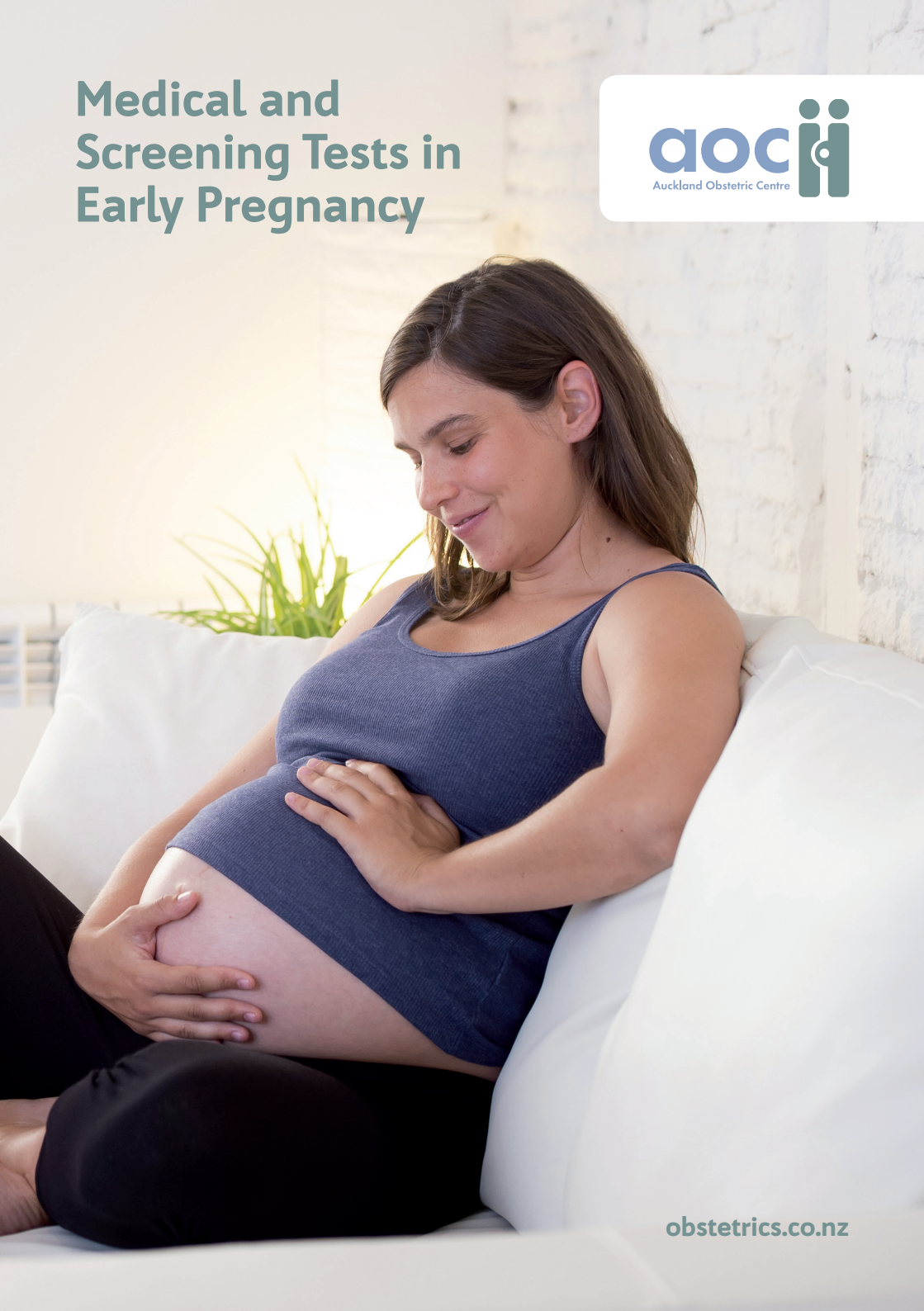
Medical and Screening Tests in Early Pregnancy

aoc
Auckland Obstetric Centre



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This leaflet explains what screening tests are available to pregnant women in the first and second trimester of pregnancy. These include your booking blood tests, screening and diagnostic tests for Down syndrome and an 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 that might interfere with cross matching blood for a transfusion or cause anaemia in a developing or new born baby are also checked for.

