
Pregnancy tests - maternal serum screening

Summary

- The maternal serum screening test is a blood test offered to pregnant women that helps determine the risk of Down syndrome, Edward syndrome and neural tube defects in their unborn child.
 - The test may be performed at around 10 weeks (combined first trimester screening test) or 14–20 weeks (second trimester maternal serum screening test).
 - In most cases that are identified as 'increased risk', the baby does not have Down syndrome, trisomy 18 or a neural tube defect.
 - NIPT is a new non-invasive pre-natal testing that will detect nearly all pregnancies affected by Down syndrome, Edward syndrome and Trisomy 13.
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The maternal serum screening (MSS) test is a blood test available to pregnant women to help determine the risk of Down syndrome (trisomy 21), Edward syndrome (trisomy 18) or neural tube defects in their unborn child. Serum is the watery part of the blood.

Maternal serum screening may be performed in the first trimester of pregnancy, when it is combined with an ultrasound. This is called the combined first trimester screening test. Alternatively, testing may be performed in the second trimester of pregnancy (second trimester MSS test).

MSS tests for various disorders

MSS can identify pregnancies that are at increased risk 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. In 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.

It is important to understand that this test is only for these disorders, and a low-risk 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 risk 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 risk of Down syndrome.

Your doctor or private obstetrician can order this test. Doctors working in public hospitals cannot order it.

Second trimester MSS test

The second trimester MSS test can identify a pregnancy with increased risk 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 and the results are combined with the woman's age risk to give a final risk of Down syndrome, Edward syndrome or neural tube defects. The result is reported as an increased or low-risk result.

Availability of the MSS test

MSS should be offered to all pregnant women. However, the decision to have testing is a personal decision. All women are at risk of having a baby with Down syndrome or Edward syndrome, although the risk increases with the age of the woman.

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

MSS test results

It is important to understand that the tests do not diagnose these conditions, but they identify women who should be offered further testing to detect if their baby is affected.

Women with an increased risk result on MSS should be offered a diagnostic test. However, it is their choice whether or not to have further testing. Chorionic villus sampling (CVS) and amniocentesis are diagnostic tests that detect chromosome abnormalities such as Down syndrome and Edward syndrome in the unborn baby.

Genetic counselling after an MSS test

In most cases where there is an increased risk MSS result, diagnostic testing will show that the unborn baby does not have the condition, but occasionally the baby does have the condition. 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
- Your obstetrician
- Victorian Clinical Genetics Services, Royal Children's Hospital Tel. (03) 8341 6201

Things to remember

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risk of Down syndrome, Edward syndrome and neural tube defects in their unborn child.

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This page has been produced in consultation with and approved by:

Victorian Clinical Genetics Services (VCGS)

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