Antenatal Screening and Diagnosis for Down’s Syndrome

Information for Patients

Maternity Services
York Teaching Hospital NHS Foundation Trust

Working together for the communities of York, Scarborough, Bridlington, Malton, Whitby, Selby and Easingwold
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If you require this information in an alternative format, for example: Braille, audio, large print or another language then please inform staff at your consultation, or ring the number for further information given on the back of this leaflet.
Introduction

It is recommended that all pregnant women should be offered a range of screening tests. Some of these tests provide specific information such as the estimated date of delivery or whether you are expecting twins. Some look at the general well being of the baby and may indicate which babies may be at a greater risk of having certain conditions such as Down’s syndrome. You can choose whether you wish to have these screening tests.

All women, whatever their age, have a small risk of delivering a baby with Down’s syndrome, though it increases with the age of the mother. It is caused by the presence of an extra chromosome in the genetic material present in each cell of the baby. Each cell contains 47 chromosomes instead of the usual 46. These chromosomes are vital for the normal function of every part of the body, with the extra chromosome in Down’s syndrome leading to physical and mental disability of varying degrees.

This booklet describes the range of optional screening and diagnostic tests available at our hospitals so that you can make an informed decision about whether you would like your baby screening for Down’s syndrome.

Choosing to have Down’s syndrome Screening is an important individual choice for you and your baby. You need to think carefully about your views about Down’s syndrome and your feelings about screening before you choose to have the test.
Risk of Down’s syndrome at term according to maternal age

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<th>Age</th>
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<tbody>
<tr>
<td>20</td>
<td>1 in 1455</td>
<td>37</td>
<td>1 in 238</td>
</tr>
<tr>
<td>25</td>
<td>1 in 1288</td>
<td>38</td>
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<tr>
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<td>1 in 874</td>
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<td>1 in 40</td>
</tr>
<tr>
<td>36</td>
<td>1 in 300</td>
<td>45</td>
<td>1 in 28</td>
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There are two categories of tests: screening tests and diagnostic tests.

**A screening test** will estimate if a woman is in a higher risk or lower risk group for having a baby with Down’s syndrome. It will not give a definite answer, but poses no risk to the baby.

**A diagnostic test** will tell you if the baby has Down’s syndrome to about 99.9% accuracy, but the test does have a risk of miscarriage.

Women who have had a screening test result putting them at higher risk of having a baby with Down’s syndrome will be offered a diagnostic test to determine if the baby is really affected.
Screening Tests

These tests only show the risk or chance of your baby having Down’s syndrome, but they do not confirm or diagnose this. There is no risk to your baby by having a screening test. A lower risk result does not guarantee a baby does not have Down’s syndrome and a higher risk result does not confirm the baby has Down’s syndrome. Screening tests are used to identify which women are at a higher risk of having a baby with Down’s syndrome because we do not have a risk-free diagnostic test.

Screening tests offered in our hospitals are:

- Early Pregnancy Scan, (dating scan)
- Nuchal Translucency scan combined test, (first trimester Down’s screening test)
- Quadruple blood test, (second trimester Down’s screening test)
- Mid Pregnancy Scan, (anomaly scan)

The Nuchal Translucency scan, combined test and the quadruple tests are the only tests offered on the NHS to screen for Down's syndrome.
Early Pregnancy Dating Scan

This is ideally performed between 10 – 14 weeks, but the accuracy is not greatly affected if done slightly outside this range.

This scan will be used to date your pregnancy and calculate the estimated date of delivery of your baby. This is more accurate than using period dates. The scan is also used to count the number of babies allowing early diagnosis of multiple pregnancies such as twins, and to check that the pregnancy appears to be progressing normally. In the rare event of a problem being suspected this would be discussed with you after the scan. You may be offered further investigations before a definite diagnosis can be given, but you always have a choice as to whether to have these investigations.

If you would like the Nuchal Translucency scan (Down’s syndrome screening test), this will be done at the same time as the dating scan, provided that your pregnancy is at the correct gestation.
Nuchal Translucency Scan Combined Screening Test for Down’s syndrome. (Performed at 11 weeks, two days to 14 weeks, one day)

This is a screening test for Down’s syndrome incorporating a scan and a blood test. If you choose to have this test, it will be performed at the same time as your dating scan.