Rubella Serology

This is to check that you are immune to Rubella (German measles). Most women have been vaccinated against rubella in childhood but a few women will have little or no immunity.

Hepatitis B

This is to check for infection or immunity to hepatitis B. Hepatitis B carriers can pass hepatitis onto their children and the babies of carriers are usually immunised shortly after birth. Occasionally, hepatitis B carriers will need treatment with anti-viral drugs in pregnancy.

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 rare in pregnant women but carriers can infect their unborn child. Treatments are very effective in reducing this risk 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.

HBA1c (Glycosylated Haemoglobin)

This is a measure of a woman's blood sugar levels over the previous weeks. High levels can indicate underlying diabetes or that there is an increased risk of developing diabetes later in pregnancy.

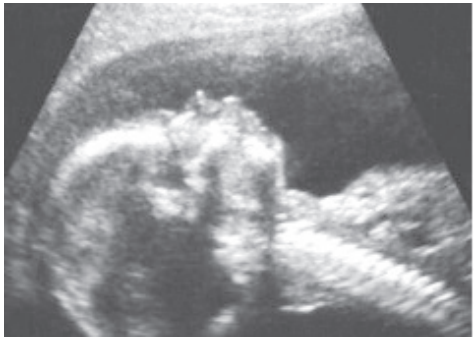
Second Trimester Tests

Vaginal Swabs

Infections such as Chlamydia, which often cause few symptoms, can be checked for in pregnancy though we usually wait until between 12 and 18 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 baby to have a surgical procedure soon after birth.



An anatomy scan at 20 weeks of pregnancy

Some couples are very anxious that a severe abnormality will be found and a termination may be discussed. 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 on a scan.

It is often possible to tell if baby is a boy or girl but this is not the main 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.

Occasionally it is not possible to obtain detailed views of every part of baby at an anatomy scan because baby is not lying in the best position for a scan. You will be asked to return for a follow-up scan a week later.

Polycose Test

This is done at 24 to 28 weeks of pregnancy to check if you are at an increased risk of developing diabetes in pregnancy (often called gestational diabetes). You don't need to be "starved" and should a normal diet on the day of the test. You will be given a drink containing 50g of glucose and an hour later some blood is taken. This is about the same amount of sugar as in a can of soft drink. 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 then have blood taken before having a drink containing 75g of glucose and then have more blood taken two hours later.

A positive polycose test is not a "panic situation" – about 10 to 15% of women will have a positive polycose test. Only about a third of women with a positive polycose test will go on to have gestational diabetes confirmed by a glucose tolerance test.

Gestational diabetes is associated with high blood sugars in pregnancy. Exposure to high sugar levels can result in baby becoming excessively large or needing extra glucose in the first few days after it is born. Women who develop gestational diabetes are also at greater risk of developing diabetes in later life.

Full Blood Count, Ferritin Levels and Antibody Screen

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.

Down Syndrome Screening

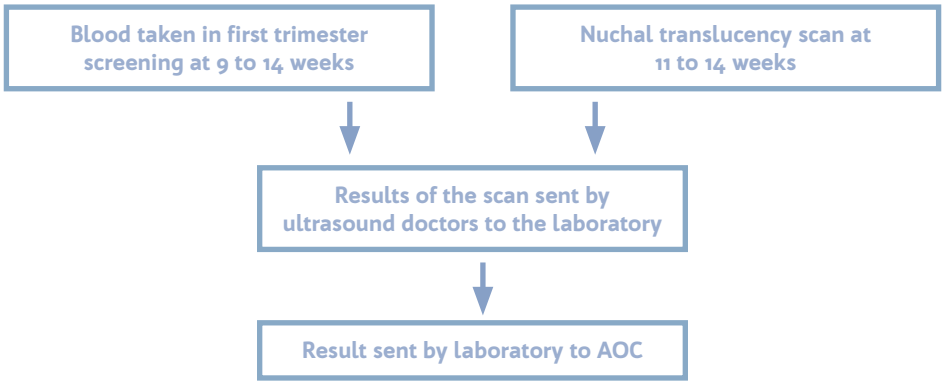
At your booking visit you will be offered a screening test for Down syndrome. This is usually a "combined screening test" in which the results of a blood test and scan are combined to produce a single result which is given as a "risk" (for example 1 in 500 or 1 in 2000). This is called MSS1 (maternal serum screening first trimester). For women booking for pregnancy care after 14 weeks it is only possible to have screening by a blood test, called MSS2 (maternal serum screening second trimester).

