small amount of vaginal loss, either bleeding and/or amniotic fluid, after the procedure. This usually resolves without treatment over 24 to 48 hours. If it persists or is heavy, please seek medical advice. It is recommended women avoid strenuous activities and sexual intercourse for 48 hours following CVS or amniocentesis. Paracetamol is safe to take in pregnancy and may be required following the procedure.

### **IS THERE ANYTHING ELSE I SHOULD KNOW?**

- If your blood group is Rh (sometimes called rhesus) negative, you will be recommended an injection of anti-D immunoglobulin after the procedure to prevent you from developing antibodies against the baby's blood cells.
- If you are HIV positive and you decide to have CVS or amniocentesis, this might increase the risk of passing HIV on to your baby. You may be offered treatment with HAART (highly active antiretroviral therapy) if you are not already taking it. This reduces the risk of the



HIV virus infecting the baby. If you carry hepatitis B or hepatitis C viruses, there is in theory a possibility that CVS or amniocentesis might increase the risk that you pass this on to your baby. There is not enough information to be sure about whether this risk is real or not.

References:

RCOG Patient Information Committee. Information for you: chorionic villous sampling and amniocentesis. 2011 Mater Mothers Hospital Patient Information. CVS and amniocentesis. 2012

