Pre-test information for parents about

Screening your baby for Neural Tube Defects and Down syndrome

from the South Australian Maternal Serum Antenatal Screening (SAMSAS) Programme

The figures quoted here are from the South Australian/Tasmanian Maternal Serum Antenatal Screening (SAMSAS/TAMSAS) Programme, operating in the Department of Genetic Medicine of the Women’s and Children’s Hospital, Adelaide, South Australia. They do not apply to other maternal serum testing centres. If consumers have results from another centre for maternal serum screening, they are advised to check with that centre for the interpretations relating to their results. (Published September 2000).
**What is screening for Neural Tube Defects and Down syndrome?**

For most parents, pregnancy ends with the birth of a normal healthy baby. In a small number of pregnancies the baby may develop with a severe problem.

Down syndrome and Neural Tube Defects are two of the abnormalities which can occur in the early 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 has one of these problems. You do not have to have these tests. Not all women choose 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 and Down syndrome?**

**Neural tube defects** are serious abnormalities which occur in 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 forms 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.

More information about spina bifida can be found from the Spina Bifida and Hydrocephalus Association of South Australia Inc., telephone (08) 8366 5900, website [www.spinabifida.asn.au](http://www.spinabifida.asn.au), and the Spina Bifida Association of Tasmania Inc., telephone (03) 6223 4537.

**Down syndrome** arises when a baby has an extra copy of one of its chromosomes (chromosome 21). This happens in about 1 in 660 babies. It is not known why it happens.
Children with Down syndrome usually have a characteristic appearance. They have varying levels of intellectual disability. They may also have one or more medical problems involving their bones, heart, bowel and thyroid gland. Some may have poor sight and hearing as well. With medical treatment and social support, children with Down syndrome may grow up in good health and with a reasonable quality of life.

More information about Down syndrome can be found the Down Syndrome Society of South Australia Inc., telephone (08) 8365 3510, website www.downssa.mtx.net, and the Down Syndrome Association of Tasmania Inc., telephone (03) 6224 0490.

**What are the tests?**

Some time when you are between 8 and 20 weeks pregnant you may be offered some screening tests to check for Down syndrome and neural tube defects in your baby. The screening tests are a *first* step in finding out whether or not your 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 specimen.

For the ultrasound scan you will need an appointment with an ultrasonologist, which your doctor will arrange for you. Ultrasound scanning is a way of seeing your baby using sound waves. There are currently no known dangers to either you or your baby in an ultrasound of this type.
What can the tests tell?

If done in early pregnancy (11-13 weeks) the blood test and the ultrasound scan together can give parents some forewarning if their developing baby might have Down syndrome. If done later in pregnancy (15-20 weeks) the blood test can give forewarning if the baby might have either Down syndrome or a neural tube defect.

Because they are screening tests they can not give a definite yes/no answer to the question ‘does my baby have a problem?’’. But they can show if there is a greater than expected chance (an increased risk) the baby might have an abnormality.

Most women who have the screening tests receive a report stating there is not at increased risk of an abnormality in their baby. A small number 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 receive a screening test report which says not at increased risk, it means there is only a very small chance (about 1 in 2500) that your baby has either Down syndrome or a neural tube defect. It does not guarantee you a perfect baby, but almost all pregnancies screened not at increased risk end in the birth of a healthy baby.

A screening test report saying increased risk means there is a chance (about 1 in 25) that your baby might have a severe abnormality. 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, will in fact go on to have a normal healthy baby. But in 1 out of 25 women receiving a report saying there is an increased risk of an abnormality, their baby will have an abnormality.

If your screening test results show your baby is at increased risk of having an abnormality, only further tests will show whether or not this is so.
**What is the further testing?**

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 done when you are around 18 weeks pregnant.

The tests which can give a definite result follow a screening report saying *increased risk of Down syndrome* are either chorionic villus sampling which is done early in pregnancy, or amniocentesis which is done later in pregnancy.

**Chorionic villus sampling** (or CVS) is usually performed when you are around 11 weeks pregnant. It involves inserting a needle into your womb and taking a very small sample of your baby’s placenta. The needle is guided under ultrasound scan so as not to damage your baby. Testing the placental tissue will show whether or not your baby has Down syndrome or any other chromosome abnormality. This testing takes around 2 weeks to get the result.

There is a small risk that there will be damage to your pregnancy as a result of the CVS, which may cause a miscarriage (about 1 in 100 tests performed).

**Amniocentesis** is usually performed around 16 weeks gestation. This test also involves inserting a needle into your womb, this time to take a sample of the fluid surrounding your baby. Again the needle is guided under ultrasound scan so as not to damage your baby. Testing the fluid will show whether or not your baby has Down syndrome or any other chromosome abnormality. This testing takes around 2 weeks to get the result.

There is a small risk that there will be damage to your pregnancy as a result of the amniocentesis, which may cause a miscarriage (about 1 in 200 tests performed).

**Will the screening tests detect all abnormalities in your baby?**

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% of all babies affected with Down syndrome. 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. Not all affected babies will be detected.
Must you have further testing?

No, you do not have to. It is your choice. Discuss this with the doctor looking after you during your pregnancy, or contact the South Australian Maternal Serum Antenatal Screening Programme on (08) 8161 7285, who will be able to refer you to counselling and support services.

What happens if your baby is found to have a neural tube defect or Down syndrome?

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 advice about what the medical consequences are likely to be, 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 who is looking after you during your pregnancy,

or

- the South Australian Maternal Serum Antenatal Screening Programme, telephone (08) 8161 7285,