Liverpool Women's NHS
NHS Foundation Trust

Specialist Screening

Serum Screening Tests

Most babies develop normally, but unfortunately about 2% have a significant abnormality. Screening tests inform you of the chance that your baby could be born with a problem. It is important that you have decided wether you would want further tests such as amniocentesis if the screening test is positive. However, agreeing to a screening test does not mean you have decided to have a termination if a problem is found. Some couples simply prefer to know in order to be prepared, whilst others may choose to terminate their pregnancy. If a problem is found, you will be offered additional support and counselling to be aware of all your options.

- Neural Tube Defects e.g. Spina Bifida
  An opening anywhere along the length of the baby's spine can result in paralysis of the lower limbs or a collection of water on the brain (hydrocephalus).
  An important warning sign is raised AFP (alphafetoprotein), measured in your blood.
  The best diagnostic test is detailed ultrasound scan.

- Down's Syndrome
  Babies with this condition have an extra chromosome 21, and are at increased risk of heart and bowel problems and learning disabilities. The overall risk increases for older mothers. The 'double blood test' - measures 'serum markers' and using other factors it can be worked out if your chance of having a
  baby with Down's Syndrome is increased.

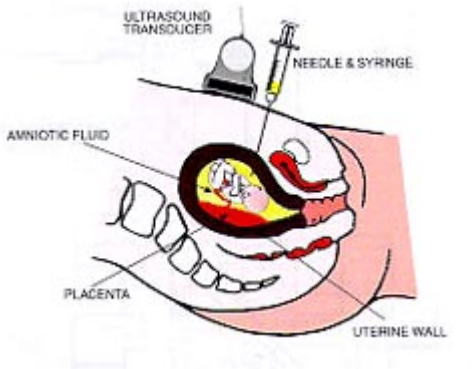
Amniocentesis

What is an Amniocentesis ?

This is a simple way of finding out if a baby has a chromosomal disorder. Amniocentesis involves the removal of fluid (liquor) from around the baby under 'direct ultrasound guidance', and it can be performed as early as fourteen weeks. The liquor contains hundreds of living cells that come from the baby's skin.

How is the test done?

The doctor does an ultrasound scan first to check that the baby's size is measured to ensure that you are at the correct dates before doing the test. Following this, the skin is cleansed with an antiseptic sterile wipe, then, a fine sampling needle is passed into the skin of the tummy, which then enters the womb. The doctor knows exactly where to put the needle in because he can see an image on the ultrasound screen. At least 20mls of clear fluid are removed with a syringe. This is then put in a clear plastic container, which is sent to the laboratory for testing.



Does having an Amniocentesis hurt?

Everybody's pain threshold is different. In our experience of performing the test, it would be true to say that most women have only found the test uncomfortable. Some say it is very similar to having an injection or blood removed from the arm.

After the test

We always advice ladies to take a seat in reception area for a short while before going home. You are advised to take things easy for the next couple of days. You don't have to stay in bed, but do try to avoid household chores or lifting of heavy objects.

When to call the Doctor

Within the next 32-48 hours, you may experience lower abdominal discomfort, similar to period pains, this is not unusual. Paracetmol can be taken to relieve this, and in most cases is effective. If, however your pains become worse, you start to bleed, experience a water type loss or develop a temperature, please contact your GP.

Possible complications

There is a 0.5%-1% chance of a miscarriage happening to you and this is on top of the average risk of miscarriage during pregnancy. Miscarriages which occur within a few days of amniocentesis are probably related to the test. For complications that happen much much later however, it would be difficult to say if these were test related.

Is the test reliable ?

Amniocentesis is 99.9% accurate for diagnosing chromosomal / genetic disorders, but like all tests it has pitfalls. No test is perfect and occasionally the test fails. This occurs in 1% of cases and is due to cells not growing (culture failure), or when the cells grow in a peculiar fashion which gives them more than one result (mosaicism). If this happens, we would let you know and suggest another test.

Results

Results usually take 3-5 weeks because, although the test is very sophisticated, it is still complex.

At the laboratory the cells are separated from the fluid and then placed in a culture dish. The cells are "incubated" and checked every day. At a specific time of development the pattern and number of chromosomes are carefully examined and a diagnosis is made. The process cannot be rushed.

