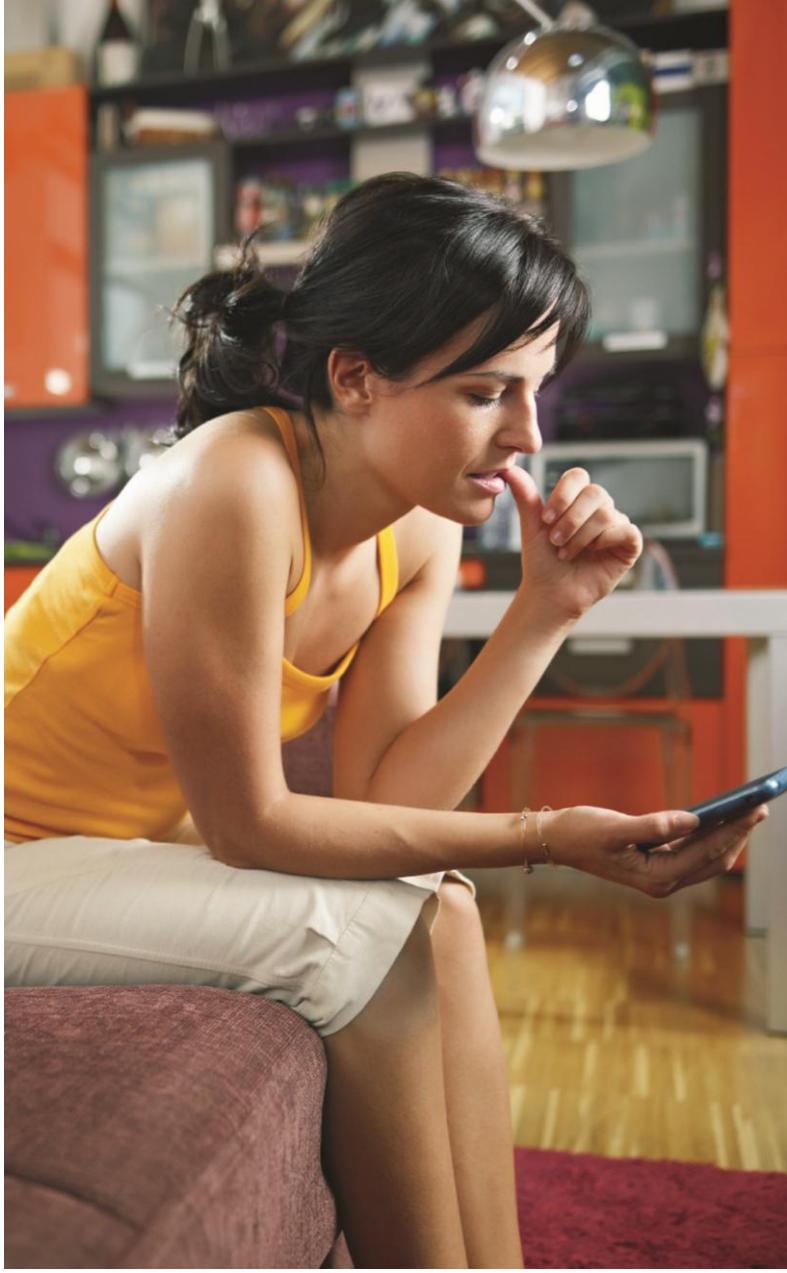
Amniocentesis and CVS



Fertility Facts

Amniocentesis and chorionic villus sampling (CVS) are techniques that can identify chromosome abnormalities in the fetus in early pregnancy – around 12 weeks for CVS and 16 weeks for amniocentesis. A normal result is reassuring if you have a higher risk of abnormality, while an abnormal result gives you the opportunity to decide whether to terminate the pregnancy or prepare for a child with special needs.

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Chromosome abnormalities

These tests are usually done to detect chromosome abnormalities in the fetus, and sometimes to identify a specific genetic disorder that the mother or father is known or is suspected to carry, such as Cystic Fibrosis.

Chromosomes carry the genetic information in the cells of the body. The most common chromosome abnormalities are an extra chromosome (such as an extra chromosome 21 in Down Syndrome), a missing chromosome (such as Turner's Sydrome when a girl has only one copy of the X chromosome) or part of a chromosome that is attached to another in what is called a translocation.

In amniocentesis the cells for analysis come from cells shed from the fetus into the surrounding amniotic fluid; in CVS the cells are taken from the chorionic villi – which are small parts of the placenta.

