Healthscope PATHOLOGY

Screening for Neural Tube Defects & Down Syndrome

Information for Parents Prior to Antenatal Risk Assessment

DOWN SYNDROME IS THE MOST COMMON CHROMOSOMAL DISORDER IN AUSTRALIA

It occurs when there are three copies of chromosome 21 (Trisomy 21) rather than the usual pair in each cell. This mistake occurs in about 1 in 600 babies and leads to a wide range of medical problems. It is not known why this happens but the risk of occurrence increases with increasing maternal age. Despite this, the majority of Down Syndrome affected pregnancies occur in younger women.

Children with Down Syndrome usually have a characteristic appearance. Their medical problems include mild to severe intellectual disability, heart defects, bone, bowel, blood and thyroid gland problems.

With parental, medical and social support, many of the problems can be recognised early and managed effectively. Many children with Down Syndrome can thus expect to enjoy a reasonable quality of life.

NEURAL TUBE DEFECTS OCCUR DURING THE DEVELOPMENT OF THE SPINAL CORD AND BRAIN.

These serious abnormalities occur in approximately 1 in 500 babies. Neural Tube Defects (NTDs) are strongly associated with folate deficiency in pregnancy. The developing nervous system requires increased amounts of this vitamin. Some pregnant women require much larger than usual amounts of folate to provide sufficient amounts to their developing baby.

The two most common forms of NTDs are Spina Bifida and Anencephaly. Babies with Anencephaly usually die soon after birth. In Spina Bifida the baby's spine has not developed properly. The defect itself may be very mild through to severe and life threatening with the development of meningitis. Babies with Spina Bifida can have a wide range of neurological problems such as paralysis or weakness of the legs, lack of bowel and bladder control, and curvature of the spine.

WHAT IS THIS BLOOD TEST FOR?

WHY SCREEN?

DO I HAVE TO BE TESTED?

Generally at sometime during the period 10–20 weeks of pregnancy, you will be offered some screening tests to check for abnormalities in your baby. These tests include blood tests and an ultrasound scan. These tests will not harm you or your baby but will provide the first step toward finding out whether your baby might have an abnormality. The screening tests do NOT provide you with a yes/no answer to the question, "Does my baby have a problem?" The tests tell you if your baby is more likely than another baby to have a problem.

If you are **between 10 and 14 weeks pregnant,** the blood test and ultrasound scan combined can indicate if there is a greater than expected risk that your baby might have Down Syndrome.

If you have any questions or require further information, please contact our Customer Service Centre.
1300 453 688

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If you are **between 14 and 20 weeks pregnant,** the blood test alone can indicate whether there is a greater than expected risk that your baby might have Down Syndrome or a Neural Tube Defect.

Approximately 95% of women who elect to have these screening tests will receive a report stating that their baby is not at an increased risk of having one of these abnormalities. Approximately 5% of women (1 in 20 pregnancies) will receive a report stating that their baby is at an increased risk of having an abnormality.

You do NOT have to have these tests. If you elect to have these screening tests and the results indicate your baby is at an increased risk of having Down Syndrome or a Neural Tube Defect, a definite diagnosis can only be made with further testing. You do NOT have to proceed to further investigations. At all stages of this process the choice is yours. We encourage you to discuss your options with the doctor or midwife looking after you and your pregnancy.

WHAT DOES 'INCREASED RISK' AND 'NOT AT INCREASED RISK' MEAN?

WILL I HAVE A NORMAL, HEALTHY BABY?

WILL THE SCREENING DETECT OTHER ABNORMALITIES IN MY BABY?

Note: This is a risk calculation not a diagnostic test.

If you receive a report that states *'not at increased risk'*, 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 your baby will be born healthy without either of these two serious defects. However, almost all pregnancies screened as *'not at increased risk'* end in the birth of a healthy baby.

If you receive a report that states *'increased risk'*, there is a greater than expected chance that your baby could have a serious abnormality. A calculated numerical risk is present in your report. Please note that approximately 24 out of 25 (96%) of women who receive an *'increased risk'* screening report will have a normal, healthy baby.

In other words, in only 1 in 25 pregnancies at increased risk of a foetal abnormality, will the baby actually have the abnormality.

These screening tests will not detect all abnormalities.

First trimester testing will identify around 85% of all babies affected by Down Syndrome. A second trimester screening will detect approximately 75% of all babies affected by Down Syndrome and 95% of all babies with a Neural Tube Defect, in conjunction with an ultrasound scan.

WHERE CAN I GET MORE INFORMATION?

For more information on any of these topics, you may wish to talk with the doctor or midwife looking after your pregnancy or contact either of the following organisations in your state:

- The Spina Bifida and Hydrocephalus Association
- The Down Syndrome Society

Providing services since 1936, Healthscope Pathology operates throughout Australia, New Zealand, Singapore and Malaysia.

Healthscope Pathology has purpose built, state-of-the-art service and molecular research laboratories which ensure the highest standard of testing.

focused on you

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