

Medical and Screening Tests in Early Pregnancy



This leaflet explains what screening tests are available to pregnant women in the first and second trimester of pregnancy. These include the booking blood tests, screening and diagnostic tests for Down syndrome and the 18-20 week anatomy scan.

There is a lot of information for you to absorb during your first visit to AOC and we hope that this leaflet will enable you to understand more about the tests that you may be offered.

BOOKING TESTS

At your first visit to AOC we will arrange for you to have a set of booking bloods taken. Ideally this is done during the first twelve weeks of your pregnancy. These tests are all done on a single blood sample and include:

A Full Blood Count

This is to check for anaemia.

Blood Group And Antibody Screen

This is to check your blood group (A, B, O or AB) and rhesus group (positive or negative). Blood group antibodies are also tested for. These might make it difficult to cross match blood if you ever needed a blood transfusion or cause anaemia in a developing baby.

Rubella Serology

This is to check that you are immune to rubella (also called german measles). Most women have been vaccinated against rubella in their early teens but a few women will have little or no immunity.

Hepatitis B

This checks for infection or immunity to hepatitis B. Pregnant Hepatitis B carriers can pass hepatitis onto their baby at delivery. There are very effective treatments for baby after it is born to prevent this if carriers are detected antenatally.



VDRL

This screens for syphilis. This is now a rare disease but if detected, treatment in pregnancy can prevent baby being infected.

HIV Screening

HIV (the virus that causes AIDS) is still rare in pregnant women but carriers can infect their unborn child. Treatments given during pregnancy are very effective in reducing the risk of fetal infection in women found to be carriers for HIV.

MSU (Mid-Stream Urine)

A urine sample can check for unexpected urine infection or the presence of bacteria in your urine that increases your risk of kidney infections later in pregnancy.

SECOND TRIMESTER TESTS

Vaginal Swabs

Infections such as Chlamydia, which often cause few or no symptoms, can be checked for in pregnancy though we usually wait until between 12 and 16 weeks to screen for this.

Anatomy Scan

At 18 to 20 weeks of pregnancy most women have an anatomy scan. This is a detailed scan to check baby's brain, heart, spine and other important organs. The great majority of babies will be normal and couples find an anatomy scan very reassuring. Occasionally, an abnormality is detected and this can have important implications for your baby's care. For example, it may be necessary to arrange for your baby to have a surgical procedure soon after birth. Some couples are very anxious that a severe abnormality will be found and a termination suggested. This is a very rare event and in most cases finding an abnormality can help paediatricians plan any treatment baby might need after it's born. It is also important to realize that not all problems and abnormalities will be detected.

It is often possible to tell if baby is a boy or girl but this is not the primary purpose of the scan. At best, the sonographer can only give you a strong idea about baby's gender. You may be offered a 4D ultrasound which produces very detailed pictures of baby but there is no particular medical reason to have a 4D scan.

Polycose Test

This screening test is done at about 26 to 28 weeks of pregnancy to check if you are at an increased risk of developing gestational diabetes (diabetes in pregnancy). You will be given a glucose drink and an hour later some blood is taken. If your blood sugar is unexpectedly high you may need a further more detailed test called a Glucose Tolerance Test (GTT). For this test you will need to miss breakfast and have blood taken before having a glucose drink and then more blood taken at one and two hours later.

Gestational diabetes is associated with high blood sugars in pregnancy. Exposure to high sugar levels can result in baby becoming excessively large. Women who develop gestational diabetes are also at greater risk of developing diabetes in later life.

Blood Group Antibody Screen, Full Blood Count And Ferritin Levels

These are also checked at the same time as your polycose test to look for signs of anaemia and the development of antibodies to baby's blood group.



