2005 Edition

A Guide to the Screening Test for Spina Bifida and Down's Syndrome

A blood test you can choose to have during your pregnancy

Happily, most babies are perfectly healthy when they are born. A small number however are born with conditions such as spina bifida or Down's syndrome. You may choose to have a test during pregnancy which can help to detect these rare conditions. The information in this leaflet may help you make this choice.

What is Spina Bifida?

Spina bifida and anencephaly are the two main types of condition together known as neural tube defects. About 1 in every 500 pregnancies is affected.

Babies with spina bifida have an opening in the bones of the spine, and the nerves to the lower part of the body are damaged. This can result in problems with walking and bowel and bladder control. Sometimes there is also a learning difficulty due to hydrocephalus (an accumulation of fluid in the brain) which often accompanies spina bifida.

There is variation in the level of disability caused and many people with spina bifida are able to lead fulfilling lives.

In babies with anencephaly the skull and brain are not properly formed. These babies generally die before or very soon after they are born.

Further information on spina bifida can be obtained from:

- **Scottish Spina Bifida Association**
  Tel: 0131 332 0743
  www.ssba.org.uk

What is Down's Syndrome?

People with Down's syndrome (sometimes referred to as 'trisomy 21') have an extra chromosome, having three copies of chromosome 21 instead of two.

Chromosomes carry genes which pass certain characteristics from parents to their children. Problems can occur when there are too many chromosomes.

Children with Down's syndrome will have learning difficulties and will need special help with their education. Many people with Down's syndrome enjoy a healthy life, but some have health problems such as heart defects or thyroid problems.

Older mothers are more likely to have a baby with Down's syndrome. For example Down's syndrome occurs:

- once in every 1500 births to women aged 20 years
- once in every 900 births to women aged 30 years
- once in every 100 births to women aged 40 years.

Further information on Down's syndrome can be obtained from:

- **Down's Syndrome Scotland**
  Tel: 0131 313 4225
  www.dsscotland.org.uk

Are Any Other Types of Chromosome Abnormality Detected?

Trisomy 18 or Edwards' Syndrome can also be detected by the screening test. Babies with Edwards' Syndrome have three copies of chromosome 18 instead of two. They have a short life expectancy and require specialised nursing. The condition affects about 1 in 6000 pregnancies and is more common in older mothers.

Further information on Edwards' Syndrome can be obtained from:

- **SOFT UK**
  Tel: 0121 351 3122
  www.soft.org.uk

Who Should Have the Screening Test?

Women choose whether or not to have the screening test. Your midwife will discuss the test with you and you will be asked to sign a consent form. It is a personal decision for you and your partner to make after you have had time to think about it. All pregnant women irrespective of age can have the test. There is no charge for this test.

You do not have to have this screening test or a follow-up test if you do not wish to. You can opt out of the process at any stage. It is your choice and whatever you decide will not affect the rest of your antenatal care in any way.

How is Testing Carried Out?

There are two stages to testing. The first stage is a screening test, offered to all. Depending on the results of this test some women (about 1 in 10) will be offered a second follow-up test.

For the screening test, a small blood sample is taken from your arm at around 15–16 weeks of pregnancy and sent to the laboratory. There the levels of two naturally occurring pregnancy hormones called AFP (alphafetoprotein) and HCG (human chorionic gonadotrophin) are measured. Within two weeks of having the screening test you should receive a letter telling you the results — whether you have a low chance or a higher chance of your baby having spina bifida or Down's syndrome.

If the screening test results are 'low chance' there is only a very small chance that your baby will have spina bifida or Down's syndrome and no further testing is indicated. 9 out of 10 women will be given a low chance result.

Several factors can affect the screening result. These include how far along the pregnancy is, the mother's weight and IVF pregnancies (especially using donor eggs or frozen embryo).

Will the Screening Test Detect all Problems?

No. The screening test can identify about 4 out of 5 spina bifida pregnancies and about 2 out of 3 Down's syndrome pregnancies. Occasionally, other rarer conditions are picked up by the screening test, but mainly it is only able to identify pregnancies where there is a higher chance of spina bifida or Down's syndrome.

What Happens if the Screening Result shows a Higher Chance of a Problem?

If the screening results are 'higher chance', this indicates that there is an increased chance that the baby has either a neural tube defect or Down's syndrome (it is very unlikely that the pregnancy will be at increased risk for both types of abnormality) and further investigation will be offered.

You will be asked to return to the hospital where the results will be discussed with you and your questions answered. The follow-up tests which you could choose to have will be explained.

HIGHER CHANCE OF SPINA BIFIDA

This screening result means that the AFP level was high and that there is some increase in the chance of spina bifida.

- About 1 woman in 20 will have this result.
- This does not mean that there is a problem.
- Most women with these results will have normal healthy babies.