This is an ultrasound scan in which particular attention is paid to a layer of fluid beneath the skin on the back of a baby’s neck, the nuchal translucency (see picture). In babies with chromosome problems such as Down’s syndrome this may appear to be thicker than average. A blood test is taken at the same time as the scan measuring two substances in the mother’s blood that are produced by the pregnancy. These substances are sometimes altered in babies with Down’s syndrome. Combining the result of this scan and blood test allows us to calculate the risk or chance of the baby having Down’s syndrome.

The nuchal translucency scan combined test will identify 85% to 90% of Down’s syndrome babies in the higher risk category. It cannot pick up 100% of Down’s syndrome babies and therefore a lower risk result does not guarantee a baby does not have Down’s syndrome.
The result is presented as a statistic. A result of 1 in 150 or any number less than this is classified as a higher risk. A result of 1 in 151 or any number greater than this is classified as a lower risk. The smaller the number, the higher the risk.

A woman receiving a higher risk result will be offered a diagnostic test, but you can choose whether you would like to proceed to this test.
Quadruple Blood Test for Down’s syndrome (Performed at 14 weeks, two days to 20 weeks)

If you have requested Down’s syndrome screening, but your pregnancy measurement is too large to perform the NT scan, which is usually beyond 14 weeks, one day, you will be offered a quadruple test. This is a blood test which measures four substances in the mother’s blood, produced by the pregnancy, which are sometimes altered in babies with Down’s syndrome.

An ultrasound dating scan is performed before the blood test to accurately date the pregnancy unless you have already had a scan for some other reason.

The quadruple test will identify as higher risk more than 75% of babies with Down’s syndrome. A lower risk result cannot guarantee a baby does not have Down’s syndrome because the test does not pick up 100% of affected babies. The result is presented as a statistic with a higher risk result being 1:150 or any number smaller than that. A result of 1:151 or any number larger than that is classified as a lower risk. If your result falls into the higher risk range you will be offered a diagnostic test, but you can choose whether you wish to proceed to this test.

If you have already had a nuchal translucency combined test, a quadruple test will not be offered.
Results from NT Combined Test or Quadruple Test

A lower risk result from your screening test will be sent to you by letter and may take up to two to three weeks.

Should your result be higher risk we will inform you by telephone to allow opportunity for discussion, so this will be received up to one to two weeks after the test. If you prefer to receive the result by other means please ask to speak to the screening co-ordinator when you attend for your scan.

If you have not received your screening result after three weeks, please contact the Screening Co-ordinator on telephone number 01904 725347.

Mid Pregnancy Anomaly Scan (Performed at 18 weeks to 20 weeks, six days)

This is a screening test that checks for possible physical problems with your baby. It also gives information on the position of the placenta (afterbirth). Scans are not guaranteed to identify all problems. Sometimes we may suspect a problem, but cannot say for certain without further tests. Occasionally babies may be born with abnormalities that were not spotted on a scan.
Diagnostic Tests

None of the screening tests will pick up 100% of Down’s syndrome pregnancies. For those who wish a test to definitely detect Down’s syndrome, a diagnostic test is needed. These are usually offered because you have received a higher risk screening result or because there is a suspected problem, identified on one of your scans. A previous pregnancy in which the screening result was higher risk, but the baby was healthy, would have no effect on this pregnancy. The two diagnostic tests available are Amniocentesis (Amnio) and Chorionic Villus Sampling (CVS). These tests allow us to look at all the baby’s chromosomes. Therefore, they can identify other chromosome problems in addition to Down’s syndrome.

There is a risk of miscarriage with these tests
Chorionic Villus Sampling from 12 weeks onwards

This test is not carried out in our hospitals, but requires that we refer you to a different NHS Trust.

It involves obtaining a very small amount of the cells from the placenta (afterbirth). The placenta develops from the baby at the very start of pregnancy and usually has identical chromosomes. A needle is passed through the mother’s skin and directed into the placenta, under ultrasound guidance. A tiny fraction of tissue is sucked through and sent to a regional laboratory. Local anaesthetic is used for this procedure.
The result is usually available within 48 hours and is extremely accurate for Down’s syndrome, so further management could be based on this result. Rarely the cells do not give a result or they may be unable to reach the placenta to gain a sample, so further testing may be needed.

