Screening for Down’s syndrome in Multiple Pregnancy

When you are expecting twins, triplets or more

July 2007

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THE MULTIPLE BIRTHS FOUNDATION

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## Screening for Down’s syndrome in Multiple Pregnancy

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Screening for Down’s syndrome in Multiple Pregnancy

All pregnant women are now offered tests for Down’s syndrome. This booklet gives you some information about Down’s syndrome and testing for it when you are expecting twins, triplets or more, so you can decide whether to have the tests.

The screening process is different in multiple pregnancy and the booklet explains this in more detail.

Choosing whether to have the tests is an important decision, for you and for your babies. You need to make the decision that is right for you.

What is Down’s syndrome?

There is no such thing as a typical person with Down’s syndrome. Like all people, they vary a lot in appearance, personality and ability. People with Down’s syndrome have learning difficulties. Some have more serious difficulties than others.

It is hard to tell in babies how much they will be affected as children, or when they are grown up. Some adults with Down’s syndrome are able to get jobs and live fairly independent lives.

However, most people with Down’s syndrome need long-term help and support.

A number of health problems are linked to Down’s syndrome. But again, people vary, and some people with Down’s syndrome enjoy good health. Problems which are linked with Down’s syndrome include heart problems and reduced hearing and vision. Many of the problems can be treated, and frequent health checks can make sure that any problems are picked up as early as possible. Most people with Down’s syndrome live to be 50 years of age and some live to be over 70. Alzheimer’s disease (a form of senile dementia) may affect people with Down’s syndrome at an earlier age than other people.

What causes Down’s syndrome?

Inside all the cells of our bodies are tiny structures called chromosomes. These chromosomes carry the genes that determine how we develop. Most people
have 23 pairs of chromosomes in each of their cells. When our bodies produce the special cells needed to make babies, the chromosome pairs divide and rearrange themselves. Sometimes these pairs of chromosomes do not divide correctly, and this causes the baby’s cells to have an extra copy of chromosome number 21. This causes Down’s syndrome (and is the reason why one of the medical names for Down’s syndrome is Trisomy 21). The extra chromosome cannot be removed from cells, so there is no cure for the condition.

**Diagnosis of a multiple pregnancy**

You will have been told that you are expecting twins, triplets or more at your first ultrasound scan.

**How identical (monozygotic) and non identical (dizygotic) twins arise:**

One of the first things to be looked at is whether your twins have one or two placentas, and whether they might be identical (monozygotic) or non identical (dizygotic or fraternal). This is called establishing the chorionicity.

**Whether the babies are identical or non identical, and whether they share one placenta, is very important in understanding your options for Down’s syndrome screening.**

**Identical Twins**

Identical twins are also known as monozygotic (MZ) because they come from a single fertilised egg (zygote) which divides. The two babies will be the same sex and both will have the same genes.

**Non Identical twins**

Non identical twins are known as dizygotic (DZ) or fraternal. They come from two separate eggs being fertilised by two separate sperm. Non identical twins are genetically no more alike than other single born brothers and sisters and they can be either the same sex or a boy and girl.
If the fertilised egg divides around 0 to 4 days after fertilisation the babies will each have their own placenta (afterbirth). There will be two inner sacs (amnions) and two outer sacs (chorions). This may be written in your notes as \textbf{DCDA} (Di Amniotic Di Chorionic).

If the division happens around 4 to 8 days the babies will share a single placenta. There will be one outer sac but each will have its own inner sac and waters (amniotic fluid). This may be written in your notes as \textbf{MCDA} (Mono Chorionic Di Amniotic).

If the division happens around 8 to 12 days the babies will share one inner sac, one outer sac and are both in the same water. This will be written as \textbf{MCMA} (Mono Chorionic Mono Amniotic).

How common is Down’s syndrome in multiple pregnancy?

If they are non identical, then the risk of Down’s syndrome for each baby individually is the same as for a single baby. This is around 1 in 800 pregnancies. If they are identical (monozygotic), because the twins have the same genes, the risk to both of having Down’s syndrome is the same as if you were having a single baby.

Around 2% of pregnancies affected by Down’s syndrome are twins.

As explained above, the majority of twins with two placentas are non identical but some (approximately one third) of identical twins can also have two placentas.

Testing for Down’s syndrome during pregnancy

What is a screening test?

A screening test is a test offered to all women that carries no risk of miscarriage. The test is usually a scan, a blood test, or a combination of both, that will give a risk or chance of the baby or babies being affected by Down’s syndrome. The result is usually expressed as “higher risk” or “lower risk”. Sometimes a number is
This booklet gives the main facts, and tells you how you can get more information if you want to know more.

You may find it helpful to read through the whole booklet and consider how you might feel, and what you might wish to do, if one baby should be found to have Down's syndrome, before making a decision.

