

Prenatal Testing Information Booklet

This pamphlet provides information about the prenatal diagnostic tests CVS and amniocentesis. Your LMC will be able to advise you as to whether it is appropriate for you to have one of the following tests.

You do not have to have any of the tests. If you agree to have a test that has been recommended to you because of particular risk factors you will be asked to sign a consent form confirming that consent. It is important that you consider the information given to you and discuss it with your LMC before making a final decision to have a prenatal test.

Tena koutou katoa, Kia orana,

Talofa lava, Malo e lelei,

Fakaalofa lahi atu, Taloha Ni,

Ni Sa Bula Vinaka,

Greetings and Welcome

Women's Health, Auckland Hospital



Chorionic Villus Sample or Amniocentesis?

Amniocentesis or Chorionic Villus Sampling (CVS) are prenatal diagnostic tests 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.

Who are these tests offered to?

- Women who have already given birth to a child with a chromosomal abnormality.
- Women whose Maternal Serum Screening (MSS) test or nuchal translucency test has given them a 'high risk' result.
- Women from 35 years of age at the expected date of delivery. These women have a higher chance of giving birth to a baby with Down Syndrome.
- Occasionally these tests may be done (through special arrangements with the genetic service) to detect other inherited conditions such as cystic fibrosis, haemophilia or certain types of muscular dystrophy.

Referral

A referral letter is required from your LMC or Doctor.

How much will it cost?

There is no charge to NZ citizens and residents who meet the above criteria.

What preparation is required?

- Prior to the procedure being performed you will have the opportunity to discuss the implications and courses of action with an obstetrician. If you agree to have this test, the doctor carrying out the procedure will ask you to sign a consent form.
- You are welcome to take more time to think about the test and have some additional sessions with a genetic counsellor or consultant obstetrician if you need to.
- An ultrasound examination is performed before an amniocentesis or CVS to determine the age and position of the baby and location of the placenta.
- It is necessary to know your blood group because if you are rhesus negative you will require an injection of anti D to prevent the formation of antibodies that may harm the babies of future pregnancies.

As these procedures are done as an outpatient, it is suggested that your partner or a support person attend.

It is not necessary to have a full bladder for either procedure and you may eat and drink normally. These procedures can be uncomfortable. Some pressure and angling of the probe is necessary to get good views of the baby. It can take some time to choose the best spot to place the needle.

Chorionic Villus Sampling

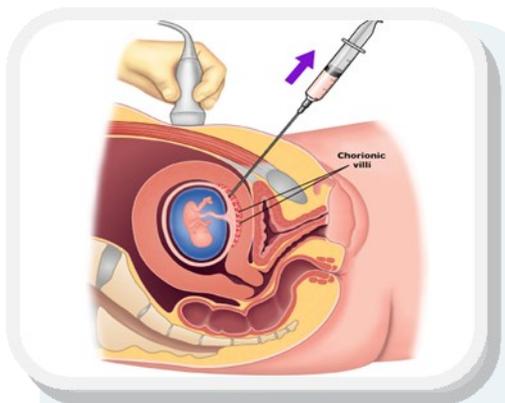
Chorionic Villus Sampling or CVS is the technique of obtaining a small sample of the developing placenta or afterbirth for testing. Chorionic villi are small threadlike projections of tissue that form within the placenta. The cells that make up the chorionic villi will almost always have the same chromosomes as the baby.

When is it performed?

A CVS is usually carried out between 11+2 and 13+6 weeks of pregnancy.

How is the procedure carried out?

An ultrasound is performed to determine the age and position of the fetus and the location of the placenta. A local anaesthetic is injected into the skin, then under ultrasound guidance a needle is passed through the abdominal wall and uterus to the placenta. Very small amounts of tissue are then drawn up into the syringe. Occasionally it is necessary to insert the needle more than once to get a sample. There is some discomfort as the needle enters the uterus.



A CVS – tissue is taken from the developing placenta.



What tests are performed on the sample and how long will it take?

Chromosome analysis is available from the tissue. The cells take time to grow and multiply before analysis. Results will be available from your LMC 10-14 days following the procedure.

The sex of the baby will also be revealed by this test and parents can decide whether they wish to have this information or not. When a female fetus is reported there is a slight chance that this result has been obtained from cells of maternal origin.

What are the risks involved?

CVS has a 0.2% (1:500) risk of miscarriage above the background risk for natural miscarriage.

In some women a result will not be available after the procedure because no sample is obtained, or because cells did not grow. Occasionally the CVS is unable to be performed because of the placental position which will be discussed with you at the time of the scan. In these circumstances you are offered an amniocentesis at 15-16 weeks.

