



Screening for Neural Tube Defect, Down syndrome and Trisomy 18

What is screening for Neural Tube Defects, Down syndrome and Trisomy 18?

For most parents, pregnancy ends with the birth of a normal healthy baby. In a small number of pregnancies the baby develops with a serious problem. Neural Tube Defects, Down syndrome and Trisomy 18 are serious abnormalities that occur early in the development of a baby. It is not known why they happen.

There are tests you can have during your pregnancy, which can show whether or not your baby is at increased risk for one of these problems. You do not have to have these tests and some women choose not to have them. Before deciding if you want these tests, you should understand what the abnormalities are, what the tests can tell you, and what the results might mean for you and your family.

What are Neural Tube Defects, Down syndrome and Trisomy 18?

Neural Tube Defects are serious abnormalities which happen during the development of the brain and spinal cord in about 1 in 500 babies. It is not known what causes them. The two most common types are *anencephaly* and *spina bifida*.

In *anencephaly* there is abnormal development of the baby's brain and skull. Babies with anencephaly usually die soon after birth. In *spina bifida* the baby's spine does not form properly. Babies with spina bifida may have paralysis of the legs, lack of bladder and bowel control, and curvature of the spine. Hydrocephalus (too much fluid around the brain) can also occur.

For more information about spina bifida contact:
The Spina Bifida and Hydrocephalus Association of South Australia, (08) 8366 5900 www.spinabifida.asn.au
Spina Bifida Association of Tasmania (03) 6275 0987.

Down syndrome and Trisomy 18 are chromosome abnormalities. Babies with Down syndrome have an extra chromosome 21 and those with Trisomy 18 have an extra chromosome 18. About 1 in 600 babies has Down syndrome and about 1 in 6000 babies has Trisomy 18.

Children with Down syndrome have varying levels of intellectual disability and a characteristic appearance. They may have medical problems involving their heart, bowel and thyroid gland. Some may have problems with eyesight and hearing. With medical treatment and social support, children with Down syndrome will usually grow up in good health and with a reasonable quality of life.

For more information about Down syndrome contact:
The Down Syndrome Society of South Australia,
(08) 8365 3510 www.downssa.asn.au
Down Syndrome Association of Tasmania (03) 6224 0490.

Trisomy 18 is usually fatal, 90% of affected pregnancies fail before term. Of the 10% reaching term, half will not survive the first week and most will not survive the first year. Children with Trisomy 18 have serious intellectual disability.

What are the tests?

Between 10 and 20 weeks of pregnancy you may be offered screening tests to look for abnormalities in your baby. Check which tests are available through your local hospital/clinic. The screening tests are a *first* step in finding out whether or not your baby *might* have an abnormality. It is important for you to know that these screening tests can *not* give you a definite yes/no answer to the question 'does my baby have a problem?'. They can only show if there is a greater than expected chance (*an increased risk*) the baby *might* have an abnormality.

The tests are a blood test and an ultrasound scan.

The blood test is done on a small sample (5 ml) of your blood. Your doctor will arrange for this to be taken. There are no known dangers to you or your pregnancy in giving this blood sample.

You will need an appointment with the ultrasonologist for the ultrasound scan. Your doctor will arrange this for you. Ultrasound scanning is a way of seeing your baby using sound waves. There are no known dangers to you or your baby in this type of ultrasound scan.

What can the tests tell?

If you are between 10 and 14 weeks pregnant, the blood test and the ultrasound scan used together can show if there is a greater than expected chance (*an increased risk*) that your baby *might* have Down syndrome or Trisomy 18.

If you are between 14 and 20 weeks pregnant, the blood test can show if there is a greater than expected chance (*an increased risk*) that your baby *might* have Down syndrome, Trisomy 18 or a Neural Tube Defect.

About 95% of women who have these screening tests receive a report stating their baby is not at increased risk of having an abnormality. A small number (5% or 1 in 20) will receive a report stating there is *an increased risk* of an abnormality in their baby.

What do 'increased risk' and 'not at increased risk' mean?

