Antenatal fetal anomaly screening and diagnosis

For Down’s, Patau’s and Edwards’ syndromes (T13/18 and T21)

Information for patients, relatives and carers

Maternity Services

For more information, please contact:
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The York Hospital, Wigginton Road, York, YO31 8HE
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or your community midwife

Caring with pride
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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 wellbeing of the baby and may indicate which babies may be at a greater chance of having certain conditions such as Down’s, Patau’s & Edwards’ syndromes. You can choose whether you wish to have these screening tests.

All women, whatever their age, have a small chance of delivering a baby with Down’s, Patau’s or Edwards’ syndrome, though it can increase 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, Patau’s or Edwards’ syndrome leading to physical and learning disabilities 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, Patau’s & Edwards’ syndrome.
What is Downs Syndrome (T21)?

In Down’s syndrome there is an extra copy of chromosome 21. Down’s syndrome is also known as trisomy 21 or T21. Down’s syndrome affects approximately one in 1,000 births.

Babies born with Down’s syndrome will have a learning disability; this means they will find it harder to learn things, including everyday tasks and communication. The degree of learning disability can vary from mild to severe and it is impossible to be able to predict their range.

There are some health problems that are common with people who have Down’s syndrome such as heart conditions, problems with their digestive system, hearing and eyesight. Some of these problems can be serious, but most can be treated. People with Down’s syndrome have a life expectancy of approximately 60 years of age with appropriate healthcare.

People with Down’s syndrome can have a good quality of life. Many can live independently and have jobs with some support from family and other support agencies.

People with Down’s syndrome have almond shaped eyes and other distinguishable facial features. They will inherit features from their parents and so not all Down’s syndrome people look the same.
What are Patau’s & Edwards’ Syndromes?

Babies with Patau’s or Edwards’ syndrome have a wide range of health problems; most of these are very serious, including major brain abnormalities.

Most babies with these syndromes may die before they are born, miscarry, be stillborn or die shortly after birth. Some babies may survive into adulthood, but this is rare.

Patau’s Syndrome (T13)

Babies with Patau’s syndrome have an extra copy of their 13th chromosome, trisomy 13 (T13).

Babies with Patau’s syndrome can have multiple problems including heart, brain, cleft lip & palate, growth, kidneys, eyes and ears and are also unable to walk or stand.

About two babies out of 10,000 births are affected with Patau’s syndrome.

Edwards’ Syndrome (T18)

Edwards’s syndrome affects approximately three babies in 10,000 births.

Babies with Edwards’ syndrome can have heart problems, unusual shaped head and facial features, growth problems and inability to walk or stand.
What should I know about screening or diagnostic tests?

Choosing to have Down’s, Patau’s & Edwards syndrome screening is an important individual choice for you and your baby. You need to think carefully about your views about Down’s, Patau’s & Edwards’ syndrome and your feelings about screening before you choose to have the test.

There are two categories of tests: screening tests and diagnostic tests.

A screening test will estimate if a woman is in a higher chance or lower chance group for having a baby with Down’s, Patau’s & Edwards’ 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, Patau’s & Edwards’ 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 chance of having a baby with Down’s, Patau’s & Edwards’ syndrome will be offered a diagnostic test to determine if the baby is really affected.
What screening tests are available?

These tests only show the chance of your baby having Down’s, Patau’s & Edwards’ syndrome, but they do not confirm or diagnose this. There is no risk to your baby by having a screening test. A lower chance result does not guarantee a baby does not have Down’s, Patau’s & Edwards’ syndrome and a higher chance result does not confirm the baby has Down’s, Patau’s & Edwards’ syndrome. Screening tests are used to identify which women are at a higher chance of having a baby with Down’s/Patau’s & Edwards’ 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 (NT) scan combined test (from 11+2 – 14+1 weeks of pregnancy. First trimester screening for Down’s, Patau’s & Edwards’ Syndrome screening test)

  With the first trimester (NT) screening you have a choice of screening options for:
  
  o Down’s, Patau’s & Edwards’ syndromes
  o Down’s syndrome only
  o Patau’s & Edwards’ syndrome only

- Quadruple blood test (from 14+2 to 20+0 weeks of pregnancy. Second trimester Down’s screening test)

- Mid Pregnancy Scan, (anomaly scan)
The Nuchal Translucency scan, combined test is the only test offered on the NHS to screen for Down's, Patau's & Edwards' syndrome.

