

Increased risk results for Down syndrome and/or trisomy 18

- We recommend that all women who receive an increase risk result are offered a diagnostic test (chorionic villi sampling (CVS) or amniocentesis – see below).
- Only a diagnostic test can confirm whether or not the pregnancy is affected:
 - An increased risk does not mean that the pregnancy is definitely affected by Down syndrome or trisomy 18.
 - Most women with increased risk results do not have an affected pregnancy
- Discuss with the patient the different scenarios that could unfold once a diagnostic result is received. For example, if her pregnancy is confirmed positive for Down syndrome or trisomy 18, she will be given the choice of whether to continue or to terminate the pregnancy
- Post test counselling is available from Genetic Health Services Victoria for women at increased risk

Brief Comparison between CVS and amniocentesis

	CVS	Amniocentesis
Gestation at testing	Between 11 & 13 weeks	15 weeks or later
Risk of miscarriage	1 in 100	1 in 200

13 week Combined Screen

- Performed when blood is collected during the 13th week of pregnancy
- Risks are calculated for Down syndrome and Trisomy 18
- 1st trimester analytes (BhCG and PAPP-A) and 2nd trimester analytes (AFP, uE3, inhibin A) are measured
- Information from the serum is combined with the CRL and NT from the scan
- Results are issued within 5 working days of the ultrasound report being received by the laboratory

The quadruple test: This test is ideally performed at 15 weeks gestation and will provide a risk result for trisomy 21 (Down syndrome), trisomy 18 (Edward syndrome) and Neural Tube Defects (NTD) such as spina bifida. If the test is performed in the 14th week a risk result for NTDs cannot be generated.

When is the best time to perform these tests?

Screening Test	Ideal Timing	Acceptable Range
1st trimester serum test	During the 10th week	9 - 13+6
NT ultrasound scan	During the 12th week	11+ 3 - 13+6
Quadruple test	15 - 17 weeks gestation	14 - 20 weeks

How to contact us

The Clinical Support Team can be contacted on:
 (03) 8341 6303/ 6356/ 6476
 Monday to Friday 9am to 5pm

The Clinical Support Team is available to assist with any concerns health professionals have regarding the MSS test.

Website details:

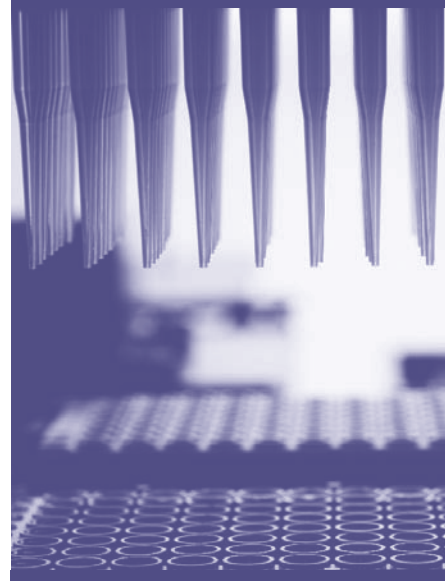
www.vcgspathology.com.au

for information about the 1st & 2nd Trimester Maternal Serum Screening

Combined First Trimester Screening for:

Trisomy 21 or Trisomy 18

Serum test and ultrasound at 10 - 12 weeks



Information for Health Professionals

Down syndrome (Trisomy 21) is the most common chromosomal cause of intellectual disability. It can occur in any pregnancy regardless of the woman's age.



What is combined screening?

Combined screening combines results of a blood test with a nuchal translucency ultrasound scan during the 1st trimester of pregnancy to determine the risk of having a baby affected with trisomy 21 or trisomy 18.

This test is a private test and is only available when requested by a private medical practitioner. The ultrasound test should be organised with a private ultrasonologist skilled in measurement of nuchal translucency.