Compared with amniocentesis, CVS can be performed earlier and therefore results are obtained earlier in the pregnancy. However, the miscarriage risk may be slightly greater, with around 1% (one in 100) of women miscarrying.
Amniocentesis from 15 weeks onwards

This test is available at our hospitals.

A sample of the amniotic fluid surrounding the baby is obtained by passing a fine needle through the mother’s skin into the womb. This is performed using an ultrasound scan to guide the needle into the fluid. No harm is done if the baby moves against the needle whilst the sample is being taken. Local anaesthetic is not used as the needle for amniocentesis is around the same size as the local anaesthetic needle.
This fluid, which contains cells from the baby, is sent to regional laboratory in Leeds and a result is usually available within 48 hours. Rarely technical difficulties mean it is not possible to obtain a sample, or the cells do not give a result, therefore a repeat sample may be needed.

The miscarriage risk is around 0.5 – 1% (1 in 100 – 200) and it is impossible to predict which pregnancies might be lost.

It is stressed that neither amniocentesis nor CVS is an infallible test. There have been rare cases of the mother’s cells being tested rather than the baby’s.

Similarly, a normal result does not guarantee a normal baby, as there are many other causes of disability, which are not detected by these tests.
Remember

• Screening tests only show the risk of Down’s syndrome. A negative or lower risk result does not guarantee that the baby will not have Down’s syndrome just as a positive, or higher risk result does not mean that the baby has Down’s syndrome.

• Diagnostic tests can confirm Down’s syndrome almost 100% accurately, but they do carry a risk of miscarriage.

• Whether you choose to accept or decline testing you will be fully supported in your decision.
For more information, you can contact:

Moira Brown, Antenatal Screening Co-ordinator for York Teaching Hospital NHS Foundation Trust

Camilla Picknett, Support Screening Midwife (York)

Jolene Boyce, Support Screening Midwife (Scarborough)

Telephone  The York Hospital:  (01904) 725347
                      Scarborough Hospital: (01723) 236308

Alternatively, refer to the leaflet: “Screening tests for you and your baby” provided by your community midwife.

Other useful organisations are:

Antenatal Results and Choices, Tel: 020 7631 0285 (www.arc-uk.org)

Down’s Syndrome Association, Tel: 0845 230 0372 (www.downs-syndrome.org.uk)
Supervisors of Midwives

Supervisors of Midwives are midwives who have undertaken additional training, whose aim is to give guidance and support to both midwives and women to ensure that the care offered is right for you and is given in the right place, by the right person and that it will benefit you and your baby (NMC 2009).

Supervisors of Midwives are available to discuss any aspect of your maternity care that may be of concern to you. You can contact us by phoning:
York: 01904 631313 or 01904 726004
Scarborough: 01723 368111 or 01723 342124
Please ask for the Supervisor of Midwives ‘on call’.
Teaching, Training and Research

Our Trust is committed to teaching, training and research to support the development of staff and improve health and healthcare in our community. Staff or students in training may attend consultations for this purpose. You can opt-out if you do not want trainees to attend. Staff may also ask you to be involved in our research.

Patient Advice and Liaison Service (PALS)

The York based team can be contacted on 01904 726262, or via email at pals.york@york.nhs.uk

The Scarborough based team can be contacted on 01723 342434, or via email at pals.scarborough@york.nhs.uk

Answer phones are available out of hours.

Would you like to comment on this leaflet?

Meeting the needs and preferences of patients and carers is at the centre of everything we do. We hope that you found this leaflet useful and informative. If you would like to comment on it, please contact Moira Brown, Antenatal Screening Coordinator, The York Hospital, Wigginton Road, York, YO31 8HE or telephone 01904 725347.
Our Commitment to You

Our ultimate objective is to be trusted to deliver safe, effective healthcare to our community. You can find further details on our website: www.york.nhs.uk.

If you require further information please contact Moira Brown, Antenatal Screening Coordinator on telephone number 01904 725347 or contact your Community Midwife or the Maternity Ward Clerk on 01723 342124 (Scarborough) or 01904 726720 (York).