The quadruple test blood test screens for Down’s Syndrome only.

**Twins/multiple pregnancies and Screening**

If you are aware you are having twins or a multiple pregnancy please inform your community midwife. They will inform the Antenatal Screening (ANNB) Co-ordinator for a further discussion regarding your choices and options for screening for Down’s, Patau’s and Edwards’ syndrome. You can contact the ANNB Screening Co-ordinator directly on either York 01904 725347 or Scarborough 01723 236308 for a discussion.

If you have opted for screening for T13/18/21 and found at scan to have a multiple pregnancy the scan will stop until you have had a further discussion with the ANNB Screening Co-ordinator. Further appointments will be made as necessary.
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. There may be a chance that the suspected anomalies are related to Downs, Patau’s or Edwards’s syndrome. 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/Patau’s & Edwards’ syndromes 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, Patau’s & Edwards’ syndrome. (Performed at 11 weeks, two days to 14 weeks, one day)

This is a screening test for Down’s, Patau’s & Edwards’ 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, Patau’s or Edwards’ 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, Patau’s or Edwards’ syndrome. Combining the result of this scan and blood test allows us to calculate the chance of the baby having Down’s, Patau’s & Edward’s syndrome.

The nuchal translucency scan combined test will identify 85 to 90 out of 100 of Down’s, Patau’s & Edwards’ syndrome babies in the higher chance category. It cannot pick up all babies with Down’s, Patau’s or Edwards’ syndrome and therefore a lower chance result does not guarantee a baby does not have Down’s, Patau’s or Edwards’ syndrome.

Sometimes the sonographer is unable to obtain the NT measurements and need a better image of the baby, this may be because the baby is in an awkward position or you are above average weight or your pregnancy is too advanced. In these cases the sonographer may ask you go for a short walk and then attempt to obtain the measurement again. If this is unsuccessful following the second attempt you may be offered the Quad Downs screening only. Screening for Patau’s & Edwards’ syndrome will be offered at the mid-pregnancy scan.
The result is presented as a statistic. Depending on the screening choices you will receive up to two results; one for Down’s syndrome and one for Patau’s & Edwards’ syndrome.

A result of one in 150 or any number less than this is classified as a higher chance. A result of one in 151 or any number greater than this is classified as a lower chance; the smaller the number, the higher the chance. If you have a lower chance result you will not be offered any further tests.

A woman receiving a higher chance result will be offered a diagnostic test (CVS or amniocentesis), but you can choose whether you would like to proceed to this test. These diagnostic tests will look at the chromosomes for all three syndromes, regardless of your initial choice.
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 chance more than 75 out of 100 babies with Down’s syndrome. A lower chance result cannot guarantee a baby does not have Down’s syndrome because the test does not pick up all affected babies. The result is presented as a statistic with a higher chance result being one in 150 or any number smaller than that. A result of one in 151 or any number larger than that is classified as a lower chance. If your result falls into the higher chance 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 chance 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 chance we will inform you by telephone to allow opportunity for discussion of the results and your options, this is usually within three working days of us receiving the result. You will be given opportunity to discuss this result with a face-to-face appointment. 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 York 01904 725347 or Scarborough 01723 236308.

