Pregnancy tests - maternal serum screening

Summary

- The maternal serum screening test is a blood test offered to pregnant women.
- It helps to inform them of the chance of their unborn child having Down syndrome, Edward syndrome or a neural tube defect.
- The test may be performed at around 10 weeks (combined first trimester screening test) or between 14 and 20 weeks (second trimester maternal serum screening test).
- In most cases that are identified as ‘increased chance’, the baby does not have Down syndrome, Edward syndrome or a neural tube defect.
- Non-invasive pre-natal testing (NIPT) is a newer test that will inform nearly all pregnancies affected by Down syndrome, Edward syndrome and Patau syndrome.

The maternal serum screening (MSS) test is a blood test available to pregnant women in Victoria. It helps determine the chance of their unborn child having Down syndrome (trisomy 21), Edward syndrome (trisomy 18) or a neural tube defect.

Maternal serum screening may be done in the first trimester of pregnancy, when it is combined with an ultrasound. This is called the combined first trimester screening test. Or, testing may be performed in the second trimester of pregnancy. This is called the second trimester maternal serum screening test.

Maternal serum screening tests for various conditions

MSS can identify pregnancies that are at increased chance of:

- **Down syndrome** – a condition that results in intellectual disability of varying degrees. It may cause some physical problems such as heart defects or difficulties with sight or hearing. Down syndrome is also known as trisomy 21 because the baby has three copies of chromosome 21, instead of the usual two
- **Edward syndrome** – a condition that results in physical problems such as growth deficiency, heart defects, digestive system defects and intellectual disability. Babies born with Edward syndrome have a poor prognosis. Many do not live beyond the first few weeks of life. Edward syndrome is also known as trisomy 18 because the baby has three copies of chromosome 18, instead of the usual two
- **neural tube defects** – the most common neural tube defects are anencephaly and *spina bifida*. With anencephaly, the brain does not develop properly and the baby does not survive. Babies with spina bifida have an opening in the bones of the spine that can result in damage to the nerves controlling the lower part of the body. This can cause weakness and paralysis of the legs, and sometimes inability to control the bowel and bladder. Neural tube defects are only screened for in the second trimester MSS test.

This test is only for these conditions. A low-chance result does not mean the baby is free of other birth defects.

**Combined first trimester screening test**

The combined first trimester screening test can identify a pregnancy with increased chance of Down syndrome (trisomy 21) and Edward syndrome (trisomy 18). This test involves the pregnant woman having a blood test between 9 and 13 weeks gestation, and an ultrasound between 11 and 13 weeks gestation. The blood test measures the amount of two different proteins that occur naturally in the mother’s blood during pregnancy.

Using ultrasound, a measurement of the amount of fluid in the skin at the back of the baby’s neck is taken. This measurement is called the nuchal translucency. All babies at this stage of pregnancy have fluid at the back of their neck, but babies with Down syndrome or Edward syndrome tend to have more fluid.

During the ultrasound, the specialist performing the scan may also report the presence or absence of the nasal
bone. If the nasal bone is absent, there may be an increased chance of Down syndrome.

Your doctor or medical specialist can order this test. There may be gap payments depending on the service you are attending – discuss this with your health professional.

Second trimester maternal serum screening test

The second trimester maternal serum screening test can identify a pregnancy with increased chance of Down syndrome, Edward syndrome or neural tube defects. The test involves the pregnant woman having a blood test between 14 and 20 weeks + 6 days gestation. However, it is best if the blood is collected between 15 and 17 weeks gestation.

The test measures the amount of four proteins in the mother’s blood. The results are combined with the woman’s age risk to give a final chance of Down syndrome, Edward syndrome or neural tube defects. The result is reported as an increased chance or low-chance result.

Availability of the maternal serum screening test

Screening for Down syndrome and other chromosome conditions should be offered to all pregnant women. However, the decision to have testing is a personal decision. All women have the chance of having a baby with Down syndrome or Edward syndrome, although the chance increases with the age of the woman.

A low-chance maternal serum screening result does not mean there is no chance of a genetic condition being present in the unborn baby. It means that the chance is low. Similarly, an increased chance result does not necessarily mean that the baby has a genetic condition. Most women with an increased chance result will not have a baby with Down syndrome, Edward syndrome or a neural tube defect.

Maternal serum screening test results

Maternal serum screening tests do not diagnose health conditions, but they identify women who should be offered further testing to detect if their baby has a genetic condition.

Women with an increased chance result on maternal serum screening should be offered a diagnostic test. However, it is their choice whether to have further testing. Chorionic villus sampling (CVS) and amniocentesis are diagnostic tests that detect chromosome conditions such as Down syndrome and Edward syndrome in the unborn baby.

Genetic counselling after a maternal serum screening test

In most cases an ‘increased chance’ maternal serum screening result does not mean the baby has the condition, just that they are at an increased risk of having the condition. This is why further diagnostic testing is encouraged to provide more information about the likely health outcomes of the baby.

If your child has been diagnosed with a genetic condition it can be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand a condition and what causes it, how it is inherited, and what a diagnosis means for your child’s health and development. Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

The Genetic Support Network of Victoria (GSNV) is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by the same genetic condition.

Where to get help

- Your GP (doctor)
- Your obstetrician
- Victorian Clinical Genetics Services (VCGS), Royal Children’s Hospital Tel. 1300 118 247