# PRE-TEST INFORMATION FOR PARENTS

## Screening for Neural Tube Defects, Down Syndrome & Trisomy 18

SA PATHOLOGY

### What is screening for neural tube defects, Down Syndrome and Trisomy 18?

Most pregnancies result in the birth of a healthy baby. However, in a small number of pregnancies, the baby has serious problems. Neural tube defects, Down syndrome and Trisomy 18 are serious abnormalities that occur early in the development of a baby.

Screening tests can determine if your baby is at increased risk for these particular conditions, so you can decide if you would like to have further diagnostic testing.

Screening tests do *not* detect all cases of these conditions or the many other abnormalities that occur in up to 2.5% of pregnancies, so a "low risk" result does not guarantee your baby will have no abnormalities. Before deciding if you want these tests, you should understand what the abnormalities are, what the tests can tell you, and what the results may mean for you and your family.

### What are neural tube defects, Down Syndrome and Trisomy 18?

**Neural tube defects** are serious abnormalities which occur during early development of about 1 in 500 babies. It is not known what causes neural tube defects, but folic acid supplementation before conception and through the early weeks of a pregnancy lowers the chance.

The two most common types are *anencephaly* and *spina bifida*. In anencephaly the baby's brain and skull fails to develop normally. Babies with anencephaly usually die soon after birth.

In spina bifida the baby's spine does not form properly. How serious the problem is depends upon on the size of the abnormality and where it occurs on the baby's back. Babies with severe 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 Association of Tasmania 1300 135 513

**Down syndrome** and Trisomy 18 are chromosome abnormalities. Babies with Down syndrome have an extra chromosome 21 and babies born with Trisomy 18 have an extra chromosome 18. Down syndrome occurs in about 1 in 600 babies, while Trisomy 18 occurs in about 1 in 6000 babies.

Children with Down syndrome have varying levels of learning and 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 have a good quality of life.

For more information about Down syndrome contact:

The Down Syndrome Society of South Australia (08) 8369 1122 www.downssa.asn.au

The Down Syndrome Association of Tasmania 1300 592 050 www.downsyndrometasmania.org.au

The Down Syndrome Association of the Northern Territory (08) 8985 6222 www.downsyndroment.com.au

**Trisomy 18** is usually fatal, children with Trisomy 18 have short lives with serious intellectual disability and other health problems. In fact 90% of Trisomy 18 affected pregnancies do not reach the end of the pregnancy.

### What are the tests?

Screening tests for these abnormalities are available between 9 weeks and 20 weeks 6 days. They do not give a definite yes/no answer and can only *determine if your baby has a greater than expected chance* (increased risk) of one of these abnormalities.

*The tests usually combine a blood test result with an ultrasound scan*. The blood test requires a small 5ml blood collection, which your doctor can arrange. There is no risk to your pregnancy from giving this blood sample.

Your doctor can organise a scan that uses sound waves to look at your baby. You will need an appointment for your ultrasound scan. There is no risk to your pregnancy from this scan.

### What can these tests tell?

The *First Trimester* screen is performed between 9 weeks and 13 weeks 6 days. The ultrasound test is usually performed between 10 and 13 weeks. Usually the information from *both* the blood test and the ultrasound scan are used to test for an increased risk of Down syndrome and Trisomy 18.

The Second Trimester screen is provided between 14 weeks and 20 weeks 6 days. The blood test can be

used to show if the baby is at increased risk of Down syndrome, Trisomy 18 and neural tube defects.

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

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

If the screening test report says "*not at increased risk*", it means there is only a *very small chance* that your baby has Down syndrome, Trisomy 18 or a neural tube defect. Remember that a "*not at increased risk*" result cannot, however, guarantee a baby born without these abnormalities.

A screening report which says "*increased risk*" means there is a greater than expected chance that your baby may have Down syndrome, Trisomy 18 or neural tube defect. The size of the chance is provided in the report. It is important to know that 24 out of 25 women receiving an "increased risk" report will go on to have a healthy baby.

If your report indicates that your pregnancy is at increased risk of having Down syndrome, Trisomy 18 or a neural tube defect, you will need to discuss with your health care provider your options, then decide whether to have further tests to show if the baby has the disorder.

## What are the further tests?

If you receive a report saying that your baby is at increased risk of Down syndrome or Trisomy 18, the further test will depend on the stage of your pregnancy. Chorionic villous sampling (or CVS) is performed before 13 weeks and amniocentesis after 15 weeks.

If the increased risk report is for neural tube defect, the further test is a detailed ultrasound scan.

Chorionic 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 the baby. Testing the piece of placenta will show whether or not your baby has Down syndrome, Trisomy 18 or other chromosome abnormality. It takes around 2 weeks to get the result. There is a small risk of miscarriage after CVS (about 1 in 150 tests performed).

Amniocentesis involves inserting a needle into your womb and taking a sample of the fluid around your baby. The needle is guided by ultrasound so as not to damage the baby. Testing the fluid will show whether or not your baby has Down syndrome Trisomy 18 or other chromosome abnormality. It takes around 2 weeks to get the result. There is a small risk of miscarriage after amniocentesis, (about 1 in 200 tests performed).

## Do you have to have further tests?

No, you do not have to have further diagnostic tests. Talk to the doctor or midwife looking after you during your pregnancy, or contact the South Australian Maternal Serum Antenatal Screening programme on (08) 8161 7285, for information on counselling and support services.

## Will the screening tests detect all affected pregnancies?

No, they will not. Testing done within the first trimester screening window of 10 to 14 weeks using the combination of blood test and ultrasound scan will detect around 85-90% of babies with Down syndrome or Trisomy 18. Testing between 14 to 21 weeks will detect approximately 95% of neural tube defects and 75% of Down syndrome and Trisomy 18 cases.

It is important to remember that not all babies affected with Down syndrome, neural tube defects and Trisomy 18 will be detected, and other abnormalities may not be detected during routine screening.

## What happens if your baby is found to have neural tube defect, Down syndrome or Trisomy 18?

If your unborn baby is found to have one of these abnormalities you will be given information about what having this disorder might mean for the baby and for the pregnancy. You should ask advice about the medical problems your baby might have if it is born, 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 about what an *increased risk* result means, 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, at the Women's & Children's Hospital, Adelaide, South Australia. They do not apply to other maternal serum testing centres.*