More information can be found at www.nhs.uk/downs
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. The scan looks for certain problems, but is 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 identified on a scan. The scan will look in detail at your baby’s brain, bones, heart, face, kidneys, abdomen and spinal cord.
There are 11 conditions that are looked at on the scan, some are more easily detected than others:

<table>
<thead>
<tr>
<th>Conditions</th>
<th>Detection rate (out of 100)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anencephaly</td>
<td>98</td>
</tr>
<tr>
<td>Open spina bifida</td>
<td>90</td>
</tr>
<tr>
<td>Cleft lip</td>
<td>75</td>
</tr>
<tr>
<td>Diaphragmatic hernia</td>
<td>60</td>
</tr>
<tr>
<td>Gastroschisis</td>
<td>98</td>
</tr>
<tr>
<td>Exomphalos</td>
<td>80</td>
</tr>
<tr>
<td>Serious cardiac anomalies includes the following:</td>
<td>50</td>
</tr>
<tr>
<td>Transposition of the Great Arteries (TGA)</td>
<td></td>
</tr>
<tr>
<td>Atrioventricular Septal Defect (AVSD)</td>
<td></td>
</tr>
<tr>
<td>Tetralogy of Fallot (TOF)</td>
<td></td>
</tr>
<tr>
<td>Hypoplastic Left Heart Syndrome (HLHS)</td>
<td></td>
</tr>
<tr>
<td>Bilateral renal agenesis</td>
<td>84</td>
</tr>
<tr>
<td>Lethal skeletal dysplasia</td>
<td>60</td>
</tr>
<tr>
<td>Edwards’ syndrome (Trisomy 18)</td>
<td>95</td>
</tr>
<tr>
<td>Patau’s syndrome (Trisomy 13)</td>
<td>95</td>
</tr>
</tbody>
</table>

It also gives information on the position of the placenta (afterbirth).
If a problem is detected on scan you will be informed and a discussion/referral with a fetal medicine specialist may be made for you. Some of problems identified on scan may require some treatment or surgery, sometimes the problems are untreatable and your baby may die either during pregnancy or shortly after birth.

Your scan will be performed by a specially trained member of staff called a sonographer. The scan is performed in a dimly lit room to be able to get good images of your baby. You will be asked to lie on a couch and expose your tummy. The sonographer will put gel on your tummy and pass the hand-held scan probe over your tummy in order to obtain the images. The scan does not hurt, but you may feel slight pressure from the probe in order to get good images of your baby. The sonographer will view this on the screen; you will be given opportunity to see your baby on a screen. Black & white pictures are taken of your baby and you may be given opportunity to purchase these.

The scan usually takes approximately 30 minutes. Sometimes the baby is in an awkward position or the baby is moving a lot or if you are above average weight, the sonographer is unable to get the best view of your baby, you may be asked to go for a walk and the sonographer will try again. If they are unable to complete the scan you will be offered another scan appointment.

A full bladder is required for the scan.
You may wish for someone close to attend the scan with you in case there is a problem found on the scan. We do not recommend that children attend the scans for this reason and childcare is not available.

You can choose if you wish to have the scan or not. There is not known to be any risk to the baby by having a scan, however if a problem is identified it means you will have to make important decisions regarding the pregnancy and any further tests.

You will be told by the sonographer if the scan of your baby appears normal. The sonographer will inform you of any suspected anomalies and will refer you to the Antenatal Screening Co-ordinator or a consultant for further discussion and referral to a specialist for further investigation or a diagnostic test (amniocentesis).

For more information regarding your mid-pregnancy scan can be found at www.nhs.uk/anomalyscan.
What diagnostic tests are available?

None of the screening tests will pick up all Down’s, Patau’s or Edwards’ syndrome pregnancies. For those who wish a test to definitely detect Down’s, Patau’s or Edwards’ syndrome, a diagnostic test is needed. These are usually offered because you have received a higher chance 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 chance, 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. Additional chromosomal testing for other chromosomal problems are only requested if there was a problem identified on scan with your baby or requested specifically by your consultant. Therefore, they can identify other chromosome problems in addition to Down’, Patau’s or Edwards’ syndrome.

There is a risk of miscarriage with these tests of around one in 100
Chorionic Villus Sampling from between 12 weeks to around 15 weeks of pregnancy

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 abdomen 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’, Patau’s & Edwards’ syndrome, so further management could be based on this result. Rarely the cells do not give a result and you may be offered an amniocentesis to confirm a diagnosis.