An anatomy scan at 20 weeks of pregnancy

DOWN SYNDROME SCREENING

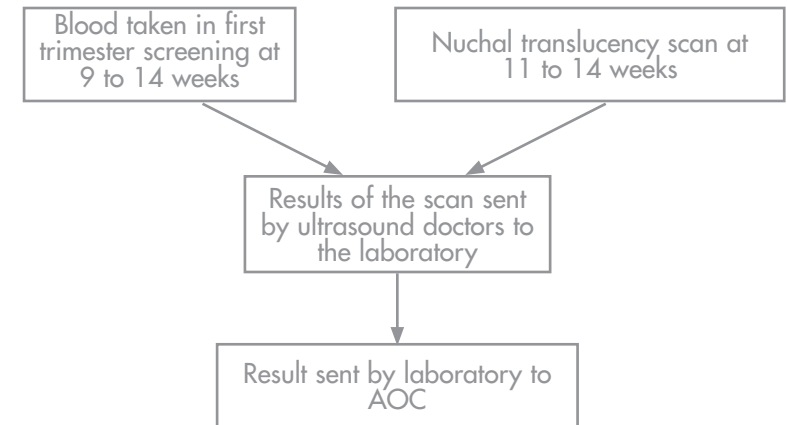
What Is Down syndrome?

A normal child has 23 pairs of chromosomes. A child with Down syndrome has an extra twenty first chromosome (the condition is sometimes called Trisomy 21). This is the result of an abnormality arising at the moment of conception. Why it occurs is unknown. It becomes commoner with increasing maternal age. For example at 30 years of age a woman has a one in 350 chance of carrying a baby with Down syndrome when she has screening tests at 11 to 14 weeks. By 40 years of age this rises to one in 100. Down syndrome is equally common in all ethnic groups. Children with Down syndrome have a varying degree of intellectual disability. Some have other serious medical problems but many children with Down syndrome will lead fulfilling lives. For more information look at the websites listed at the end of this leaflet.

First Trimester Combined Screening

Pre-natal screening for Down syndrome consists of a blood test taken at between 9 and 14 weeks of pregnancy (ideally between 10 and 12 weeks) and an ultrasound scan (called a nuchal translucency scan) performed at 11 to 14 weeks of pregnancy. A separate blood test at 14 to 20 weeks of pregnancy can also be offered to women who did not have any screening tests earlier in their pregnancy.

These screening tests start with a risk based on your age. They then calculate your personal risk based on the levels of two substances in your blood and an ultrasound measurement taken from the back of baby's neck. The two substances in your blood (called pregnancy-associated plasma protein (PAPP-A) and beta-human chorionic gonadotrophin (β -hCG)) tend to occur at different levels in pregnancies affected by Down syndrome. The thickness of the skin at the back of baby's neck (the nuchal translucency measurement) tends to be increased in babies that have Down syndrome. By putting all these measurements together your personal risk of carrying a baby affected by Down syndrome is calculated.



Together these tests are referred to as *first trimester combined screening*.

Both these tests are completely safe and will provide you with a risk measurement. They are screening tests – they will provide an accurate assessment of your risk but won't give you a definite yes or no answer. This risk is given to you as a number - for example 1 in 100 or 1 in 500.

Used together as a screening test about 80 to 90 per cent of babies affected by Down syndrome will be picked up. It is impossible for all babies affected by Down syndrome to be picked up because even for women at very low risk, an occasional baby will have Down syndrome (for example, for a risk of one in one thousand, every thousandth baby will have Down syndrome).

One purpose of the screening tests is to help many women avoid amniocentesis. The risk of having a baby with Down syndrome increases with age but most older women who might have considered an amniocentesis because of their age will still have a reassuring result from these tests.

Overall about 3% to 5% of women will opt to have an amniocentesis or CVS after the first trimester combined screening test. This proportion will be slightly higher in older women because their initial pre-test risk based on their age alone will be higher.

AMNIOCENTESIS AND CHORIONIC VILLUS SAMPLING



A nuchal translucency scan

Second Trimester Serum Screening

Women booking for antenatal care after 14 weeks of pregnancy (too late for a nuchal translucency scan) can still have a screening blood test (this is called *second trimester serum screening*). This can be done at up to 20 weeks of pregnancy (ideally between 14 and 18 weeks). This test also takes your risk based on your age alone and calculates a personal risk based on the levels of the different substances checked in your blood.

