

Important points

- Prenatal diagnostic tests give a reliable answer about certain problems with a baby
- **Chorionic Villus Sampling (CVS)** is a test in which a small sample of the *chorionic villus* or *chorion*, (the material lining the uterus that will develop into the placenta), is sampled and tested. This test is offered when the woman:
 - Is in her mid 30s or older since she has an increased risk of having a baby with a chromosome problem based on her age alone. This applies to any pregnancy, even if it is not the woman's first
 - Has had a screening test such as a first trimester test that indicates that the baby has an 'increased risk' of having chromosomal problems
 - Has had a child/previous pregnancy with a chromosomal problem, eg. Down syndrome
 - Is at risk for having a baby with a genetic condition because either she and/or her partner may pass on particular faulty gene(s) to the baby. Not all genetic conditions that could affect the baby, however, can be diagnosed during pregnancy
- CVS is done under ultrasound guidance; the sample is taken either through the abdomen or, less commonly, the vagina depending on the way the baby is lying and the preference of the doctor doing the test
- There is a very small chance that the result may be difficult to interpret and the test may need to be repeated
- CVS is usually done between the 11th and 13th weeks of pregnancy and should not be done before the 10th week
- The risk of miscarriage due to the test is less than 1%, or less than 1 chance out of every 100
- **Amniocentesis** is a test in which the fluid (*amniotic fluid*) that surrounds the baby in the womb (*uterus*) containing cells shed by the developing baby, is sampled and tested. This test is offered when the woman has any of the indications listed above for CVS:
 - Amniocentesis is done under ultrasound guidance
 - Is usually done between 15 and 19 weeks of pregnancy
 - The risk of miscarriage due to the test is less than 1%, or less than 1 chance out of every 100
- **Cordocentesis** (*fetal blood sampling*) may be recommended for the diagnosis of infection and some specific genetic conditions, where there is uncertainty about the results of the amniocentesis or if a rapid result is required
- The miscarriage risk **related to the test** is about 2% (1 in 50 pregnancies tested), but it may be higher if there are other problems associated with the pregnancy
- Where the baby has a problem identified with any prenatal diagnostic test genetic counselling is essential. Parents will be given all the information, they will be supported during and after their decision-making (see Genetics Fact Sheet 3)

Every woman hopes for a healthy baby. In some cases, a baby may have a serious either physical or intellectual problem.

There are a number of different prenatal (meaning before birth) tests and procedures available to assess the development of the baby. Each has advantages, disadvantages and limitations.

The importance of counselling in association with prenatal testing

Counselling before any prenatal test is done, whether it is a screening test or a diagnostic test, is strongly recommended. It provides an opportunity to discuss:

- How and when the tests are done
- The advantages and disadvantages of each test
- Any risks to the baby that may result from each test
- Any further testing which may be offered and what this further testing may mean for the parents and the baby

Counselling before a test is done will help the woman decide which test, if any, is best for the woman and the baby.

This Fact Sheet discusses CVS and Amniocentesis. Other related Genetics Fact Sheets are:

- Genetics Fact Sheet 17 – An overview
- Genetics Fact Sheet 17A – Ultrasound
- Genetics Fact Sheet 17B – First and second trimester screening

1. Chorionic villus sampling (CVS)

What is a CVS?

CVS stands for Chorionic Villus Sampling. The *chorionic villus* or *chorion* is the material lining the uterus that develops during the pregnancy into the placenta.

- The cells that make up the chorion are mostly the same as the cells of the baby, as both the placenta and the baby originate from the same fertilised cell
- A tiny sample of the chorion will therefore enable doctors to look closely at the baby's cells, in particular at the baby's chromosomes or, in some cases, particular genes

Why should parents' consider having a CVS in pregnancy?