There is a miscarriage risk of around 1% (one in 100) with this procedure and it is impossible to predict which pregnancies might be lost. Most miscarriages will occur within a few days following the test, but can be up to two weeks later. There is nothing you can do to prevent a miscarriage following the procedure.

There is also a risk (less than one in 1,000) that women may develop an infection following the procedure.

There may be occasions where the specialist is unable to perform the procedure and you will be re-offered another appointment for a CVS or offered an amniocentesis.

Compared with amniocentesis, CVS can be performed earlier and therefore results are obtained earlier in the pregnancy.
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 abdomen 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 1% (one in 100) and it is impossible to predict which pregnancies might be lost. Most miscarriages will occur within a few days following the test, but can be up to two weeks later. There is nothing you can do to prevent a miscarriage following the procedure.

There is also a risk (less than one in 1,000) that women may develop an infection following the procedure.

Occasionally (approximately less than seven out of a 100 women) we are unable to obtain sufficient fluid at the first attempt and the needle is re-inserted. If this second attempt fails, we will offer another appointment to attempt the procedure again.

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 chance of Down’s, Patau’s & Edwards’ syndromes. A lower chance result does not guarantee that the baby will not have Down’s, Patau’s or Edwards’ syndrome, just as a higher chance result does not mean that the baby has Down’s, Patau’s or Edwards’ syndrome.

- Diagnostic tests can confirm Down’s, Patau’s & Edwards’ syndrome with almost complete (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.
Who can I contact for more information?

Camilla Picknett, Antenatal Screening & Newborn Screening Coordinator for York Teaching Hospital NHS Foundation Trust.

Clare Hodgson, Support Screening Midwife (York)

Jolene Boyce, Support Screening Midwife (Scarborough)

Telephone: York Hospital: 01904 725347

Scarborough Hospital: 01723 236308

Further information & Support

Alternatively, refer to the leaflet: “Screening Tests for You and Your Baby” provided by your community midwife. It is also available online (available in 12 different languages/easy read/audio versions) at:


www.nhs.uk/downs

www.nhs.uk/anomalyscan
Diagnostic Testing

CVS/Amniocentesis information for parents online at:

www.nhs.uk/conditions/chorionic-villus-sampling/pages/introduction

www.nhs.uk/conditions/amniocentesis/pages/introduction


Other useful organisations are:

Antenatal Results and Choices, Tel: 020 7713 7356
www.arc-uk.org

Down’s Syndrome Association, Tel: 0333 1212 300
www.downs-syndrome.org.uk

Support Organisation for Trisomy 13/18 (SOFT)
www.soft.org.uk
Tel: 0330 088 1384
Tell us what you think of this leaflet

We hope that you found this leaflet helpful. If you would like to tell us what you think, please contact: Camilla Picknett, Antenatal & Newborn Screening Coordinator, The York Hospital, Wigginton Road, York, YO31 8HE or telephone 01904 725347.

Teaching, Training and Research

Our Trust is committed to teaching, training and research to support the development of health and healthcare in our community. Healthcare students may observe consultations for this purpose. You can opt out if you do not want students to observe. We may also ask you if you would like to be involved in our research.

Patient Advice and Liaison Service (PALS)

PALS offers impartial advice and assistance to patients, their relatives, friends and carers. We can listen to feedback (positive or negative), answer questions and help resolve any concerns about Trust services.

PALS can be contacted on 01904 726262, or email pals@york.nhs.uk.

An answer phone is available out of hours.
Providing care together in York, Scarborough, Bridlington, Malton, Selby and Easingwold communities

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Braille  Audio e.g. CD
Large print  Electronic

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Date first issued  November 2003
Review Date  January 2020
Version  11 (issued February 2018)
Approved by  Obstetrics & Gynaecology Clinical Governance Group
Document Reference  PIL 228 v11

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