

Genetic counselling with a VCGS genetic counsellor is available to anyone considering this screening test (1300 118 247). VCGS genetic counsellors can also discuss other prenatal screening and testing options with you. More information about this test is available at vcgs.org.au/mss

Purpose of the test

VCGS offers screening for some conditions that can be identified during pregnancy. Maternal serum screening has two options: screening during the first trimester or screening during the second trimester.

- **Combined first trimester screening (CFTS):** provides an estimate of the chance that a pregnancy has a chromosome condition such as Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) or Patau syndrome (trisomy 13). An optional extra with CFTS is screening for early-onset pre-eclampsia (EO-PE). Early detection and treatment can improve pregnancy outcomes.
- **Second trimester screening (2ndTMSS):** provides an estimate of the change that a pregnancy has Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) or a neural tube defect, such as spina bifida.

Test process

CFTS combines a blood test with an ultrasound.

1. A blood test between 9 weeks + 0 days and 13 weeks + 6 days (if having PE as well, blood must be collected between 11 weeks and 13 weeks + 6 days). This measures two proteins called PAPP-A and free beta hCG. These are found naturally in the blood during pregnancy. A change in the level of these proteins may indicate a greater chance of a chromosome condition. The blood sample can be collected at any 4Cyte Pathology centre.
2. A nuchal translucency ultrasound (NT scan). This ultrasound is performed by a specialist ultrasonographer between 11 weeks + 1 day and 13 weeks + 6 days. The ultrasound measures the thickness of a fluid filled space at the back of baby's neck and also the presence or absence of the nasal bone. An enlarged space and/or absence of the nasal bone may indicate a greater chance of a chromosome condition.

The results of the blood test and the NT ultrasound are combined with the patient's age and weight to give a combined estimate of the chance the pregnancy may have a particular condition.

2ndTMSS is a blood test only.

Blood is collected between 14 and 20 weeks. Although if the sample is collected between 14 and 15 weeks, no estimate will be given for neural tube defects.

Limitations of the test

- Maternal serum screening is not a diagnostic test. Therefore, not all pregnancies with a chromosome condition will be identified. Screening only looks for a small number of particular chromosome conditions: no screening test can identify all possible conditions that may affect a pregnancy.
- A screen negative result means it is very unlikely the pregnancy has a chromosome condition. The majority of women will receive a screen negative result. As with all screening tests, there is always a small chance the result is incorrect (false negatives).
- A small number of women will receive a screen positive result. This does not mean the pregnancy definitely has a chromosome condition - only that there is a higher chance. If you receive this result, you will be offered diagnostic testing to confirm the result. In most cases, the screen positive result turns out to be a 'false positive' and the pregnancy does not have the chromosome condition. If you receive a screen positive result, there are two options for further testing. You can have *percept* non-invasive prenatal test (NIPT), which is a more accurate screening test or you can have a diagnostic test. A diagnostic test requires an invasive procedure such as a CVS or amniocentesis.

Privacy, confidentiality & use of information

- Your test results will be kept confidential. Results will only be released to your healthcare provider, other healthcare providers involved in your medical care, or to another healthcare provider as directed by you, or otherwise as required or authorised by applicable law.
- Collecting information on your pregnancy after testing is part of our laboratory's standard practice for quality purposes and test evaluation. VCGS may contact your healthcare provider to obtain this information.

Retention & use of samples

- In line with best practices and clinical laboratory standards, leftover de-identified specimens (unless prohibited by law), de-identified genetic material, as well as other information learned from your testing, may be used by VCGS for purposes of quality control, laboratory operations, laboratory test development, laboratory improvement, and generation of new scientific knowledge. All such uses will be in compliance with applicable law.

Financial responsibility

- You are responsible for fees incurred with VCGS for services performed.