The parents may consider having a CVS in the pregnancy for one of the following reasons, if the woman:

- Is in her mid 30s or older since she has an 'increased risk' of having a baby with a chromosome problem based on her age alone. This applies to any pregnancy, even if it is not the woman's first (see Genetics Fact Sheets 17 & 6)
- Has had a screening test such as a first trimester test (see Genetics Fact Sheet 17B) that indicates that the baby has an 'increased risk' of having chromosomal problems
- Has had a previous child/pregnancy with a chromosomal problem, eg. Down syndrome (see Genetics Fact Sheet 28)
- Is at risk for having a baby with a genetic condition because either she and/or her partner may pass on particular faulty gene(s) to the baby. Not all genetic conditions that could affect the baby, however, may be diagnosed during pregnancy

When will the CVS be done?

The test is usually done between the 11th and 13th week of pregnancy.

- As this is very early in pregnancy, it is important for the woman to see her doctor as soon as she realises she is pregnant
- In special circumstances, the CVS may be carried out later in the pregnancy
- A CVS should not be done before the 10th week of pregnancy

What happens during the CVS?

In order for the doctor to be able to get the best possible picture of the baby using an ultrasound scan, the uterus needs to be in the correct position. This can be helped by drinking one litre of non-fizzy fluid about one hour before the test and having a comfortably full bladder.

An ultrasound scan is done before and during the test (see Genetics Fact Sheet 17A). It enables the doctor to locate the position of the chorion to be sampled. It also allows the doctor to watch the sampling on the screen and makes sure the procedure is carried out safely.

The amount of chorion needed for the test is extremely small. Only about 1/1000 (or 0.1%) of the total chorion is sampled during this test.

There are two different methods used to sample the chorion, trans-abdominal and trans-vaginal. The selection of the most appropriate method will be discussed with the woman.

Trans-abdominal CVS

Trans-abdominal CVS (Figure 17C.1) is performed in a similar way to amniocentesis (see later).

The chorion sample is obtained by passing a fine needle through the abdominal wall and into the chorion using ultrasound for guidance.

A local anaesthetic is usually used on the mother's skin prior to the procedure to numb the area where the needle is passed.

Trans-vaginal CVS

Trans-vaginal CVS (Figure 17C.2) is very similar to having a Pap smear test and is done less commonly than abdominal CVS.

While the ultrasound is used on the abdomen to locate the baby in the uterus, a speculum is inserted into the vagina so that the cervix can be clearly seen by the doctor.

The cervix is swabbed to remove mucus that is usually present and a fine tube is passed through the cervix into the uterus. The image of the tube can be clearly seen in the ultrasound picture and thus it can be guided to where the placenta is forming. A small sample of the chorion is removed and sent to the laboratory for testing.

What will the CVS tell the parents about the baby?

It is now possible to diagnose a large number of genetic conditions during pregnancy. It is not, however, possible at present to test the baby's cells for all the possible genetic abnormalities that could affect him or her.

The woman's doctor or genetic counsellor will discuss with the parents the range of genetic conditions that can be found using the CVS test and what each of these might mean for the parents and the baby.

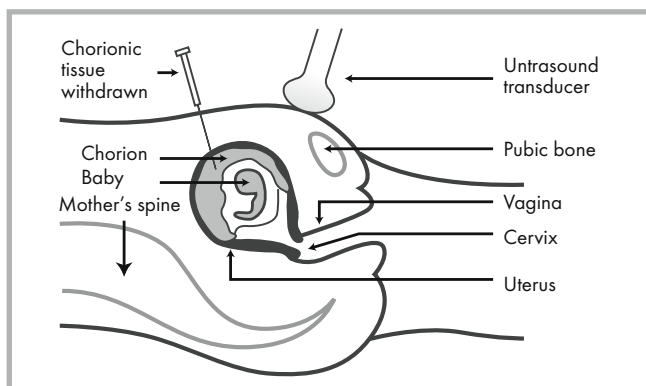


Figure 17C.1: Chorionic villus sampling in which about 10-20mg of chorionic villi are removed through the abdomen usually during the 11th to 12th week of pregnancy. Source: Vogel, F. & Motulsky, A.G. (1986). Human Genetics, 2nd end. Springer-Verlag, Berlin.

