Testing if you are pregnant

Most children born in Australia are born healthy. But about 2 or 3 in 100 are born with a condition that means they will need medical care.

Some of the more common conditions are:

- Congenital heart disease
- Disorders of the kidney and bladder
- Hip dislocation at birth
- Club foot
- Down syndrome and other chromosome alterations
- Spina bifida and related conditions, which are known as neural tube defects and which are problems in the development of the spinal cord and/or brain
- Cleft lip and/or palate
- Developmental delay.

Some of these can be detected in pregnancy, while others cannot.

Screening tests and/or diagnostic tests are available for some of these disorders. They are not compulsory – it is your choice whether or not to have these tests.

Before having any tests, you need to consider what you would do if a test came back positive or indicates some degree of risk. What would you do if a test showed your unborn child had an abnormality? Would you consider a termination of pregnancy? Would you not consider one? Would you just like to know, even if you don't plan to do anything about it? Or would you rather not know?

Screening tests

These tests do not give a firm diagnosis, but aim to give parents an idea of whether or not they have a higher than normal risk of having a child with the disorder being screened for.

None are perfect – sometimes screening tests miss the conditions they are meant to detect.

If a screening test picks up an increased risk, then there are diagnostic tests – chorionic villus sampling, amniocentesis or ultrasound – that can sort out whether or not the baby has the disorder.

First trimester screening test

This test is designed to identify women at increased risk of having a baby with Down syndrome, but it can sometimes also identify other problems.

The test has three parts. The first is a blood test at 10 to12 weeks of pregnancy. The second is an ultrasound, called a nuchal translucency or NT test, at 11 to13 weeks. The third part is the woman's age, which is also taken into account.

These three pieces of information are combined to calculate the risk that the baby has Down syndrome. Couples with an increased risk will be offered genetic counselling to consider their choices; the choice of whether or not to have a diagnostic test – either chorionic villus sampling or amniocentesis – to check the baby's chromosomes.

Nuchal translucency test

Nuchal translucency is used to estimate if a baby is at an increased risk of having a chromosomal abnormality. It uses ultrasound to see and measure a fluid filled sac at the back of the unborn baby's neck during early pregnancy.

The nuchal translucency test, which is part of the first trimester screening test, can sometimes be done on its own, without the blood test. This ultrasound is carried out between 11 and 13 weeks of pregnancy and is reasonably accurate, but not as accurate as the combined first trimester screening test.

Second trimester maternal serum screening test

This blood test is best done between 15 and 17 weeks of pregnancy, but it can be carried out between 14 and 20 weeks. The second trimester screening test is suitable for women who did not have either the first trimester screening test or the nuchal translucency test. It can tell parents whether the baby is at increased risk of Down syndrome (and/or some other chromosomal alterations) or a neural tube defect, which is a problem in the development of the spinal cord and/or brain.

The second trimester test is not as accurate as the first trimester screening test.

Ultrasound

Most pregnant women will have an ultrasound at 18 to 20 weeks. This ultrasound checks the baby's growth, the stage of pregnancy, and the amount of amniotic fluid, the position of the baby and placenta. This ultrasound also looks for physical problems such as neural tube defects, heart and kidney malformations, cleft lip and limb abnormalities.

Diagnostic tests

These tests aim to give a firm diagnosis of a potential problem. They are more accurate than screening tests. Like all tests, they are only looking for specific gene alterations or chromosome alterations (see fact sheet on '*What is a gene?*'). They are not perfect and they may occasionally miss something.

Chorionic villus sampling

This test can detect chromosomal alterations as well as genetic conditions, which your doctor knows to look for because they have happened before in the family, or because a genetic screening test has shown that you could have an affected baby.

Chorionic villus sampling is usually done at around 11 weeks of pregnancy and preferably by 13 weeks. Usually, a needle is guided through the abdomen to the tissue that will form the placenta, and a small fragment of tissue is removed. Occasionally a plastic tube is guided through the vagina and cervix instead. In both cases, the procedure is monitored by ultrasound so that the needle or plastic tube is kept away from the baby. Most women find it uncomfortable rather than painful.

Women who have chorionic villus sampling have a slightly increased risk of miscarrying afterwards. The risk is between 1 in 100 and 1 in 200.

The results are usually available in two to three weeks.

Amniocentesis

This test can detect chromosomal alterations. The test can also detect gene alterations, which your doctor knows to look for because they have happened before in the family, or because a genetic screening test has shown that you could have an affected baby.

Amniocentesis is usually done at 15 to16 weeks of pregnancy and preferably before 20 weeks. A needle is guided into the fluid around the baby and a small amount of fluid is removed. The procedure is monitored by ultrasound so that the needle is kept away from the baby. Most women find it uncomfortable rather than painful.

Women who have amniocentesis have a slightly increased risk of miscarrying afterwards. The risk is about 1 in 200.

The results are usually available in two to three weeks.

Ultrasound

A detailed ultrasound may be used to look for certain disorders that have happened before in the family. Ultrasound can also be carried out at any time if problems arise in the pregnancy.

Contacts and further information

- All states and the ACT have familial cancer services. Contact them through your local state or territory health department.
- Your local hospital antenatal clinic.
- MyDr at **http://www.mydr.com.au**
- HealthInsite at **http://www.healthinsite.com**
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email **gtis-australia@unimelb.edu.au** or visit Biotechnology Australia's website at **http://www.biotechnology.gov.au**