This later test is not quite as reliable as the combined first trimester test. There is no need to have this later test done if you have already had the earlier combined test.

Women who have a higher than expected risk of carrying a baby with Down syndrome may wish to have a diagnostic test to confirm or rule out whether they do definitely have a baby with Down syndrome. For some couples making this decision is easy but for most it is a big step and we are very happy to talk about how these tests are undertaken and help you make a decision that is right for you.

Amniocentesis and chorionic villus sampling (CVS) are the prenatal diagnostic procedures used to detect whether a baby has a chromosomal abnormality such as Down syndrome. They can also detect the presence of some inherited genetic disorders for couples known to have an increased risk of these conditions. Amniocentesis and CVS enable the testing of fetal chromosomes found in fetal and placental cells.

Amniocentesis

Amniocentesis is usually performed from 15 weeks of pregnancy. During the procedure a fine needle and syringe is used to draw off a small amount of amniotic fluid (about 10-15 millilitres, equal to about two teaspoons) from inside the amniotic sac. Amniotic fluid is the fluid that surrounds baby. It is made by the fetal kidneys and contains cells that have been shed from baby's skin, bladder and lungs. At the time of amniocentesis an ultrasound is performed to help the doctor see where the needle is placed and avoid harming baby. The procedure takes about 10 minutes to set up and just a couple of minutes to do.

Chorionic Villus Sampling (CVS)

CVS can be undertaken from 11 weeks of pregnancy. Chorionic villi are small thread-like projections of tissue that form within the developing placenta. The cells that make up the chorionic villi will almost always have the same chromosomes as baby. This means that if baby has a chromosomal abnormality, the cells of the chorionic villi will also show the same abnormality.

During a CVS a fine needle is used to draw off a small sample of this tissue from the placenta. An ultrasound scanner is used to guide the doctor performing the CVS. The procedure can sometimes take a few minutes longer than an amniocentesis.

Risks of Amniocentesis and CVS

Both these procedures have a small risk of miscarriage.

The risk of miscarriage after an amniocentesis is about 1 in 150 to 1 in 200. The risk of miscarriage after CVS is about 1 in 100 to 1 in 150.

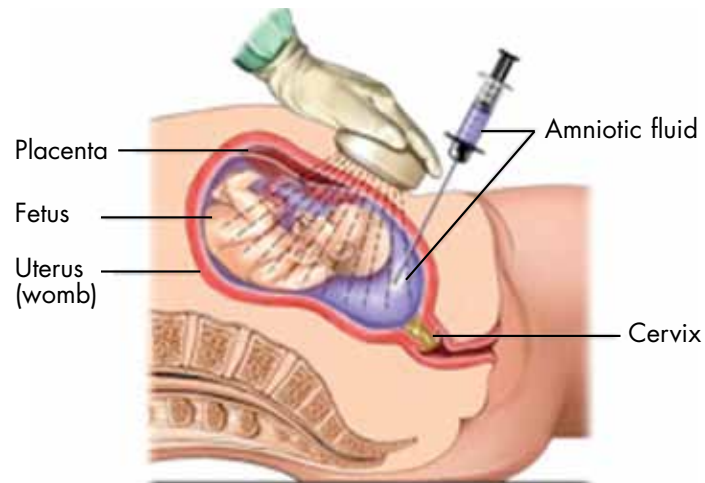
Very rarely amniotic fluid can start leaking vaginally after either procedure. This usually settles with rest and time.

Occasionally, a result is obtained that can't be interpreted because the sample of fetal cells also has some maternal (your) cells mixed in with it. In about 5% of cases a planned CVS can't be performed or no sample can be obtained. This is usually because the placenta is difficult to access.

Much less commonly, it is not possible to obtain fluid at an amniocentesis and a second procedure needs to be done at a later date.