If you received a screening test report which says *not at increased risk*, it means there is only a very small chance that your baby has either Down syndrome, Trisomy 18 or a Neural Tube Defect.

Not at increased risk does not guarantee you a perfect baby, but almost all pregnancies screened *not at increased risk* end in the birth of a healthy baby.



Pre-test information for parents

Screening for Neural Tube Defect, Down syndrome and Trisomy 18

A screening test report saying *increased risk* means there is a greater than expected chance that your baby *might* have a severe abnormality. The calculated risks are given on your report. It is important for you to know that, on average, **24 out of 25 (or 96%) of women who receive a screening test result saying there is an increased risk of an abnormality in their baby, go on to have a normal healthy baby.** Only 1 out of 25 women receiving a report saying there is an *increased risk* of an abnormality, will have a baby with an abnormality. If your screening test results show your baby is at *increased risk* of having Down syndrome, Trisomy 18 or a Neural Tube Defect, further tests will be needed to show whether or not this is so.

What are the further tests?

If you are **between 10 and 14 weeks pregnant**, the tests which can give a definite result following a screening report saying *increased risk of Down syndrome or Trisomy 18* are either chorion villus sampling, which can be done between 10 and 14 weeks, or amniocentesis, which can be done after 15 weeks.

If you are **between 14 and 20 weeks pregnant**, the test which can give a definite result follow a screening report saying *increased risk of Neural Tube Defect* is a detailed ultrasound scan. This is most accurate between 18 and 20 weeks. The test which can give a definite result following a screening report saying *increased risk of Down syndrome or Trisomy 18* is amniocentesis which is done between 15 and 20 weeks.

Chorion villus sampling (or CVS) involves inserting a needle into your womb and taking a very small piece of your baby's placenta. The needle is guided by ultrasound so as not to damage your baby. Testing the piece of placenta will show whether or not your baby has Down syndrome, Trisomy 18 or another chromosome abnormality. It usually takes around 2 weeks to get the result.

There is a small risk of miscarriage as a result of CVS (about 1 in 100-200 tests performed).

Amniocentesis also involves inserting a needle into your womb, this time to take a sample of the fluid around your baby. Again the needle is guided by ultrasound so as not to damage your baby.

Testing the fluid will show whether or not your baby has Down syndrome, Trisomy 18 or another chromosome abnormality. It usually takes around 2 weeks to get the result.

There is a small risk of miscarriage as a result of amniocentesis (about 1 in 200 tests performed).

Must you have further testing?

No, you do not have to. It is your choice. Talk to the doctor looking after you during your pregnancy, or contact the South Australian Maternal Serum Antenatal Screening Program on (08) 8161 7285, which can refer you to counselling and support services.

Will the screening tests detect all affected pregnancies?

No, they will not. If done when you are between 11 and 13 weeks pregnant, the ultrasound and blood tests together will detect around 85-90% of all babies affected with Down syndrome or Trisomy 18. If done when you are between 15 and 20 weeks pregnant, blood testing and ultrasound scanning will detect over 95% of all babies who have a neural tube defect, and about 75% of all babies who have Down syndrome or Trisomy 18. Not all affected babies will be detected.

What happens if your baby is found to have a Neural Tube Defect, Down syndrome or Trisomy 18?

If your unborn baby is found to have one of these conditions you will be given information about the likely effects the abnormality may have on the rest of your pregnancy and on the baby which may be born. You should ask about the medical consequences, what treatment is available, and what support services are available to you and your family.

You will then have to decide whether to continue your pregnancy or to end it early.

Where can I get more information?

More information about antenatal screening and what an *increased risk* result may mean, can be obtained from the doctor or midwife who is looking after you during your pregnancy

or

the South Australian Maternal Serum Antenatal Screening Program,

(08) 8161 7285

samsas.program@health.sa.gov.au

www.wch.sa.gov.au/samsas.html

The figures quoted here are from the South Australian Maternal Serum Antenatal Screening (SAMSAS) Program, SA Pathology, operating at the Women's & Children's Hospital, Adelaide, South Australia. They do not apply to other maternal serum testing centres. (Revised October 2008)