Chorionic villus sampling (CVS)

Chorionic villus sampling (CVS) is performed from 11 weeks of pregnancy. An ultrasound is first performed to date the pregnancy and check that the position of the placenta and fetus is suitable for performing the procedure. Occasionally the procedure may not be possible and your doctor will discuss this with you. A sterile needle is guided into the placenta and a small sample is taken for testing. CVS has a miscarriage risk of 1%. Sometimes, a test result may be difficult to interpret and it may be necessary to undergo further testing, such as amniocentesis, to clarify the result.

Amniocentesis (Amnio)

Amniocentesis is performed from 16 weeks of pregnancy. Under ultrasound guidance, a needle is inserted through the abdomen into the amniotic sac around the fetus and fluid is taken for testing. Amniocentesis has a miscarriage risk of 1%.

Test results

CVS and amniocentesis test the fetal chromosomes. Other genetic testing can occur where indicated. A rapid chromosome screening test takes 2-5 days. This only assesses for a handful of chromosomal disorders including Down syndrome, Edward syndrome, and Patau syndrome, amongst others. This test can also determine fetal gender. Normal rapid screening results are very reassuring; however it is important to wait for the final chromosome results which may take 2–3 weeks for confirmation. The time taken for other genetic test results may vary depending on the test.

What is genetic testing?

If you are considering a pregnancy or are pregnant, it is advisable to obtain a referral to a genetics service if you have a personal and/or family history of an inherited disorder (e.g. cystic fibrosis, Fragile X or Duchenne muscular dystrophy). Tests on couples or their family members may be required before prenatal diagnostic testing can be offered in a pregnancy.

Limitations of prenatal screening and testing

Prenatal screening and diagnostic tests are designed to detect disorders in a fetus before birth. Some conditions can be treated after birth. However, chromosome abnormalities and some other genetic disorders cannot be reversed, which may have serious consequences for the baby. In these situations, some couples may wish to have information prior to the birth of their baby so they have time to prepare; other couples consider requesting a termination of the pregnancy. No prenatal test can give a full guarantee that the baby will be normal in every way. However the majority of couples will have a healthy child.

Genetic Health Queensland (GHQ)

For more information about Genetic Health Queensland, educational material and details about making appointments, please contact the main office or the genetic counsellor at one of the outreach centres.

Royal Brisbane and Women's Hospital (Main Office)	07 3646 1686
Office hours are 8 am to 5 pm.	
Royal Brisbane and Women's Hospital Prenatal Service	07 3646 2269
Cairns and Townsville	07 4433 1464
Bundaberg, Rockhampton and Mackay	07 4150 2794
Nambour	07 5470 6068
Toowoomba	07 4616 6995
Gold Coast	07 5687 1515

Genetic Health Queensland

C/-Royal Brisbane and Women's Hospital
Butterfield Street, Herston QLD 4029

Phone: 07 3646 1686
Fax: 07 3646 1987
Email: ghq@health.qld.gov.au
Web: www.health.qld.gov.au/ghq

QH453 07/16

Genetic Health Queensland

Prenatal screening and testing

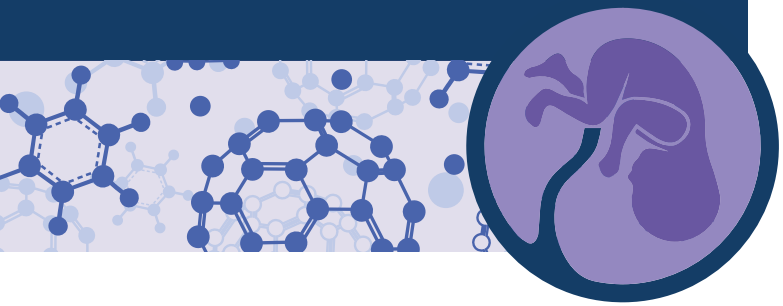
Queensland Government

What are prenatal tests?

Prenatal tests are medical investigations performed during a pregnancy to check on the health of the developing fetus (baby).

The most common tests are:

- nuchal translucency ultrasound at 12–14 weeks combined with a blood test (biochemistry) from the mother
- detailed ultrasound at 18–20 weeks
- chorionic villus sampling (CVS) from 11 weeks
- amniocentesis from 16 weeks.



What is combined nuchal translucency screening?

This screening test is performed between 12–14 weeks of pregnancy, and is an optional ultrasound scan and blood test for all pregnant women. An ultrasound involves the use of high frequency sound waves to create images of the fetus. Ultrasound is non-invasive and has been shown not to cause harm to the fetus.

Your blood test, ultrasound scan and age are combined to calculate the risk of your fetus having Down syndrome or other less common chromosome problems. Down syndrome is more likely if your fetus has extra fluid at the back of the neck, if the nasal bone can not be seen and/or your blood test is out of the normal range. Assessing the nasal bone is a new feature and it improves the accuracy of the screening. It is important to ask if the centre where you are having your screening performed also includes a nasal bone in their risk assessment.

You are not required to have a risk assessment for Down syndrome. However a 12–14 week ultrasound can give you other important information about your pregnancy such as whether you are having twins. Ask your doctor or genetic counsellor for more details.

What is the second trimester blood test/triple test?

This screening blood test can be taken between 15–20 weeks of pregnancy. This blood test provides you with a risk assessment for Down syndrome (if you could not have the combined first trimester test) and spina bifida.

What is non-invasive prenatal testing?

This screening blood test identifies fetal DNA in the blood stream of the mother, and tests for Down syndrome as well as some other common chromosome problems. If you have a positive test result for Down syndrome, it is more than 98% likely that the fetus has Down syndrome. A negative test result means that the fetus has a 1:10000 chance of having Down syndrome. If positive, this test does not replace invasive testing, and the results should be confirmed with either a chorionic villus sampling or amniocentesis.

This test may not be possible for all pregnancies, such as triplets or in pregnancies where one twin has died. Please discuss with your doctor or genetic counsellor if this test is a good screening test for you.

What is the ultrasound at 18–20 weeks?

An ultrasound at 18–20 weeks is a routine scan to assess the growth and development of the fetus. At this time, some structural abnormalities (such as spina bifida, cleft lip/palate and heart defects) may be identified on scan. Ultrasound may detect certain signs in the fetus which suggest an increased risk of a chromosomal or other genetic problem. It is not possible to detect all structural abnormalities or all chromosome problems on ultrasound. This scan is not very good at screening for Down syndrome.

Do I need a prenatal diagnostic test?

Chorionic villus sampling and amniocentesis are prenatal diagnostic tests. These are invasive tests which allow for the analysis of the baby's chromosomes and, in some cases, genetic testing for inherited conditions.

Prenatal diagnostic testing may be offered if:

- the mother is 35 years of age or older at delivery and has had no other screening tests
- there is an increased risk of Down syndrome or other chromosome problems from screening tests in pregnancy
- there is an abnormal finding on ultrasound
- there are concerns the mother has had certain infections during pregnancy
- the mother has had a condition herself which could put the fetus at risk
- Or if either parent:
  - » is a carrier of a chromosome problem
  - » has a personal and/or family history of an inherited disorder
  - » has had a pregnancy or child with an inherited or chromosome disorder.

Is counselling available before testing?

Discussion with your doctor, obstetrician, and/or a genetic counsellor is recommended before any prenatal diagnostic test is performed. You will receive:

- information to enable you to make an informed decision regarding prenatal testing
- a thorough explanation of the prenatal tests and the options available to you
- a discussion regarding the possible outcomes of testing and what your options are
- information on how long test results will take and who will give the results to you.