There is also an alternative screening test available, which is a blood test called Non-Invasive Pre-natal Testing or "NIPT". This measures the levels of baby's DNA from chromosome 21 in your blood and can be done at any time after 10 weeks. It would still be important to have a scan at 11-13 weeks to check early anatomy. There is no public funding for this tests at present.

Many couples find their screening options confusing. We are here to help you understand the options that are available to you.

What is Down syndrome?

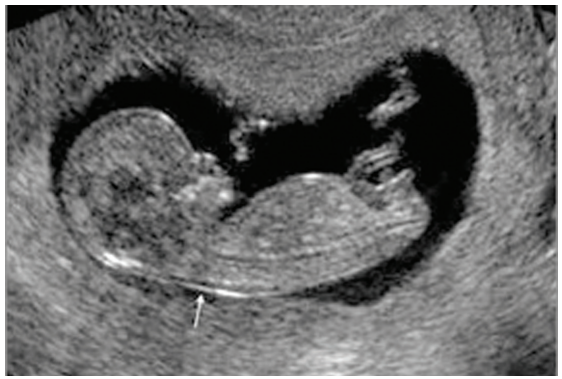
Normally we have 23 pairs of chromosomes. A child with Down syndrome has an extra copy of the twenty first chromosome (the condition is also called Trisomy 21). This is the result of a problem arising at the moment of conception. Why it occurs is unknown. It becomes commoner with increasing maternal age. For example, at 30 years of age a woman has a one in 600 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 70. 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 about Down syndrome look at the websites listed on our useful websites and information page on our website.



First Trimester Combined Screening

For women booking for antenatal care during the first trimester, screening for Down syndrome consists of a blood test taken at between 9 and 14 weeks of pregnancy and an ultrasound scan (called a nuchal translucency scan) performed at 11 to 13 weeks and 6 days of pregnancy.

This screening test starts with a risk based on your age. Your individual risk is calculated using the levels of two proteins in your blood, an ultrasound scan measurement taken from the back of baby's neck (nuchal translucency) and the presence or absence of the nasal bone ('present' means it is brighter than the skin overlying it). The two proteins in your blood (called pregnancy-associated plasma protein (PAPP-A) and beta-human chorionic gonadotrophin (Beta-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. Babies with Down's syndrome more commonly have an 'absent nasal bone'. This means that the nasal bone is less bright than the overlying skin so it is deemed 'absent' for the purpose of the screening test but will actually still be present. By putting these measurements together your individual risk of carrying a baby affected by Down syndrome is calculated. Together these tests are referred to as first trimester combined screening test or the "integrated test" (MSS1).



A nuchal translucency scan

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 85 per cent of babies with 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). A risk greater than 1in 300 is considered high risk and you will be offered the opportunity to have a diagnostic test (see below for more information).

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

Overall about 2% to 4% of women will opt to have an amniocentesis or chorionic villus sampling (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.

The combined screening test will also screen for other rarer trisomies including trisomy 13 and 18. The nuchal translucency scan can occasionally pick up early signs of an anatomical problem with baby too.

Second Trimester Serum Screening for Down Syndrome

Women booking for antenatal care after 14 weeks of pregnancy (too late for MSS1) can still have a Down syndrome screening blood test. This is called second trimester serum screening. This can be arranged 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 measuring the levels of three different proteins in your blood.

There is no need to have this later test if you have already had the earlier combined test or NIPT.