Should I have an Amniocentesis or CVS?

CVS appears to have a slightly higher risk of miscarriage associated with it and is technically more difficult to perform. Women are generally guided towards amniocentesis rather than CVS unless their risk of having a child affected by a chromosomal abnormality is particularly high. There are a few rare genetic conditions where a CVS is the preferred diagnostic procedure.



Amniocentesis – fluid is taken from the amniotic sac around baby

Is the procedure painful?

Both procedures can be mildly uncomfortable. It should be no worse than a blood test.

Who will do my amniocentesis or CVS?

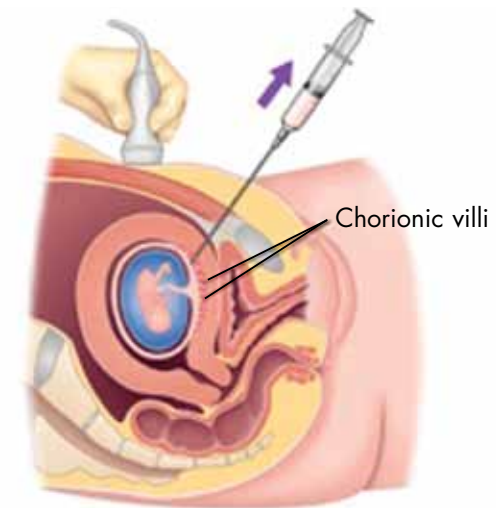
Most women will be referred to the Amniocentesis clinic at National Women's Auckland City Hospital for these procedures. The service is free to all New Zealand residents.

You can be seen urgently within a few days of referral if required. The doctors performing these procedures have all undergone sub-specialty training.

There is also a private service available at Ascot hospital. Most women under our care having an amniocentesis or CVS opt to have their procedure at National Women's.

What happens if I am rhesus negative?

Women with a rhesus negative blood group will be given an injection of "Anti-D" at the time of the procedure. This is to protect a woman with a rhesus negative blood group from developing antibodies against the blood cells of a baby with a rhesus positive blood group. Your blood group will have been checked at the time you booked for antenatal care.



Chorionic villus sampling – tissue is taken from the developing placenta

Can I bring my partner?

We encourage you to bring your partner or a support person. These procedures are often done at a time when women are anxious about a possible abnormality. It is important to have someone else with you for support.

Can I ask questions?

The doctors performing your amniocentesis or CVS will always explain the procedure to you and are happy to answer any questions. They usually won't have access your full pregnancy notes and so may not be able to answer specific questions relating to your care. You are most welcome to call us at AOC if you have any questions concerning your amniocentesis or CVS that you forgot to ask us in clinic.

Recovery after Amniocentesis or CVS

You should plan to have the rest of the day off work and have a quiet day at home. You should avoid strenuous activity for 48 hours.

If you have a sedentary job then you can work the next day. We can provide you with a sick note if you are concerned about the possibility of strenuous work the next day.

There is little evidence that intercourse after either procedure increases your risk of miscarriage but we would suggest that you avoid intercourse for 48 hours.

What happens to the sample?

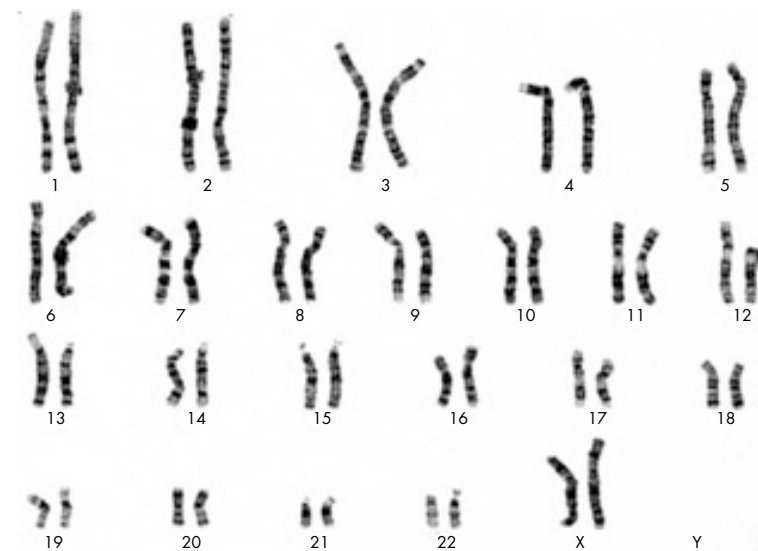
The cells collected by amniocentesis or CVS can be used to test for Down syndrome and other chromosomal abnormalities.