Chorionic Villus Sampling

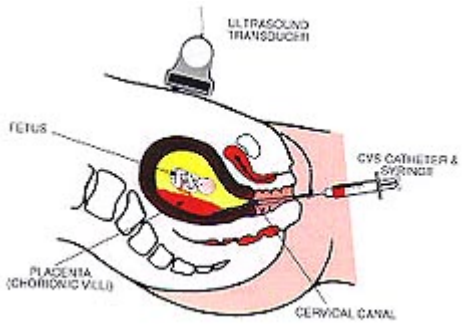
What is Chorionic Villus Sampling?

Chorionic villi are small "finger-like" projections from which the placenta or after-birth is formed, and it is this tissue which is tested to see if a baby has a chromosomal or inherited disorder. It is a much newer type of prenatal test and is performed from ten and a half weeks.

The Transcervical Method - How is it done?

It is very similar to having a cervical smear and isn't too uncomfortable. First, an ultrasound scan is performed to locate the position of your placenta. (You will need to have a full bladder for the scan.) After this you will be asked to lie down flat on your back with your knees apart. To help reduce the risk of infection, the doctor will cleanse inside and around the vaginal area with an antiseptic solution. Then a speculum (a small metal object which holds the vagina walls apart), is inserted into the vagina so that the neck of the womb can be clearly seen.

Next, to take a specimen of Chorionic villi, a fine plastic tube is placed into the vagina and cervix. On reaching the placenta, small fragments of villi are gently sucked out with a syringe. The plastic tube is withdrawn and the sample is inspected under a microscope. If there are enough villi, then the test is complete and the sample is quickly sent to the laboratory. If the doctor feels that there is insufficient tissue, then the test is repeated and more tissue collected.



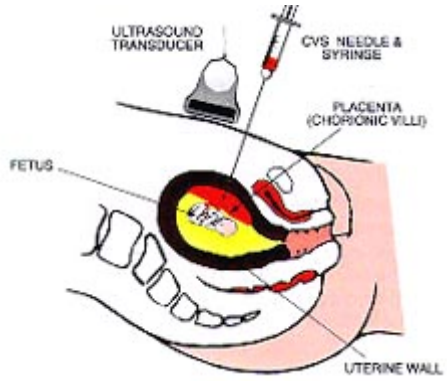
The Transabdominal Method

This test if very similar to having an amniocentesis performed and you don't necessarily need to have a full bladder. Whilst lying on your back, the doctor performs an ultrasound scan first see where your placenta is situated. Next, he cleans your tummy with an antiseptic solution, and then puts a small amount of lignocaine.

After a few minutes, your skin will become 'numb' and then the test can begin. With one hand, the doctor puts the sampling needle through your tummy and into the placenta, and with the other he holds the ultrasound probe (under direct guidance) which enables him to see where the needle is.

Once the needle tip is in place, he takes a sample of Chorionic villi, by passing a fine needle down inside the first. Gentle suction and movement is applied and you shouldn't feel any discomfort from this. The inner needle is removed and he then checks the sample under a microscope. If the sample is small, then he just repeats the test again, by passing down the inner needle again.

This time if there is sufficient tissue, the needle in your tummy is removed. To give you as much reassurance as possible, a quick scan is performed to show you your baby's heartbeat.



Does C.V.S. hurt?

Everybody's pain threshold is different. In our experience of performing the test, it would be true to say that the test is slightly uncomfortable, but not painful.

After the test

We always advice ladies to take a seat in reception area for a short while before going home. You are advised to take things easy for the next couple of days. You don't have to stay in bed, but do try to avoid household chores or lifting of heavy objects.

When to call the Doctor

Within the next 32-48 hours, you may experience lower abdominal discomfort, similar to period pains, this is not unusual. Paracetmol can be taken to relieve this, and in most cases is effective. If, however your pains become worse, you start to bleed, experience a water type loss or develop a temperature, please contact your GP.

Possible complications

There is a 2% chance of a miscarriage happening to you and this is on top of the average risk of miscarriage during pregnancy. Miscarriages which occur within a few days of Chorionic Villis Sampling are probably related to the test. For complications that happen much much later however, it would be difficult to say if these were test related.

Is the test reliable?

Chorionic Villus Sampling 99.8% accurate for diagnosing chromosomal / genetic disorders, but like all tests it has pitfalls. No test is perfect and occassionally the test fails. This occurs in 1% of cases and is due to cells not growing (culture failure), or when the cells grow in a peculiar fashion which gives them more than one result (mosaicism). If this happens, we would let you know and suggest another test.

Results

Results usually take about one week. This initial result is known as the "direct preparation" and following this, about two weeks later, is a second report called the "cultured preparation"