

Increased risk results for Down syndrome and/or trisomy 18

- An increased risk result in the first trimester is a risk value:
 - greater than or equal to 1 in 300 for Down syndrome
 - greater than or equal to 1 in 175 for trisomy 18
- An increased risk result means that there is an increased likelihood of Down syndrome and/or trisomy 18 for this pregnancy
- 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.
- Only a diagnostic test can confirm whether the pregnancy is affected. We recommend that all women who receive an increased risk result are counselled and offered a diagnostic test (chorionic villus sampling (CVS) or amniocentesis – see below).



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Information for Health Professionals

MATERNAL SERUM SCREENING

Combined First Trimester Screening For:
 Trisomy 21 or Trisomy 18
 Serum test & Ultrasound

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

- Discuss with the patient the different scenarios that could unfold once a diagnostic result is received.
- If the patient's 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
- Genetic Counselling is available from Victorian Clinical Genetics Services Ph: 1300 934 355

When is the best time to perform these tests?

Screening test	Ideal timing	Acceptable range
1st trimester serum test	Between 10 and 11 weeks gestation	9 to 13+6
Nuchal translucency ultrasound scan	Between 12 and 13 weeks gestation	11+1 to 13+6
Quadruple test	15 – 17 weeks gestation	14-20 weeks

How To Contact Us:

FOR ALL QUERIES RELATING TO MATERNAL SERUM SCREENING
 call: **1300 934 355**
 website: **vcgs.org.au**



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 with trisomy 21 or trisomy 18.

This test 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: Complete the pathology request form

Please document your patient's:

- Date of birth, weight and gestation (LMP or agreed EDD)
- Whether the patient has had a previous pregnancy affected with Down syndrome
- Current pregnancy information: if there has been a threatened miscarriage; IVF information: if fresh or frozen cycle, the egg pick-up date, patient's own egg or donor egg, DOB of the donor (if a donor egg is used)
- Date and location of the nuchal translucency ultrasound

Maternal Serum Screening request slips can be ordered through the Maternal Serum Screening Laboratory on 1300 934 355

STEP 2: Patient has the serum screening test (optimally performed between 10 and 11 weeks gestation)

- The blood is collected in a plain tube (5-7mL) between 9 and 13 weeks 6 days gestation
- The blood test measures two proteins:
 - Free beta human chorionic gonadotrophin (free beta-hCG)
 - Pregnancy associated plasma protein A (PAPP-A)
- Changes in the level of these proteins may indicate that there is an increased risk that the fetus has Down syndrome or trisomy 18
- A risk cannot be calculated using the blood sample alone.

- Blood can be collected at a pathology collection centre. 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: 1300 934 355)
- There are out of pocket costs for the blood test, for more information contact 1300 934 355
- All blood samples should be sent to the Maternal Serum Screening laboratory at the Victorian Clinical Genetics Services, Royal Children's Hospital in Melbourne

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.

STEP 3: Patient has the ultrasound (ideally between 12 and 13 weeks gestation)

- The ultrasound is performed between 11 weeks 1 day and 13 weeks 6 days gestation
- Performed by an accredited ultrasonologist/ultrasonographer
- Three fetal measurements used:
 - Crown rump length (CRL) must be between 45 and 84mm inclusive
 - Nuchal translucency (NT)
 - Nasal bone (NB)

The nuchal translucency is a collection of fluid at the back of the fetal neck in the first trimester of pregnancy. 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. The presence or absence of the nasal bone is also noted and included in the risk calculation. Absence of the nasal bone may indicate an increased risk of Down syndrome.

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 or the requesting doctor needs to fax a copy of the ultrasound report to the Maternal Serum Screening laboratory in order for a risk to be calculated. (Fax 03 8341 6389)
- The ultrasound report needs to include the date of scan, the CRL, NT measurements and the presence or absence of NB
- The laboratory will combine the results of the blood test with the measurements from the ultrasound to generate the combined risk result

STEP 5: The risk estimates for the pregnancy

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

STEP 6: Getting the results

Low risk result:

- If the ultrasound practice indicates that the patient is waiting, the result will be faxed to the ultrasound practice on the same day and sent to the referring doctor
- If the patient is not waiting, the result is faxed to the referring ultrasound clinic and/or the referring doctor within 48 to 72 hours of the ultrasound report being received by the laboratory
- The laboratory will not issue results directly to patients

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 for requesting practitioners. To access this service, please contact the laboratory on 1300 934 355 or by email: vcgs.mss@vcgs.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 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 for women who receive low risk results from maternal serum screening.