The most common causes of a high AFP result are:

- the pregnancy was further along than was thought
- there are twins
- normal variations between women in the level of AFP in the blood.

A follow-up test, usually a detailed ultrasound scan, is offered which can show whether an abnormality is present or not. It is your choice whether or not to have a detailed ultrasound scan.

HIGHER CHANCE OF DOWN'S SYNDROME

This screening result means that the combination of AFP and HCG results together with a woman's age show a higher chance of Down's syndrome.

- About 1 in 20 women will have this type of screening result.
- This result does not mean that there is a problem
- Most women with these results will have normal healthy babies.

The most common causes of this type of screening result are:

- the pregnancy is not as far along as was thought
- normal variations between women in the level of AFP and HCG in the blood.

A follow-up test, usually *amniocentesis* is offered to find out whether the baby has Down's syndrome. It is your choice whether or not to have an amniocentesis.

What are Follow-up Tests?

These tests are used where the screening test result indicates that further investigation of the pregnancy should be considered. They include:

Ultrasound (also known as 'Scanning'): Almost all women will have a routine ultrasound scan at some stage in their pregnancy. Sound waves are used to make a picture of the baby on a special TV screen. It can show the age and position of the baby and also if there are twins.

A more detailed scan can also detect certain conditions such as spina bifida and hydrocephalus. There are no known harmful effects to the baby from ultrasound.

Amniocentesis: This follow-up test is used where the screening result shows an increased chance of Down's syndrome. For most women amniocentesis takes only a few minutes and causes little discomfort.

First ultrasound is used to find the baby's position in the womb. A needle is then inserted into the fluid (called amniotic fluid) surrounding the baby. The needle is carefully positioned to avoid the baby. A small amount of fluid is removed and sent to the laboratory where it is used to examine the baby's chromosomes.

The results of the amniocentesis test take up to three weeks but increasingly a newer test is being offered which can shorten the time to a few days. These results will tell whether the baby has Down's syndrome or normal chromosomes. There is a 1 in 100 risk of miscarriage following amniocentesis.

What Happens if the Follow-up Test Finds a Problem?

In most cases follow-up testing finds a healthy baby. If spina bifida or Down's syndrome is found this will be discussed fully with you and your partner and you will be able to choose what you feel is best for you. Some couples may decide to continue with the pregnancy while others will feel that termination of pregnancy is the best course of action.

There should be no pressure to influence you in your decision. The hospital staff will provide help and support whatever you decide. Further help and family support can also be obtained from the Scottish Spina Bifida Association or the Scottish Down's Syndrome Association.

What Happens to the Samples and Information?

The test results are confidential and only health care professionals closely involved in your care will be informed of your results. No one else will be told about your results without your consent.

Samples are retained, so that if necessary tests can be checked. These samples may be used anonymously for purposes such as monitoring and developing new tests. Occasionally it is necessary to use identifiable specimens in which case your permission would always be sought.

Published by Health Scotland, Woodburn House,
Canaan Lane, Edinburgh EH10 4SG.

© NHS Health Scotland, 2002, 2004.
ISBN: 1-84485-129-X

Photography: Steve Glyn Jones

FURTHER INFORMATION ON ALL THESE TESTS IS AVAILABLE FROM YOUR MIDWIFE, OTHER HOSPITAL STAFF OR GP.

You can also obtain additional information from:

Antenatal Results and Choices (ARC)
Tel: 0207 631 0285
email: info@arc-uk.org
www.arc-uk.org

Contact a Family Directory (Scotland)
Tel: 0131 475 2608
email: scotland@cafamily.org.uk
www.cafamily.org.uk

This information is also available in alternative languages and formats. Please contact Health Scotland or your local NHS Board for further details.

هذه المعلومات متوفرة أيضاً بلغات وتنسيقات أخرى.
الرجاء الاتصال بالصحة الاسكتلندية (Health Scotland) أو مجلس خدمة الصحة الوطنية (NHS Board) المحلية لديك للحصول على المزيد من التفاصيل.

這些訊息還以其他語言和格式提供。
有關更多詳細資訊，請與蘇格蘭衛生部 (Health Scotland) 或者您當地的全民健康委員會 (NHS Board) 聯絡。

یہ معلومات متبادل زبانوں اور شکلوں میں بھی دستیاب ہے۔ مزید تفصیلات کے لئے براہ مہربانی
ہیلتھ سکاٹ لینڈ (Health Scotland) یا اپنے مقامی این ایچ ایس بورڈ (NHS Board) سے رابطہ کریں۔

Details of organisations and websites are included for your information. Inclusion in, or exclusion from this information leaflet does not imply the endorsement or otherwise of an organisation by the Scottish Executive Health Department.

NHS SCOTLAND

2058 2/2005