Pregnancy tests - maternal serum screening

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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.

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

- The maternal serum screening (MSS) 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.

References


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A-Z of genetic conditions

Ambiguous genitalia

The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.

Angelman syndrome

The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.
- Ankylosing spondylitis
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.

- Barrett's oesophagus
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.

- Bipolar disorder
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings.

- Central nervous system birth defects
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects.

- Charcot-Marie-Tooth disease (CMT)
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.

- Cleft palate and cleft lip
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.

- Congenital adrenal hyperplasia (CAH)
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available.

- Creutzfeldt-Jakob disease (CJD)
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.

- Cri du chat syndrome
  Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems.

- Cystic fibrosis (CF)
  When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria.

- Digestive tract birth defects
  Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

- Down syndrome
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

- Dwarfism
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

- Eczema (atopic dermatitis)
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.

- Essential tremor
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

- Fragile X syndrome
  The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

- Friedreich's ataxia
  To the casual observer, a person with Friedreich ataxia may seem to be drunk.

- Genetic factors and cholesterol
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

- Haemochromatosis
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

- Haemophilia
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

- Hair
Human hair grows one centimetre every month.

Hearing problems in children
The earlier that hearing loss is identified in children, the better for the child’s language, learning and overall development.

Huntington's disease
The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

Kabuki syndrome
Kabuki syndrome affects males and females equally and there is no cure.

Kennedy's disease
Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

Kidneys - medullary cystic kidney disease
Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

Kidneys - polycystic kidney disease (PKD)
Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

Klinefelter syndrome
Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don’t occur.

Leukodystrophy
Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

Long QT syndrome
You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

Marfan syndrome
Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

McCune-Albright syndrome
The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

Muscular dystrophy
People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

Neurofibromatosis
Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

Neonatal syndrome
Neonatal syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

Osteoporosis in children
Osteoporosis in children is rare and usually caused by an underlying medical condition.

Phenylketonuria (PKU)
PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

Prader-Willi syndrome
A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

Premature and early menopause
The symptoms of premature or early menopause are the same as for menopause at any age.

Rett syndrome
People with Rett syndrome have a keen desire to communicate.

Spinal muscular atrophy (SMA)
A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

Tay-Sachs disease
Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians...

- **Thalassaemia**
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated...

- **Tourette syndrome**
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders...

- **Treacher Collins syndrome**
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss...

- **Trisomy disorders**
  Children affected by trisomy usually have a range of birth defects, including delayed development and intellectual disabilities...

- **Tuberous sclerosis**
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity...

- **Usher syndrome**
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing...

- **Von Willebrand disease**
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth...

- **Williams syndrome**
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life...

- **Wilson disease**
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes...

**Genes and genetics explained**

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  Children inherit physical characteristics such as eye colour from their parents through their genes...

- **Gene therapy**
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases...

- **Genetic disorders**
  Genetic disorder is caused by an altered or faulty gene or set of genes...

- **Genetic services in Victoria**
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare...

**Genetic testing**

- **DNA profiling**
  DNA profiling is a way of establishing identity and is used in a variety of ways...

- **Egg freezing**
  You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances...

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- **Genetic testing for inherited cancer**
  A predisposition to certain cancers can be inherited via altered genes...

- **Newborn bloodspot screening**
  Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions...
Pregnancy tests – chorionic villus sampling

Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities.

Pregnancy tests - maternal serum screening

Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis.

Related Information

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  Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities...

- Pregnancy tests - ultrasound
  Ultrasound is used during pregnancy to check the baby's development and to help pick up any abnormalities...

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Last updated: August 2014

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