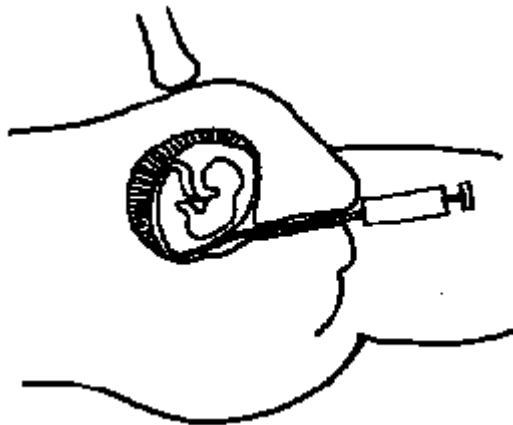


# Prenatal-diagnosis

Everyone hopes for a happy, healthy baby, but unfortunately some couples have a greater than average chance of having a baby with serious physical or mental disabilities. Your doctor has suggested you consider the special tests that are available while you are pregnant, to see if your baby has such a problem.

## Chorionic villous sampling

Otherwise known as CVS,



this test involves the collection of a tiny piece of placental tissue, or chorion, at about 10 weeks. Cells are grown from this over the next 2-3 weeks and analysed to see if your baby's chromosomes are normal. Chromosomes are the parts of a cell that determine whether we are male or female, as well as our other characteristics. Usually a cell will have 46 chromosomes, of which two are the sex chromosomes. Sometimes a baby will have 47, called Trisomy, and the risk of this is greater if the mother is over 35 years.

Trisomy 21, or Down's Syndrome, is the most common and occurs when the baby has an extra chromosome 21. Other inherited diseases can sometimes be detected by analysing the

genes, or parts of chromosomes. Gene structure is so complex that a routine screening for every abnormality is not possible, so be sure to discuss with your doctor any diseases in your families that might need to be tested for.

On rare occasions the test may need to be repeated, and less than 1% of women will need their test confirmed by amniocentesis. It is important to remember not all birth defects can be

detected genetically, and that no test is 100% accurate. However, CVS is generally considered to be both safe and effective.

It is usually performed around 10 weeks when the placenta can be identified by ultrasound (U/S). The sample may be collected through the mother's abdomen, similarly to amniocentesis, or through the vagina; dependant on the size and site of the placenta. Local anaesthetic is used to numb the area, and a thin metal tube (needle or cannula) is guided by U/S into the uterus to the edge of the placenta. A very fine needle is then passed through this tube into the placenta and a syringe is used to draw up a tiny fragment of tissue.

When performed by a

specialist the risk of subsequent miscarriage is about 1-2%.

## Amniocentesis

An amniocentesis obtains some of the amniotic fluid which surrounds the baby and is usually performed at 16-18 weeks. The fluid is rich in shed fetal cells which can be cultured for similar genetic testing to CVS. The fluid is also rich in chemicals excreted by the baby, which can be tested in later pregnancy for such things as maturity of the baby's lungs.

An U/S first examines the baby for any physical problems, and a safe spot is chosen for the insertion of the guiding needle. Usually the skin is made numb with local anaesthetic, but some women feel a short sharp jab as the needle enters the muscle of the womb. A finer needle is then inserted

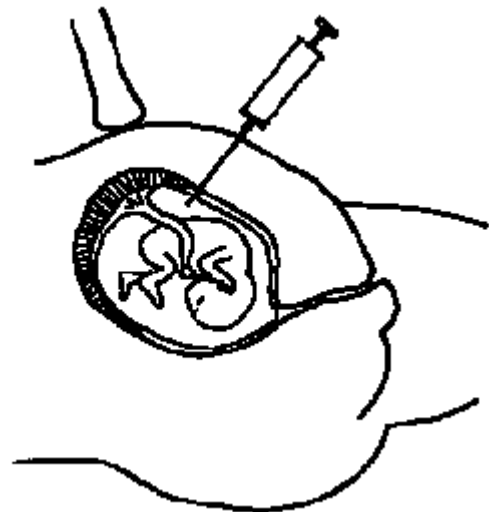
introducing infection, or of the membranes leaking, or bleeding. Advice should be sought if these complications occur, or if there is pain not controllable by paracetamol.

Performed by a specialist, the miscarriage rate is about 0.5-1%.

## Options

Ideally, both parents will have thought about the choices they might make before they have the tests performed. If the test does detect an abnormality, it is important that both parents understand the outcome, and discuss their emotional responses as well as any treatment options.

Termination of pregnancy can be performed, but many parents choose to proceed with the pregnancy forewarned that their baby will require more care and attention over the years.



through the guide and about 15mls of fluid drawn off. Most women comment that the procedure was not as bad as they thought it would be.

Every effort is made to reduce complications, but there is a small risk of

The further advice of your doctor, with genetic and emotional counselling, is recommended.