

Maternal serum screening

The maternal serum screening test (MSS) is a blood test, available to pregnant women, that helps to determine the risk of certain abnormalities with their unborn child. The term 'serum' means the watery part of the blood.

Maternal serum screening can be done in the first trimester of pregnancy, when it is combined with ultrasound. This is called a **combined first trimester screening test**. It can also be done in the second trimester of pregnancy and is called the **second trimester maternal serum screening test**.

A range of disorders are screened for

Maternal serum screening can help detect the risk of a range of disorders in the unborn child, including:

- **Down syndrome** – also known as trisomy 21, because the baby has three copies of chromosome 21 instead of two. Characteristics include intellectual impairment and physical defects such as heart abnormalities.
- **Edward syndrome** – also known as trisomy 18. Edward syndrome occurs when the baby has three copies of chromosome 18 instead of two. Characteristics may include malformations of the neural tube, hands and feet, and intellectual impairment.
- **Neural tube defects** – these are only screened for as part of the second trimester maternal serum screening test. Neural tube defects result in a failure of the brain or spinal cord to develop properly. For example, spina bifida is characterised by the incomplete closure of the spinal vertebrae. The spinal cord and nerves protrude through the gap, instead of growing down into the abdominal organs and legs, leading to paralysis of the legs.

MSS should be offered to all pregnant women

Maternal serum screening should be offered to all pregnant women. They will then have the choice of being screened or declining the test. The conditions that are screened for can occur in any pregnant woman of any age, although chromosomal abnormalities such as Down syndrome occur more frequently in older mothers.

It is important to realise that both tests only indicate an increased or lesser risk of a particular condition. They do not detect whether that condition is definitely present. If the test suggests an increased risk, the pregnant woman can then decide whether to undertake further diagnostic tests such as an amniocentesis or chorionic villus sampling (CVS).

Combined first trimester screening

Combined first trimester screening is a test that involves the pregnant woman having a blood test at around the 10th week of pregnancy and a nuchal translucency ultrasound during the 12th week of pregnancy. The nuchal translucency is a pocket of fluid at the back of the baby's neck, which can be best seen between 11 weeks and three days and 13 weeks and six days of pregnancy.

This test shows a risk for Down syndrome and trisomy 18, and identifies women who should be offered further testing. Your doctor or private obstetrician can order this test. Doctors working in public hospitals cannot order it.

Second trimester maternal serum screening

Generally, the MSS test is performed between 15 and 18 weeks into the pregnancy. If the blood test is taken at 15 weeks, the results will be obtained more quickly. The test measures four chemicals in the blood and the results are combined with the woman's age risk to give a final risk of Down syndrome, trisomy 18 and neural tube defects.

When MSS may be used

A pregnant woman may consider undergoing MSS to give her information about her pregnancy and to determine the risk of certain conditions being present in her baby.

It is important to remember that a screening test does not diagnose these conditions. A 'low risk' result does not mean there is no chance of the particular condition being present.

If the test gives an increased risk result

An 'increased risk' result does not necessarily mean the baby has the particular condition. It is important to remember that MSS only indicates an increased risk, not the presence of the disorder. This risk figure is used to help decide if a diagnostic test should be done.

The diagnostic test detects whether the baby has the condition or not. The diagnostic tests are chorionic villus sampling or amniocentesis. A detailed ultrasound scan at 18–20 weeks is the diagnostic test if the second trimester screening results indicate an increased risk for neural tube defects.

Genetic counselling

In most cases, diagnostic testing will show that the unborn baby is healthy. Occasionally, the baby does have the particular condition for which it was tested, such as Down syndrome. Genetic counselling is available for parents to help them make informed decisions about the pregnancy. Information is also available through relevant support networks.

Where to get help

- Your doctor
- Obstetrician
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6201

Things to remember

- The maternal serum screening test (MSS) is a blood test, offered to pregnant women, that helps determine the risk of certain abnormalities in their unborn child.
- The test may be performed at around 10 weeks (combined first trimester screening test) or 15–18 weeks (second trimester maternal serum screening test).
- In most cases that are identified as 'increased risk', the baby is normal.

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Genetic Health Services Victoria

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