Step 1: The pathology request form: Please document

- Date of birth, weight and gestation (LMP or agreed EDD)
- Previous pregnancy information:
 - Pregnancy affected with Down syndrome?
- Current pregnancy information:
 - Threatened miscarriage?
 - IVF pregnancy? The age of the egg used to achieve the pregnancy will affect her combined risk result (Egg pick-up date, donor DOB)
- Date and place of the nuchal translucency ultrasound

Combined first trimester screening request pads can be ordered through the Maternal Serum Screening Laboratory on 03 8341 6356/6357/6303

Step 2: The serum screening test (optimally performed during the 10th week)

- The blood is collected in a plain tube (5-7.5mL) between 9 and 13 weeks 6 days gestation (see-13 week combined screen)
- The blood test measures two proteins
 - Free beta human chorionic gonadotrophin (free beta-hCG)
 - Pregnancy associated plasma protein A (PAPP-A)
- A change in the level of these proteins indicates that there is an increased risk that the fetus has Down syndrome or trisomy 18

- Private pathology services should be asked to forward the blood
 - All blood samples should be sent to the Maternal Serum
- Screening Laboratory at the Victorian Clinical Genetics Services, Royal Children's Hospital in Melbourne
- Alternatively, your patient is welcome to make an appointment with the Maternal Serum Screening Team to have blood taken at the Royal Children's Hospital (Ph: 8341 6303 or 8341 6356)
 - There are out of pocket costs for the blood test

Step 3: The ultrasound (ideally at 12 weeks)

- The ultrasound is performed between 11 weeks 3 days and 13 weeks 6 days gestation
- Performed by an accredited ultrasonologist/ultrasonographer
- Two fetal measurements used
 - Crown rump length (CRL) – must be between 45 and 84mm inclusive
 - Nuchal translucency (NT)
- All fetuses at this stage of pregnancy have some nuchal fluid, but on average babies with Down syndrome or another chromosomal abnormality have a greater amount of fluid
- **Patients should not be given the NT risk without combining the serum results as the combined screen has a higher detection rate and lower false positive rate than NT risk alone.**

There are usually out of pocket costs for the ultrasound scan. Individual practices need to be contacted regarding costs

Step 4: After the ultrasound

- The ultrasound practice needs to fax a copy of the ultrasound report to the Maternal Serum Screening Laboratory (Fax: 03 8341 6389)
- The ultrasound report needs to include the date of scan, the CRL and NT measurements
- The laboratory will generate the combined risk result
- The ultrasound practice should inform the laboratory if the patient is waiting for her results or if she will receive her results on another day

Step 5: The risk estimates for the pregnancy

- The results from the serum test and the nuchal measurement are divided by their gestational age specific medians and reported as Multiples of the Median (MoM).
- This is used with the woman's age-related risk to give a final risk for Down syndrome and trisomy 18.

Getting the results

- If the patient is waiting the result will be faxed to the ultrasound practice and posted to the referring Doctor
- If the ultrasound practice indicates that the patient is not waiting the combined risk result will be generated usually within 48 hours. The result is faxed to the ultrasound practice and/or the referring doctor.

Low Risk Result

- Low risk results are usually faxed to the referring ultrasound clinic and/or the referring doctor within 48 hours of receiving the ultrasound report
- A copy of the result is also mailed to the referring doctor

Increased Risk Result

- Increased risk results are phoned and faxed to the ultrasound clinic and/or the referring doctor as a matter of urgency on the day that the result is issued.

On-line reporting

- Maternal Serum Screening results are available on request by email. To access this service please contact Ivan Francis on (03) 8341 6273 or by email: ivan.francis@ghsv.org.au

The meaning of risk results and what next

- Results are always reported as a risk figure (1 in X) and are identified as either low risk (less than 1 in 300 for Down syndrome and 1 in 250 for trisomy 18) or increased risk
- This test can detect 9 out of 10 pregnancies affected with Down syndrome (90% detection rate) but the actual detection rate increases with age

Low risk results for Down syndrome and trisomy 18

- A low risk result means it is unlikely that this pregnancy has Down syndrome or trisomy 18 but does not exclude the possibility. This test does not exclude other fetal abnormalities.
- No further diagnostic tests are recommended on the basis of screening.