Screening by Free Fetal DNA in Maternal Blood

A newer screening test for Down syndrome is also available privately. This is a blood test that can be done after 10 weeks. The test measures the levels of baby's DNA circulating in your blood. In all pregnancies very small amounts of "cell-free fetal DNA" originating from the placenta can be detected in maternal blood. Different levels of fetal DNA related to chromosome 21 will be detected in a pregnancy affected by Down syndrome. This test is sometimes called "non-invasive pre-natal testing" or NIPT. NIPT will detect over 99% of babies affected by Down syndrome and is less likely to give a non-reassuring or "false-positive" result than the combined screening test. It can also detect high levels of DNA associated with trisomy 18 and 13. Test results typically take five to eight days to come back.

So far, the test has been mainly used by women with a non-reassuring combined screening test result who are keen to avoid an amniocentesis or older women with a higher risk based on their age. It is not reliable enough to be considered a diagnostic test – not all women with a positive NIPT test result will have a baby with Down syndrome. However, a negative test reduces the chances that a woman is carrying a baby with Down syndrome to well below 0.1 per cent (less than 1 in a thousand). It is not currently part of the publicly funded national screening programme.

There are a number of different companies providing NIPT testing. AOC currently uses the Victorian Clinical Genetic Services (VCGS) in Australia called PERCEPT. Patients make an appointment for blood collection with us and we then arrange for this to be couriered to Australia. You can read more about the PERCEPT Prenatal test by clicking their link in our useful websites and information page on our website.

Screening Costs

The screening programme is funded by the Ministry of Health. Some community-based ultrasound facilities will add a part-charge for a nuchal translucency scan. Nuchal translucency scanning can also be done at Auckland City Hospital at no cost. NIPT testing is not funded by the Ministry of Health.

Should I Have Screening Tests For Down Syndrome?

Screening tests for Down syndrome are all optional. The great majority of women who have the combined test will receive a reassuring result, but some women will be given a risk assessment that means they need to consider having a diagnostic test (an amniocentesis or CVS).

Every year a few couples that we see will be told that their baby has Down syndrome. Couples who find out they are going to have a child with Down syndrome will sometimes decide to have a termination, but for other couples, continuing with their pregnancy is the right decision.

It is important that you consider what choices you might make yourself when having any screening test. We all feel our role at AOC is to support couples in their decision making and provide information to help them.

If you are unsure about your screening options, we are very happy to talk to you by phone between appointments or arrange extra visits to clinic if you need more help in deciding what tests to have. We can also provide you with additional written information to help guide your decision making.

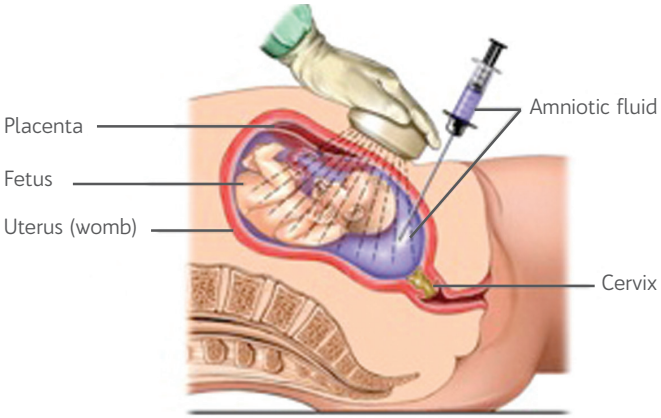
Amniocentesis and Chronic Villus Sampling

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 can talk to you 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.



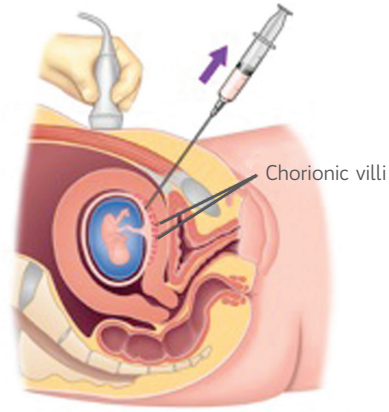
Amniocentesis
Fluid is taken from the amniotic sac around baby.