After amniocentesis cells in the amniotic fluid are cultured (stimulated to grow and divide) before the chromosome tests are performed. When enough cells have grown the chromosomes are checked to see if the extra chromosome number 21 found in babies with Down syndrome is present. Other much rarer chromosomal abnormalities where parts of other chromosomes are absent or present as extra copies can also be checked for.

It takes about 10 to 14 days after an amniocentesis for a complete set of results to be available.

After CVS chorionic villi cells are cultured in the laboratory until enough cells are available to check the chromosomes.

It takes about 7 to 14 days after a CVS for a complete set of results to be available.



In the laboratory fetal cells are cultured and the number of chromosomes is checked. This is a sample from a baby girl (two x chromosomes) with a normal number of chromosomes.

What is FISH testing?

A more rapid test can also be done on the sample obtained at amniocentesis or CVS. This is called FISH (fluorescent in-situ hybridisation).

In this laboratory procedure the chromosomes from a very small sample of cells are tested without the need for culturing the cells.

This can give a result within 24 to 48 hours. FISH can detect babies affected by Down syndrome and abnormalities of some other chromosomes but only some of the chromosome pairs are checked and part of the sample is also cultured so that the full range of chromosomes (this is called baby's karyotype) can be checked.

There is no Ministry of Health funding for FISH testing for Down syndrome and National Women's charges \$400 to \$450 for FISH.

The only advantage of FISH is that the result will be available sooner. For some women, especially those with a particularly high risk of a chromosomal abnormality, this may be important.

Can the test be wrong?

Rarely, the cells won't grow in the laboratory (the culture "fails") and no result can be obtained.

Occasionally, the results are difficult to interpret because parts of the cell culture show cells with normal number of chromosomes and other parts don't. This is called a "mosaic pattern". In this situation more tests may be needed.

Great care is taken to ensure all samples are correctly labelled and the correct results are given to the correct woman.

It is also important to remember that most genetic and inherited diseases can not be detected before baby is born and these test only look at baby's chromosomes which for most inherited or genetic condition appear normal.

How will I receive my results?

Most couples prefer us to phone them as soon as the result is available but you may prefer to come into clinic to receive your result in person. Please let us know if you wish to come into clinic for your result.

Should I have genetic counselling?

All of us at AOC are happy to discuss prenatal testing with you.

National Women's also offers a genetic counselling service. Relatively few couples making decisions about having a diagnostic test for Down syndrome feel that they need additional genetic counselling.

For couples whose baby may be affected by rarer chromosomal or genetic problems then this service can provide useful information to help in decision making about pre-natal testing.

Signs of a problem

Call us at AOC if you experience:

- Bleeding or fluid loss vaginally
- Persistent cramping pains
- A fever

We are very happy to see you to check baby's heart beat if you are worried in the days after an amniocentesis or CVS.

Useful contact numbers

Auckland Obstetric Centre

Tel. 09 367 1200

Fax. 09 367 1201

National Women's Ultrasound Department

Tel. 09 631 0782

Fax. 09 631 0782

Useful websites

Auckland Obstetric Centre – www.obstetrics.co.nz

New Zealand Down Syndrome Association – www.nzdsa.org.nz

National Screening Unit – www.nsu.govt.nz

