

Summary of tests available to pregnant women

TEST	TEST CAN DETECT INCREASED RISK OF:	WHEN TO HAVE THE TEST:
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Blood Tests

1st trimester combined screening	Down syndrome Trisomy 18	Blood test: 9-13 weeks gestation (best between 10 & 11 weeks) Ultrasound: 11-13 weeks gestation (best between 12 & 13 weeks)
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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	Down syndrome Trisomy 18 Neural tube defects	Blood test: 14-20 weeks gestation (best between 15 & 17 weeks)
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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 and 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)	Down syndrome other chromosomal conditions Some physical abnormalities	11-13 weeks gestation
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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)	Physical & structural abnormalities including: spina bifida, heart defects & limb defects	18-20 weeks gestation
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This scan is used to identify physical and structural abnormalities including spina bifida, heart defects and limb defects.

Chorionic Villus Sampling (CVS)

Chorionic Villus Sampling (CVS)	Chromosomal conditions Including: Down syndrome Genetic conditions including: cystic fibrosis	11-12 weeks gestation
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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

Amniocentesis	Chromosomal conditions Including: Down syndrome Genetic conditions Including: cystic fibrosis	15-18 weeks gestation
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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.

Diagnostic tests available for diagnosis of specific conditions

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 vaginal). 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.



* The nuchal translucency scan should be done between 11 weeks & 1 day 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.
1 The blood test for 2nd trimester maternal serum screening should be done between 14 weeks & 0 days and 20 weeks & 6 days

Prenatal Testing

Testing During Pregnancy:
 Ultrasound
 Maternal Serum Screening
 Chorionic Villus Sampling
 Amniocentesis



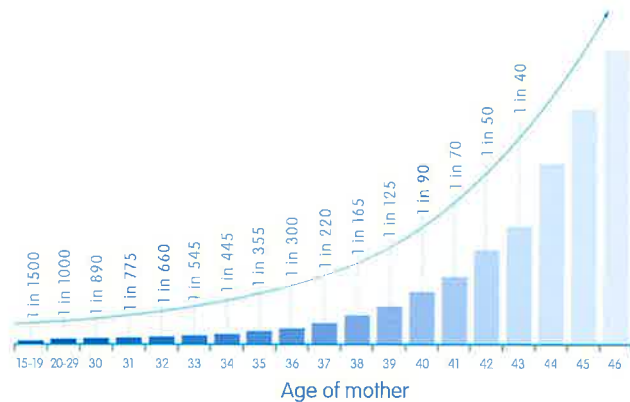
For further information or to arrange a consultation please contact
VICTORIAN CLINICAL GENETICS SERVICES
 Maternal Serum Screening: **1300 934 355**
 The Royal Women's Hospital: **03 8345 2180**
 Monash Medical Centre: **03 9594 2026**
 The Royal Childrens Hospital: **03 8341 6201**
 Tasmania: **03 6222 8296**
 website : vcgs.org.au

Prenatal testing information

Most babies are born healthy, however a few will have birth defects. About four in every 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 genetic 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 above 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 in pregnancy

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 in the mother's blood and indicate the risk of a woman having a baby with Down syndrome and some other chromosomal conditions.

Blood tests do not diagnose these conditions, but indicate whether there is an increased or decreased risk in this pregnancy. There are two different types of maternal serum screening tests. You can have one or the other, not both.

1st trimester combined screening is a blood test done at 9-13⁺ weeks of pregnancy (ideally between 10 and 11 weeks gestation), 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 (ideally between 15 and 17 weeks gestation). This test will indicate the risk of having baby with Down syndrome, trisomy 18 or neural tube defects (ie spina bifida or anencephaly).

If the blood sample is collected during the 14th week of pregnancy, no neural tube defect risk is given.

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 a neural tube defect 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 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 fluid 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 condition.

The specialist performing the scan may also report whether the nasal bone is present or absent. If the nasal bone is absent it can indicate an increased risk of having a baby with Down syndrome. 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.