After the CVS

There may be some discomfort due to bruising and some cramps may be experienced. 2 Paracetamol tablets can be taken to ease these cramps (they will not harm the baby).

If there is any loss of blood or fluid from your vagina, or strong pains, you should consult your doctor or midwife immediately as you could be at risk of having a miscarriage.

We recommend you rest quietly at home for the remainder of the day and have someone drive you home. Do not perform any tasks involving heavy lifting or strenuous activity for 24 hours.

Amniocentesis

Amniocentesis is when a small sample of amniotic fluid (the fluid surrounding the baby) is removed and sent for testing.

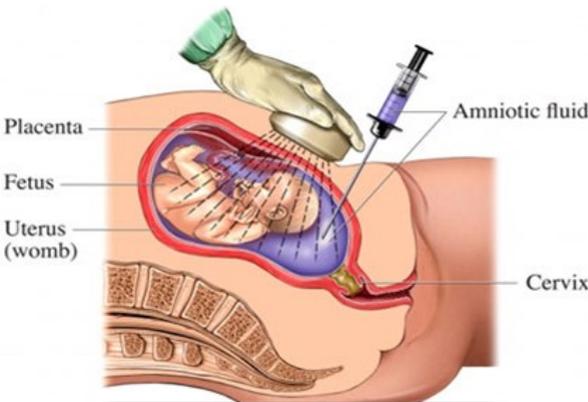
When is it performed?

An amniocentesis is usually carried out from 15+ weeks of pregnancy.

How is the procedure carried out?

An ultrasound scan is performed to find the position of the fetus, the placenta and a suitable amount of amniotic fluid. The ultrasound will also be used to guide the placement of the needle.

A patch of skin is cleaned on your abdomen with antiseptic. A fine needle is then guided through the abdomen and into the amniotic fluid. This may be mildly uncomfortable but usually only lasts for about 30 seconds. A small amount of fluid is withdrawn.



Amniocentesis – fluid is taken from around the fetus.



What tests are performed on the fluid and how long will it take to get the results?

Chromosome analysis – cells from the baby are present in the amniotic fluid. These are grown and cultured before being analysed. This takes 10-14 days, after which time the results will be available from your LMC. Most women will be 17-19 weeks pregnant when these results are known.

The sex of the fetus will be revealed by this test and parents can decide whether they wish to have this information or not. As with CVS, when a female fetus is reported there is a slight chance that this result has been obtained from cells of maternal origin.

What are the risks involved?

Amniocentesis is a relatively safe procedure but there is a small risk of miscarriage. About 0.1% (1 in 1000) of women will miscarry as a result of the procedure.

What do I look for after amniocentesis?

There may be some discomfort and some women experience period like cramps, but these usually resolve within 24 hours. 2 Paracetamol tablets can be taken to ease these cramps (they will not harm the baby).

We recommend you rest at home with no heavy lifting or strenuous activity after the amniocentesis. If you have any bleeding or fluid leaking from your vagina, you should contact your doctor, midwife or the hospital immediately as you could be at risk of having a miscarriage.



FISH test

A more rapid test can be done on the sample obtained at amniocentesis or CVS. This is called FISH (fluorescent in-situ hybridisation) and there is no need to culture the sample. This test only detects babies affected by Down syndrome, Trisomy 18 and 13 or some sex chromosome abnormalities. This can usually give a result within two working days.

There is no Ministry of Health funding for FISH testing and the cost for this is \$300 unless there is a clinical need.

Amniocentesis and CVS only detect chromosomal abnormalities so other abnormalities may not be detected.

What are my options if there is an abnormality?

If your results reveal an abnormality you will be told what the problem means for the child and if treatment is available. You and your partner can then decide whether you wish to continue the pregnancy. Social work counsellors are available as well as information about community based support groups.

Can the test be wrong?

Rarely, the cells won't grow in the laboratory and no result can be obtained. Occasionally the results are difficult to interpret because of a mosaic pattern. This is where some cells show normal chromosomes and others don't. In this situation more tests may be needed.



Need more information?

For further information about matters discussed in this booklet please discuss with your Lead Maternity Carer (LMC) or contact these numbers:

National Women's Health Ultrasound Department

Amniocentesis/CVS bookings phone: (09) 307 2811

Fax: (09) 307 2868

National Women's Fetal Medicine Unit

Phone (09) 307 4949 ext 24951

Northern Regional Genetic Service at Auckland Hospital

Phone: (09) 307 4949 ext 25870 or 0800 476 123

Fax: (09) 307 4978



[Pregnancy and Early Family Care - Auckland DHB](#)

The National Women's Health website has good, accurate and up to date information which you may find helpful.

nationalwomenshealth.adhb.govt.nz

Women's Health,
Auckland City Hospital