

Prenatal Testing

testing during pregnancy



Ultrasound
Maternal Serum Screening
Chorionic Villus Sampling
Amniocentesis

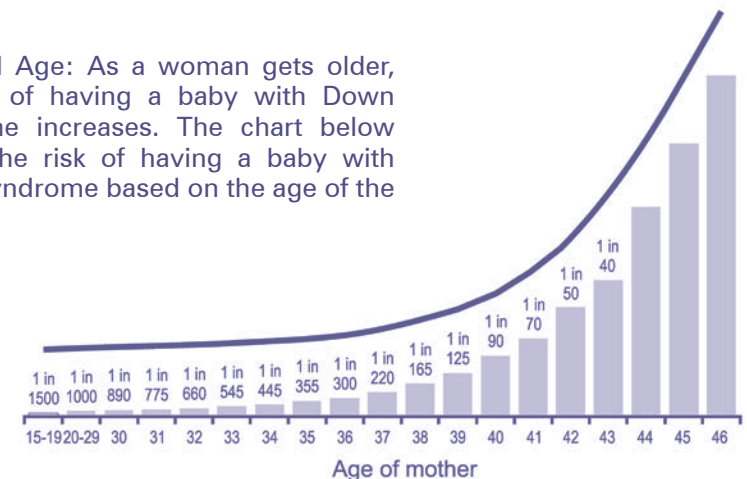
Prenatal Testing Information

Most babies are born healthy, however a few will have birth defects. About four in one hundred babies will be born with a birth defect. Women may choose whether or not to have testing during pregnancy to find out their risk of having a baby with a birth defect.

There are two types of tests: "Screening" tests are available to all pregnant women, and "Diagnostic" tests are specific tests for diagnosis and are offered to women who may be at an increased risk of having a baby with Down syndrome or another condition. There are out of pocket expenses for some of these tests.

Genetic Counselling is recommended before any tests are performed. Your doctor or midwife may provide this, or an appointment can be made with a Genetics Clinic. The doctor or genetic counsellor can provide information about the tests, the conditions tested for, and the advantages and disadvantages of prenatal testing, taking into account your medical, family and pregnancy history. They can also discuss your attitudes and personal beliefs about testing and options if a problem is detected.

Maternal Age: As a woman gets older, her risk of having a baby with Down syndrome increases. The chart below shows the risk of having a baby with Down syndrome based on the age of the mother.



Screening tests (non-invasive tests) are available to all women

Screening tests are available to all pregnant women. They are also called non-invasive because there is no risk of miscarriage from the procedures. There are two types of screening tests: blood tests and ultrasound scans.

Blood tests from the mother called "maternal serum screening" will measure the level of particular hormones and indicate the risk of a woman having a baby with Down syndrome or another problem.

Blood tests do not diagnose these conditions, but indicate whether or not there is an increased or decreased risk in this pregnancy.

There are two different types of maternal serum screening tests. You can only have one or the other, not both.

1st trimester combined screening is a blood test done at 9-13 weeks of pregnancy (best during 10th week), combined with a nuchal translucency ultrasound scan done at 11-13* weeks of pregnancy.

This test will indicate the risk of having a baby with Down syndrome and another chromosome condition called Trisomy 18. As this is a combined test, a risk cannot be calculated using the blood sample alone.

If this test indicates an increased risk the woman will be offered a diagnostic test, either CVS or amniocentesis (see below).

2nd trimester maternal serum screening also called the quadruple test is a blood test done at 14-20 weeks of pregnancy (best during 15th week). This test will indicate the risk of having a baby with Down syndrome, Trisomy 18 or neural tube defects (ie. spina bifida or anencephaly).

If this test indicates an increased risk for Down syndrome or Trisomy 18 the woman will be offered amniocentesis. If this test indicates an increased risk for spina bifida or anencephaly the woman will be offered an ultrasound examination to look for neural tube defects.

Ultrasound scans involve the use of sound waves to create a picture of the baby, which is viewed on a television screen.

Current knowledge suggests that there is no harmful effect on the mother or the developing baby. Ultrasound does not involve X-rays. Ultrasound scanning can take place at any time during a pregnancy.

For testing purposes there are two ideal times for doing an ultrasound scan.

1st trimester ultrasound also called nuchal translucency ultrasound is done at 11-13* weeks of pregnancy. This scan measures the skin thickness at the back of the baby's neck (nuchal translucency). When this measurement is increased it can indicate an increased risk of having a baby with Down syndrome or another problem. When an ultrasound is performed at an accredited centre the result can be combined with the first trimester blood test.

If this test indicates an increased risk the woman will be offered diagnostic testing, either CVS or amniocentesis.

2nd trimester ultrasound sometimes called the fetal anomaly scan is done at 18-20 weeks of pregnancy. This scan is used to identify structural problems such as limb or heart defects, in the baby. It does not detect all abnormalities.

If problems are detected the woman will be offered further testing.

Diagnostic tests (invasive tests) are for diagnosis of specific conditions

Diagnostic tests are for diagnosis of specific conditions. They are also called invasive because they carry a risk of miscarriage from the procedure. This risk is in addition to the natural risk of miscarriage at the time of testing.

