

Amniocentesis

Your doctor has referred you for an amniocentesis. Ample time will be available at your visit to discuss any questions you may have. You may find it helpful to read the description of the indications for the procedure and its risks and benefits below.

WHAT IS AN AMNIOCENTESIS?

Amniocentesis is a procedure where a small quantity of the fluid surrounding the fetus in the uterus is withdrawn with a needle. The sample is withdrawn through a needle passed through the mother's abdominal wall. It is then sent to the laboratory for genetic testing

WHEN & HOW IS IT PERFORMED?

An amniocentesis is usually performed from the 15th week of pregnancy onwards. Sometimes the procedure may need to be deferred for a week or two for technical reasons.

After an ultrasound scan, the woman's abdomen is cleaned with a sterile solution and then a sterile sheet is placed over her. The ultrasound probe is placed in a sterile bag and, whilst the doctor is scanning, a very fine needle is inserted through the abdominal wall into the uterus. A syringe is attached to the needle and approximately 20 mls of amniotic fluid is withdrawn. The needle is then removed. The procedure only takes one or two minutes. This may be a little uncomfortable but is similar to having a blood test.

If the woman has the Rhesus negative blood group, she will have an injection of Anti-D after the procedure.

WHAT ARE THE INDICATIONS?

In New Zealand, women are offered screening with the combination of a scan (Nuchal Translucency) and a blood test to assess the risk of the fetus having Down Syndrome (Trisomy 21). Women who have had a high risk result with this screening may wish to have an amniocentesis to determine whether the fetus has this genetic abnormality.

Some women may not have a high risk of a genetic abnormality but wish to have an amniocentesis for reassurance.

WHAT IS THE TESTING?

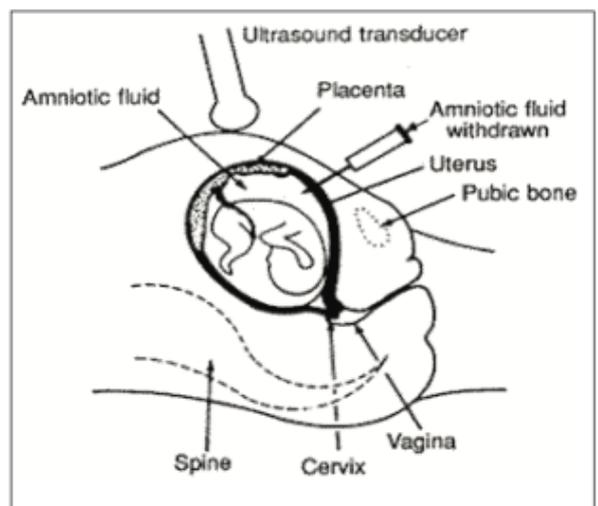
Cells shed from the skin of the fetus and float in the amniotic fluid. These cells are identified in the laboratory and used for analysis.

Every cell of the body contains genetic material stored in the nucleus. There are thousands of genes which code for many features and characteristics. The genes are stored in long strands called chromosomes. Each cell has 46 chromosomes which are made up of 23 pairs. One of each pair comes from the mother and one from the father. In the laboratory, the number and size of the chromosomes is examined. This is called a Karyotype. The individual genes are too small to see and different testing is done for these if it is indicated.

Trisomy 21 occurs when there are three chromosomes at pair number 21 rather than two. This brings a lot of extra genetic material and genes which lead to the condition known as Down Syndrome.

The two other conditions which are seen most commonly are **Trisomy 13 and 18**. They have an extra chromosome at pair 13 and 18 respectively.

The results will go to your Lead Maternity Carer (LMC) and will take 10 to 14 days. You can choose to find out the sex of your baby.



Amniocentesis CONTINUED

RISKS OF THE TESTING

There is a risk of miscarriage with amniocentesis of approximately 1 in 1000. **Bleeding or cramps after 12 hours are abnormal and you should call your LMC.** Very rarely the membranes can rupture and the woman leaks a small amount of clear fluid. This usually reseals and is not a problem.

LIMITATIONS OF TESTING

This test will not diagnose many abnormalities (e.g. cleft lip & palate, spina bifida and cystic fibrosis). Careful scanning will be used to screen for structural abnormalities and an anatomy scan is recommended even when a Karyotype is reported as normal.

THE TESTING PROCESS

Specimen testing is either done through Auckland District Health Board or IGENZ (a private testing laboratory) or a combination of the two.

Women who are New Zealand residents and are identified as high risk (i.e. maternal age etc) have their Karyotype costs covered through the ADHB. You can also choose to have an additional test done through IGENZ called Fluorescent Insitu Hybridisation or FISH). This is an optional test which specifically targets certain chromosomes and produces limited results within a shorter time frame than the standard full test.

Low risk Karyotype testing is done through IGENZ. You can also opt to have the FISH test done.

PRICING

The criteria for an amniocentesis and levels of risk can vary from patient to patient. Once an appointment has been made for you, our booking staff will contact you at a later date with specific pricing details.

