Prenatal Testing
Special tests for your baby during pregnancy

There are a number of different prenatal (before birth) tests to check the development of your baby. Each test has advantages and disadvantages. This information is for

- people who are planning a pregnancy
- people who are already pregnant and want information about these tests.

There is no test that gives a 100% guarantee of a healthy baby. The tests below can give some information about your baby’s health. They do not find all potential health problems.

Some form of prenatal testing is usually offered to all pregnant women in NSW. You can choose

- whether you want any testing at all (you may not want any tests) or
- which tests are best for you (after talking to your doctor, midwife or genetic counsellor first).

Some people decide to have prenatal testing because they want to know if their unborn baby has a condition that causes serious physical and/or intellectual problems. Some people decide not to have prenatal testing. The choice is always yours.

Types of prenatal tests
There are several different tests that may be available to you during pregnancy. Some of them are screening tests and some are diagnostic tests.

Screening Tests
Screening tests give a result that tells you the ‘risk’ of your baby having certain conditions that cause intellectual and/or physical problems. These tests do not give definite answers about your baby’s health. If a screening test result suggests your baby has an ‘increased risk’ of a problem, you may consider having a diagnostic test. Talk about the advantages and disadvantages of further testing with your doctor, midwife or genetic counsellor. Screening tests do not harm the baby or the mother.

Diagnostic Tests
Diagnostic tests give a result that tells you more definitely whether or not the baby has a condition that causes intellectual and/or physical problems. These tests give
a very reliable answer about the health of your baby. Some diagnostic tests (CVS and amniocentesis) have a small chance (less than 1%) of causing a miscarriage.

If a diagnostic test shows your baby has a problem, you can learn about how it will affect the baby. You can then decide whether you want to continue the pregnancy or not. Your doctor, midwife or genetic counsellor will give you information and support while you are making this decision and afterwards.

The list of tests below tells you about the prenatal tests available at different stages of pregnancy.

For more information about genetic counselling and prenatal testing, ask your doctor, midwife or genetic counsellor, or contact The Centre for Genetics Education, Tel: (02) 9926 7324, or go to www.genetics.com.au

If you need help making phone calls in English, ring the Translating and Interpreting Service (TIS) on 131 450.

You can find more information in your language on the Multicultural Health Communication website at http://mhcs.health.nsw.gov.au

Tests available at different stages of pregnancy

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Stage of pregnancy: 11-13 weeks of pregnancy

Name of test
Chorionic villus sampling (CVS)

Type of test
This is a diagnostic test.

How is this test done?
With the help of ultrasound, a small sample of the placenta (afterbirth) is taken through the mother’s abdomen, using a thin needle or through the cervix (the entrance to the womb), using a thin flexible tube.

What does this test look for?
This test can check for a range of intellectual and/or physical problems that the baby may have. These are known as chromosomal problems. Sometimes more testing may be needed.

Are there any risks to the baby or mother?
There is a small chance (less than 1%) that this test may cause a miscarriage. The mother may feel some discomfort during the test.

Stage of pregnancy: 11½-13½ weeks of pregnancy

Name of test
Nuchal translucency (ultrasound) test with or without testing of the mother’s blood

Type of test
This is a screening test

How is this test done?
Using an ultrasound, a special measurement (nuchal translucency) is taken of the baby. Also, a sample of the mother’s blood may be taken for testing.

What does this test look for?
This test can tell if a baby has an increased risk of certain intellectual and/or physical problems.

About 5% of babies tested may have an increased risk result. Most of these babies will NOT have a problem.

If the nuchal translucency test is done without the blood test:
About 75% of babies who have an intellectual and physical problem called Down syndrome will have an increased risk result. 25% of babies with Down syndrome will be missed by this test.

If the nuchal translucency ultrasound is done together with the blood test:
About 80-90% of babies who have Down syndrome will have an increased risk result. 10-20% of babies with Down syndrome will be missed by this test.
Are there any risks to the baby or mother?
This test does not harm the baby or the mother.

Stage of pregnancy: 15-18 weeks of pregnancy

Name of test
Maternal serum triple test

Type of test
This is a screening test

How is this test done?
A sample of the mother’s blood is taken for testing.

What does this test look for?
This test can tell if a baby has an increased risk of certain intellectual and/or physical problems.

About 5% of babies tested may have an increased risk result. Most of these babies will NOT have a problem.

About 60% of babies who have Down syndrome will have an increased risk result. 40% of babies with Down syndrome will be missed using this test.

If this test is done at the same time as a detailed ultrasound scan, it will also identify about 95% of babies who have spinal problems or neural tube defects.

Are there any risks to the baby or mother?
This test does not harm the baby or the mother.

Stage of pregnancy: 15-19 weeks of pregnancy

Name of test
Amniocentesis

Type of test
This is a diagnostic test.

How is this test done?
With the help of ultrasound, a small sample of the amniotic fluid (fluid surrounding the baby in the uterus or womb) is taken through the mother’s abdomen, using a thin needle.

What does this test look for?
This test can check for a range of intellectual and/or physical problems that the baby may have. These are called chromosomal problems.
Are there any risks to the baby or mother?
There is a small chance (less than 1%) that this test may cause a miscarriage. The mother may feel some discomfort during the test.

Stage of pregnancy: 18 weeks of pregnancy

Name of test
Detailed fetal anomaly ultrasound scan

Type of test
This is a diagnostic test.

How is this test done?
An instrument like a microphone is pressed on to the mother's abdomen. This shows a picture of the developing baby.

What does this test look for?
This test can check the size of the baby and many physical features such as the heart, brain and kidney development.

Are there any risks to the baby or mother?
This test does not harm the baby or the mother.