Diagnostic tests are offered when:

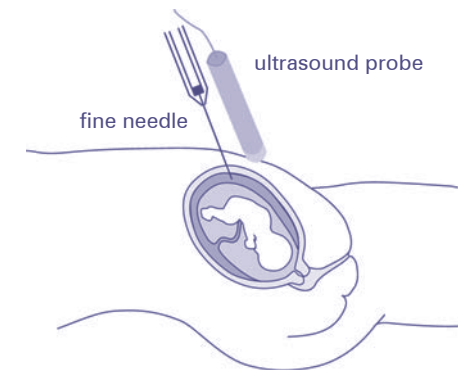
- ❖ There is a family history of a particular condition, or
- ❖ Screening tests have indicated an increased risk of a particular condition, or
- ❖ The fetal anomaly scan has indicated an increased risk of a particular condition, or
- ❖ A woman is 37 years of age or older and does not choose to have screening

These specific diagnostic tests are done on samples of material taken from the placenta (CVS) or amniotic fluid (amniocentesis). After the procedures women should rest for the remainder of the day.

Samples obtained by these procedures are then sent to a laboratory for chromosome testing for conditions such as Down syndrome or genetic (DNA) testing for conditions such as cystic fibrosis. For chromosome testing the full result will usually take two weeks, but in some cases it is possible to get a preliminary result in 24-48 hours (FISH test). If diagnostic testing is chosen you only need to have one or the other, not both.

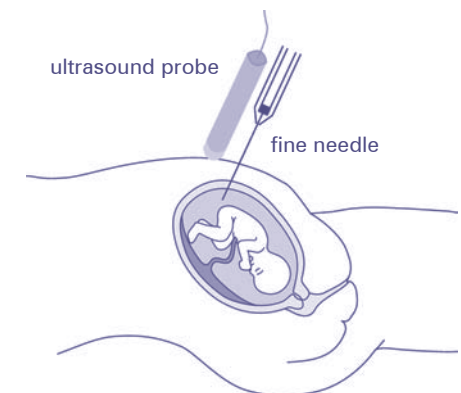
Chorionic Villus Sampling (CVS)

CVS is an outpatient procedure (ie. You do not have to be admitted to hospital) generally performed between 11 and 12 weeks of pregnancy. Under ultrasound guidance a fine needle is inserted through the woman's abdomen into the placental tissue and a small sample of the tissue is withdrawn. (CVS is occasionally done through the vagina). The risk of miscarriage after a CVS is less than 1 in 100 or 1%.



Amniocentesis

Amniocentesis is an outpatient procedure generally performed between 15 and 18 weeks of pregnancy. Under ultrasound guidance a fine needle is inserted through the woman's abdomen and a small sample of amniotic fluid is obtained. The risk of miscarriage is less than 1 in 200 or 0.5%. Amniocentesis is sometimes used to provide information about neural tube defects.



Summary table of tests available in pregnancy

Screening tests available to all pregnant women

Test	When	Detects
Blood Tests		
1st trimester combined screening	Blood test: 9-13* weeks of pregnancy (best during 10th week) Ultrasound: 11-13* weeks of pregnancy (best during 12th week)	This test indicates your risk of having a baby with Down syndrome or Trisomy 18. If the risk is increased a diagnostic test such as CVS or amniocentesis is offered.
2nd trimester maternal serum screening	14-20 weeks of pregnancy (best during 15th week)	This test indicates your risk of having a baby with Down syndrome, Trisomy 18 or neural tube defect. If the risk is increased for Down syndrome or Trisomy 18 amniocentesis is offered. If the risk is increased for neural tube defect an ultrasound scan is offered.
Ultrasound scans		
1st trimester ultrasound (nuchal translucency scan)	11-13* weeks of pregnancy	This scan indicates your risk of having a baby with Down syndrome or other chromosome problem by measuring the thickness of the skin at the back of the baby's neck. If the risk is increased a diagnostic test such as CVS or amniocentesis is offered.
2nd trimester ultrasound (fetal anomaly scan)	18-20 weeks of pregnancy	This scan is used to identify physical and structural abnormalities including spina bifida, heart defects and limb defects.

Diagnostic tests available for diagnosis of specific conditions

Test	When	Detects
Chorionic Villus Sampling (CVS)	11 - 12 weeks of pregnancy	Samples of placenta obtained by this procedure can be used to diagnose chromosomal conditions such as Down Syndrome or genetic conditions such as cystic fibrosis.
Amniocentesis	15 - 18 weeks of pregnancy	Samples of amniotic fluid obtained by this procedure can be used to diagnose chromosomal conditions such as Down Syndrome or genetic conditions such as cystic fibrosis.

* The nuchal translucency scan should be done between 11 weeks & 3 days and 13 weeks & 6 days. We recommend that this scan is performed at a RANZCOG accredited practice

The blood test for 1st trimester combined screening should be done between 9 weeks & 0 days and 13 weeks & 6 days.

For further information or to arrange a consultation please contact Genetic Health Services Victoria

at The Royal Women's Hospital 03 8345 2180
at Monash Medical Centre 03 9594 2026
In Tasmania 03 6222 8296

at the Royal Children's Hospital
Maternal Serum Screening 03 8341 6303 or 03 8341 6356
Website <http://genetichealth.net.au>