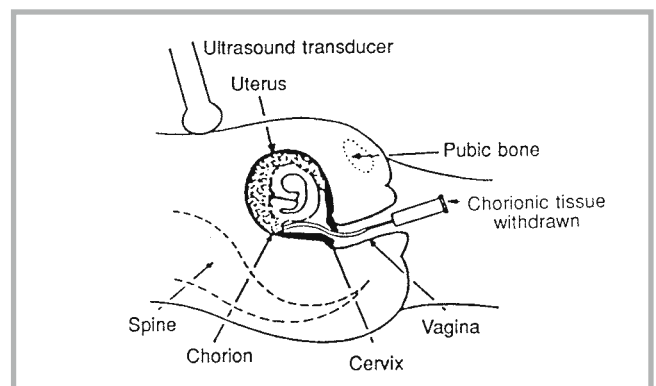


Figure 17C.2: Chorionic villus sampling in which about 10-20mg of chorionic villi are removed through the vagina and cervix usually during the 11th to 12th week of pregnancy. Source: Vogel, F. & Motulsky, A.G. (1986). Human Genetics, 2nd end. Springer-Verlag, Berlin.

It is important to remember that a normal test result cannot exclude every possible problem with the baby. Not all genetic conditions can be detected by these tests.

How reliable is the CVS test?

The CVS test is very accurate. It is, however, performed early in pregnancy and at this stage, the chorion (early placenta), is immature. Occasionally, the doctor is unable to obtain enough chorion on the first attempt. Samples of the chorion are examined immediately to make certain there is sufficient for the laboratory. If not, another sample may need to be taken at this time.

In rare cases, the laboratory may be unable to get a definite answer and then a second CVS may have to be done at a later time.

In other cases, there will be a need for an amniocentesis to be done to confirm a diagnosis. This means that a very small number of women (less than 1%) have both a CVS and an amniocentesis.

Does the CVS identify all genetic conditions?

NO. Although it is a diagnostic test, the CVS has some testing limitations.

- Most major chromosomal problems can be identified. However, not all subtle chromosome problems will be picked up
- Genetic or DNA testing (see Genetics Fact Sheet 21) for specific conditions can also be done in cases where this is indicated. Not all genetic conditions, however, can be detected using prenatal testing.

The woman's doctor or genetic counsellor will discuss the limitations of the CVS test with the parents prior to the woman undergoing the procedure.

When will the results be available?

The cells making up the chorionic villi usually have the same genetic information as the developing baby. They are allowed to grow and multiply in a laboratory until they are mature enough for testing.

It usually takes about 2-3 weeks for the result.

Does the CVS harm the mother or the baby?

There is a slight risk of miscarriage associated with having the CVS test. This risk is **in addition** to the 'background risk' of miscarriage. There is a risk in all pregnancies of miscarriage due to natural causes. It is important to discuss the background risk of miscarriage with the doctor.

The risk of miscarriage **due to having a CVS test** is less than 1% (occurring in less than 1 in 100 pregnancies tested with CVS). The specific risk figure depends on the experience of the doctor doing the test and the difficulty he or she has in getting the sample of chorion.

It is therefore important that a CVS test is only carried out by a doctor experienced in this technique and should be done after the 11th week of pregnancy.

Some women experience cramping after CVS. These symptoms are normal and should only last a day or so. The woman, however, should contact her doctor if she is concerned.

Parents are often worried that the tube or needle used to collect the sample of chorion will hurt the baby. In rare cases, problems with the normal development of the limbs has occurred when the testing has been done too early in pregnancy ie. earlier than 10 weeks. Therefore, as noted before, CVS should not be done before 10 weeks of pregnancy to minimise this rare problem.

All procedures are carried out in sterile conditions. The use of the ultrasound scanner during the procedure ensures that the tube or needle is outside the sac that the baby is in at all times.

What happens if the result of the CVS shows the baby has a problem?

In the few cases where the test does show that the baby has a problem, the parents will be given as much information as possible to enable them to make a choice about whether to continue the pregnancy. Some parents opt to continue their pregnancy and choose to use the information to be better prepared in pregnancy and after the baby is born.

If parents decide to terminate the pregnancy, details of the procedure required (including need for a general anaesthetic and time spent in hospital) will be discussed. The method of termination will vary depending on how advanced the pregnancy is and the policy of the particular hospital or clinic.