Chance of chromosome abnormalities

If you have had a baby with Down Syndrome, the risk of having another affected infant depends on the chromosome pattern in the previously affected infant. If this was the most common form of Down Syndrome (standard trisomy 21), then the risk is 1% plus your age related risk (see below).

If Down Syndrome has occurred in other family members, the risks of recurrence may be very low and a blood test should be performed on the affected relative (this will often have been done already) before amniocentesis. If the affected family member had standard trisomy 21, the risk to your child is very low and amniocentesis may not be indicated.

For a healthy couple with no previous history of this abnormality, the risk of giving birth to an infant with a chromosome abnormality is related to the age of the mother. The father's age has only a very small effect. The risks of having an affected pregnancy at 16 weeks are approximately as follows:

Woman's age	20	30	35	37	40	45
Risk - 1 in	1500	800	170	120	60	25

It is important to remember these tests will not diagnose many common abnormalities. About 1 in 50 births in women

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of any age is associated with a congenital abnormality of some sort.

The risk estimate for a particular pregnancy at 12 weeks can be refined by using a combination of blood tests and an ultrasound scan that looks at the skin around the fetus' neck – known as nuchal transparency. This approach does not make a diagnosis, but gives a better estimate of the risks of an abnormality for younger women. For instance, the blood test and nuchal scan may indicate a higher risk than predicted from the woman's age.

WHAT THE TESTS INVOLVE

CVS Local anaesthetic is injected into the skin of the abdominal wall. An ultrasound machine is used to visualise the fetus and the placenta. A fine needle is inserted into the wall of the uterus, and then into the edge of the placenta. When the needle is in place, a syringe is attached and a sample of cells is removed by gentle suction. A full bladder is not necessary for this procedure. Sometimes more than one attempt is needed to obtain suitable tissue. Occasionally, the CVS is unsuccessful. If this occurs, a further attempt might be made a week or so later or amniocentesis might be recommended.

Amniocentesis Amniocentesis is usually performed around the 15th week of pregnancy, but if there is not enough fluid the procedure may need to be deferred for a week or two to make it easier and safer. Otherwise it is very much like CVS, except the needle drains a little amniotic fluid.

No special preparation is needed for either test. A little discomfort after the test is normal; it is a good idea to have you partner or a friend drive you home. Partners and support people are very welcome to be present during the procedure, and it is useful for them to share the discussion beforehand. They will be able to see the ultrasound scan.

Risks of Amniocentesis and CVS

The chance of miscarriage is approximately 0.5% to 1% on top of the usual risk of miscarriage in any pregnancy at 12 weeks. This risk is similar for CVS and amniocentesis. If miscarriage does occur, it normally will happen within a week or two of the procedure.

Because the needle is kept well away from the fetus, risk of fetal damage from the procedures is very small. If CVS is done before 9 ½ weeks, there is a chance of abnormalities in the development of the fetal limbs due to tiny blood clots becoming stuck in a developing limb. Because of this a CVS will not be done before 11 weeks of pregnancy as determined by ultrasound dating.

After CVS and Amniocentesis

If your blood group is Rhesus negative, you will be given an injection to stop you forming antibodies against your baby's blood cells. This could only happen if your baby is Rhesus positive and some fetal cells are spilt into your circulation. This is very unlikely, but it is better to take this precaution.

A quiet day is all we suggest. If there is vaginal blood or fluid loss, you should report it to your LMC right away, and rest in bed until the problem settles. Some soreness around the needle site occurs occasionally and cramps are common.

When are the results known?

Your doctor will be sent the results of the test, and will then tell you.

The complete chromosome studies take about two weeks or sometimes a little longer. Very occasionally the cells from CVS or amniocentesis fail to grow and a chromosome result is not possible. With CVS there is a 1-2% chance of a confusing result because both fetal and maternal cells grow.

If one of these things happens, we can usually tell you within two weeks of the test. If this happened after CVS, your doctor would usually recommend an amniocentesis as a follow up. If it happened after an amniocentesis, then we would offer to repeat the amniocentesis. Having problems with getting a result does not mean you have a higher chance of chromosome abnormalities.

The test result will include the baby's gender. If you do not want to know this, it is important to let your doctor know.

Rapid results, which look at chromosome 21, 18, 13, X and Y, are available in three working days. These are the chromosomes most commonly found to be abnormal. Unless the screening test result is very high risk, then you will normally have to pay extra for this test (around $300-$400).

Cost of the tests

The standard tests are free to those with a higher risk of abnormalities.