



Government of South Australia

Children, Youth and Women's
Health Service



Women's
& Children's
Hospital



Pre-test information for parents about

Screening for Neural Tube Defects and Down syndrome

Information from the South Australian Maternal Serum Antenatal
Screening (SAMSAS) Programme,
Department of Genetic Medicine,
Women's and Children's Hospital, North Adelaide 5006

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.

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 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 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.

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 in 1 out of 25 women receiving a report saying there is an *increased risk* of an abnormality, will their baby actually have an abnormality.

If your screening test results show your baby is at *increased risk* of having Down syndrome or a Neural Tube Defect, only further tests will show whether or not this is so.

What are the further tests?

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 done when you are around 18 weeks pregnant.

The tests which can give a definite result following a screening report saying *increased risk of Down syndrome* are either **chorion villus sampling** which is done when you are around 12 weeks pregnant, or **amniocentesis** which is done when you are around 16 weeks pregnant.

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 under ultrasound so as not to damage your baby. Testing the piece of placenta will show whether or not your baby has Down syndrome or any other

chromosome abnormality. This testing takes around 2 weeks to get the results.

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 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 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 also 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).

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.

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

Tasmania Inc., *telephone* (03) 6275 0987 can provide you with more information about spina bifida.

Down syndrome arises when a baby has an extra copy of one of its chromosomes (chromosome 21). This happens in about 1 in 600 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.

The Down Syndrome Society of South Australia Inc., *telephone* (08) 8365 3510, *website* www.downssa.asn.au, and the Down Syndrome Association of Tasmania Inc., *telephone* (03) 6224 0490, can provide you with more information about Down syndrome.

What are the tests?

Some time when you are between 10 and 20 weeks pregnant you may be offered some screening tests to check 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 a **ultrasound scan**.

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. Neural Tube Defects and Down syndrome are two of the severe 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.

The Spina Bifida and Hydrocephalus Association of South Australia Inc., *telephone* (08) 8366 5900, *website* www.spinabifida.asn.au, and the Spina Bifida Association of

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.

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
email cywhs.samsas@cywhs.sa.gov.au
website www.wch.sa.gov.au/samsas.html

The figures quoted here are from the South Australian Maternal Serum Antenatal Screening (SAMSAS) 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 2006).