All aspects should be fully discussed with the woman's doctor or genetic counsellor. Parents will be offered support and information before, during and after making decisions of this kind.

Genetic counselling is recommended (see Genetics Fact Sheet 3).

What other tests are available later in pregnancy?

Even if the CVS test is normal, a specialised ultrasound at around 18 to 20 weeks of pregnancy is still recommended to check the baby's physical development (see Genetics Fact Sheet 17A).

2. Amniocentesis

What is an amniocentesis?

An amniocentesis is a procedure used to sample the fluid (*amniotic fluid*) that surrounds the baby in the womb (*uterus*). This fluid contains cells shed by the developing baby.

After a small sample of the fluid is collected, the baby's cells can be removed from the fluid and grown in the laboratory for testing.

Why should parents' consider having an amniocentesis in pregnancy?

The parents may consider having an amniocentesis in the pregnancy for one of the following reasons, if the woman:

- Is in her mid 30s or older since she has an increased risk of having a baby with a chromosome problem based on her age alone. This applies to any pregnancy, even if it is the woman's first (see Genetics Fact Sheets 17 & 6)
- Has had a screening test such as a first trimester test (see Genetics Fact Sheet 17B) that indicates that the baby has an 'increased risk' of having chromosomal problems

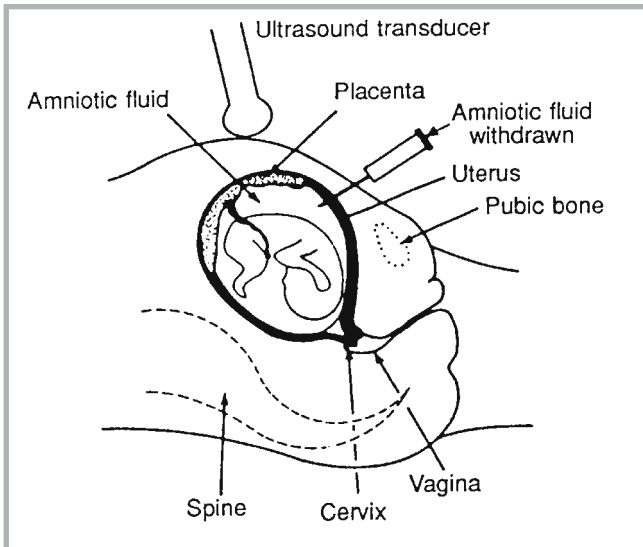


Figure 17C.3. Amniocentesis in which amniotic fluid (containing cells from the baby) is sampled during the 15th-19th week of pregnancy. Source: Vogel, F. & Motulsky, A.G. (1986). *Human Genetics*, 2nd edn. Springer-Verlag, Berlin.

- Has had a previous child/pregnancy with a chromosomal problem, eg. Down syndrome (see Genetics Fact Sheet 28)
- Is at risk for having a baby with a genetic condition because either she and/or her partner may pass on particular faulty gene(s) to the baby. Not all genetic conditions that could affect the baby, however, may be diagnosed during pregnancy

When will the amniocentesis be done?

The test is usually done between the 15th and 19th week of pregnancy (ie. between 15 and 19 weeks). At this time, there is enough amniotic fluid to sample.

What happens during the amniocentesis?

An ultrasound scan is done before, and during the test. The doctor cleans the woman's abdomen with antiseptic fluid and puts a very thin needle into the uterus through the abdomen. The ultrasound is used to check the position of the needle, the position of the baby and determine if there is a suitable amount of amniotic fluid.

A small sample of the amniotic fluid (about 15mls) is taken up through the needle. The woman may feel some slight discomfort during the test (Figure 17C.3).

The amniotic fluid contains cells shed by the developing baby. These cells are allowed to grow and multiply in a laboratory until they are mature enough to test.

Occasionally, the doctor is unable to obtain enough amniotic fluid on the first attempt. If this happens, another sample may need to be taken at this time.

What will the amniocentesis tell the parents about the baby?

While it is now possible to diagnose a large number of genetic conditions prenatally, it is not feasible to test the cells for all the possible abnormalities that could affect the baby.