Chorionic Villus Sampling (CVS)

CVS can be undertaken from 11½ 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.



Chorionic villus sampling
Tissue is taken from the developing placenta.

Risks of Amniocentesis and CVS

Both these procedures have a small risk of miscarriage. The risk of miscarriage above the background rate after an amniocentesis is about 1 in 1000 and after CVS is about 1 in 500.

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 up to 4% 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 higher risk of miscarriage associated with it and can be technically more difficult to perform. Some women prefer to have amniocentesis rather than CVS because of this, however some women prefer to have a CVS as it can be performed at an earlier gestation allowing reassurance at an earlier stage if the result is normal, or allowing more time for decision making if the result is abnormal. Sometimes an amniocentesis will be required following a CVS for clarification of results.

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.

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?

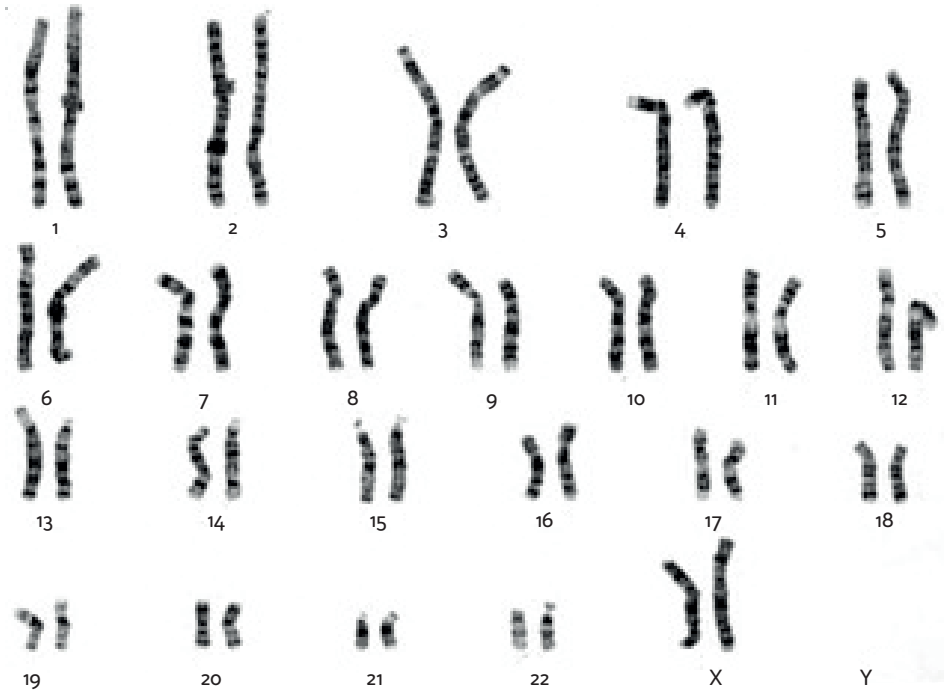
The cells collected by amniocentesis or CVS can be used to test for Down syndrome or other chromosomal alterations.

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 up 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 up 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.

FISH can detect babies affected by Down syndrome and some other chromosomal trisomies but only the chromosome pairs most commonly associated with trisomy are checked. Part of the sample is also cultured so that the full range of chromosomes (this is called baby's karyotype) can be checked.

It takes about 24 to 48 hours after a FISH test for a result to be available.

There is limited Ministry of Health funding for FISH testing for Down syndrome. Women with a risk of higher than 1 in 50 for Down syndrome or with structural abnormalities can have FISH testing for free. National Women's charges $300 for women with a lower risk.

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 cannot be detected before baby is born and these tests only look at baby's chromosomes which for most inherited or genetic condition appear normal.

How will I receive my results?

Many 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 after amniocentesis and CVS

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
P: 09 367 1200 | **F:** 09 367 1201

National Women's Ultrasound Department
P: 09 631 0782 | **F:** 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

This booklet was prepared from information provided by National Women's Health and the Royal Australian and New Zealand College of Obstetricians and Gynaecologists.

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