The woman's doctor or genetic counsellor will discuss with the parents the range of genetic conditions that can be identified using amniocentesis and what each of these might mean for the parents and the baby.

It is important to remember that a normal test result does not exclude every possible problem with the baby. Not all genetic conditions can be detected by this test.

How reliable is an amniocentesis?

Chromosome studies on amniotic fluid following amniocentesis are very accurate.

Very occasionally, more than one amniocentesis is necessary to obtain a diagnosis. This happens when the doctor is unable to obtain enough fluid on the first attempt, or when the laboratory is unable to make an analysis of the fluid.

Does the amniocentesis identify all genetic conditions?

No. Although it is a diagnostic test, the amniocentesis has some testing limitations. Most major chromosomal problems can be identified. Not every subtle chromosome abnormality, however, will be picked up.

Genetic testing for specific genetic conditions can also be done in some cases where this is indicated, although, not all genetic conditions can be detected using prenatal testing.

The woman's doctor or genetic counsellor will discuss the limitations of the amniocentesis test with the woman prior to the test being done.

When will the test results be available?

The amniotic fluid is sent to the laboratory and the baby's cells are removed.

They are allowed to grow and multiply until they are mature enough for testing.

The results are usually available in about two to three weeks.

Does the amniocentesis harm the mother or the baby?

There is a slight risk of miscarriage associated with having the amniocentesis test. This risk is in addition to the 'background risk' of miscarriage. There is a risk of miscarriage in every pregnancy due to natural causes. This risk is less at this stage of pregnancy than earlier on. It is important to discuss the background risk of miscarriage with the doctor.

The risk of miscarriage **due to having an amniocentesis** test is less than 1% (occurring in less than 1 in 100 pregnancies tested with amniocentesis). The specific risk figure depends on the experience of the doctor doing the test and the difficulty he or she has in getting the sample.

It is therefore important that an amniocentesis is only carried out by a doctor experienced in this technique.

It is not uncommon to have cramping on the day of the test. A small number of women may experience bleeding and some leaking of amniotic fluid. The woman's doctor should be contacted if this occurs.

Parents are often concerned that the needle will hurt the baby, but the use of the ultrasound during the procedure enables the needle to be inserted in a position away from the baby.

All procedures are carried out in sterile conditions.

What happens if the result of the amniocentesis shows the baby has a problem?

Most often, the baby will not have the particular genetic condition for which it was tested. In those few cases where the test does show that the baby has a problem, the parents will be given as much information as possible to enable them to make a decision about whether to continue the pregnancy.

If parents decide to terminate the pregnancy, details of the procedure required (including need for a general anaesthetic and time spent in hospital) will be discussed. The method of termination will vary depending on how advanced the pregnancy is and the policy of the particular hospital or clinic.

All aspects should be fully discussed with the woman's doctor or genetic counsellor. Parents will be offered support and information before, during and after making decisions of this kind.

Genetic counselling is recommended (see Genetics Fact Sheet 3).

What other tests are available later in pregnancy?

Even if the amniocentesis shows that the baby does not have certain problems, a detailed ultrasound is usually offered between

18 and 20 weeks of pregnancy to check the baby's physical development (see Genetics Fact Sheet 17A).

3. Cordocentesis

Where there is uncertainty about the results of the amniocentesis, or a rapid result is required, **cordocentesis** (CORD-O-CEN-TEE-SIS, also called *fetal blood sampling*) may be recommended. Cordocentesis is also a prenatal test used for the diagnosis of infection and some specific genetic conditions.

A sample of the baby's blood is taken by passing a fine needle into the umbilical cord using ultrasound guidance.

There is a miscarriage risk **related to the test** of about 2% (1 in 50 pregnancies tested), but it may be higher if there are other problems associated with the pregnancy.

This test is not performed very frequently and the woman's doctor will discuss it with the parents if he or she feels it is indicated. The risk of miscarriage associated with the test is an important factor to consider when deciding whether to have this test or not.

Other Genetics Fact Sheets referred to in this Fact Sheet: 3, 6, 17, 17A, 17B, 21, 28

Information in this Fact Sheet is sourced from:

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