What screening is recommended for a multiple pregnancy?

Blood tests alone to screen for Down's syndrome are not currently recommended for multiple pregnancies, because markers in the blood are affected by the presence of more than one baby.

The screening test recommended when a mother has a multiple pregnancy should include a special ultrasound examination called a “Nuchal Translucency scan”, which can only be done between 11 weeks to 13 weeks and 6 days. The amount of fluid lying under the skin at the back of each baby’s neck is measured. A computer programme uses the measurement, the length of the baby (Crown rump length, CRL) and the mother’s age to work out the risk for Down’s syndrome for each baby. It is sometimes known as
the NT test. The results of the NT test may be used in combination with results from a blood test and the mother’s age, to calculate the risk of each baby being affected by Down’s syndrome.

If you have become pregnant using donated eggs, it is the approximate age of the woman who donated the eggs which will be needed to calculate the risk for your babies.

What happens if I decline Down’s syndrome screening?

If you decline screening, your care will not be affected and will continue as routine antenatal care. You will be offered an anomaly scan at around 18 - 20 weeks. This screening test is a detailed ultrasound scan but it cannot detect Down’s syndrome in your babies. If any abnormalities are seen on this scan, you may be offered a diagnostic test or referral to a specialist unit if you wish.

What happens if I am too late for a nuchal translucency scan?

If you are too late for your nuchal translucency scan (more than 13 weeks and 6 days) it will not be possible to accurately assess each baby’s individual risk of Down’s syndrome.

What happens next if I have a higher risk result and I am offered a diagnostic test?

Your midwife or doctor will discuss the results with you and answer any questions that you have. If you have a higher risk result, you will be offered a diagnostic test which would tell you definitely whether either of the babies has Down’s syndrome or not.

If you decide to have a diagnostic test, you should be referred to a specialist fetal medicine unit where the staff have specialist experience in looking after women with multiple pregnancies.

A specialist in the unit will discuss your care and give you advice on which diagnostic test is appropriate for you. You will have time to make up your mind about what to do next.

You have two options:

You can decide not to have a diagnostic test. The other option is to have the diagnostic test, knowing this may have other implications which you will need to think carefully about, and which carries an increased risk of miscarriage.

If you need any further information please refer to the organisations at the back of this booklet.
Diagnostic tests for Down’s syndrome

What can you tell me about diagnostic tests?

There are two diagnostic tests, amniocentesis and chorionic villus sampling. All diagnostic tests in a multiple pregnancy should be done in a fetal medicine unit. This is so that a detailed scan can be performed to accurately identify each baby's position prior to undergoing any diagnostic test.

What is chorionic villus sampling? (CVS)

CVS can be performed from 11 weeks and at any time during pregnancy. An ultrasound scan is used to guide a fine needle through your abdomen or through your vagina. A small sample of tissue is taken from the placenta of each baby. The sample is analysed in the laboratory in the same way as an amniocentesis. However, if the babies share one placenta, only one sample will be taken as the babies will both have the same chromosomes.

Are there any risks with chorionic villus sampling?

There is a 3-4% risk of miscarriage in twin pregnancies which means for every 100 women who have the test, 3 or 4 will miscarry.

What is amniocentesis?

Amniocentesis can be performed from 15 weeks of pregnancy. Under continuous ultrasound guidance a fine needle will be inserted through your abdomen, and a sample of fluid surrounding each baby (amniotic fluid) will be taken. The fluid contains cells from each baby, which will be examined in the laboratory. Very occasionally samples do not produce a result because the cells do not grow or the results are not clear. If this happens, you will be offered a second amniocentesis.

Are there any risks with amniocentesis?

There is a 2.5% risk of miscarriage in twin pregnancies. This means that for every 100 women who have amniocentesis, 2 or 3 will miscarry.

Are these procedures painful?

Many women find the procedures uncomfortable but they should not be painful. You will be advised to take things easy for a day or two afterwards. If possible you should avoid activities that involve lifting, bending or stretching. You may have some discomfort in your lower abdomen for a day or two after the procedure. This is normal and you can take paracetamol to relieve the discomfort.
How long does it take to get the results?

It can take up to 14 days to get the results of diagnostic tests. Some hospitals offer new “molecular” tests as part of the diagnostic test. These are usually known by their initials – PCR. These tests provide some information within two to three days, but you will still have to wait up to 14 days for the full diagnostic result. Waiting for the results can be an anxious time, so do call your midwife or one of the support organisations listed at the end of the booklet, if you need to talk.

How would I get my results?

Before you have the diagnostic tests done, your midwife or doctor will discuss how you want to receive the results.

What are the possible results from diagnostic tests?

The babies do not have Down’s syndrome

This is the result that most women get. Some women are happy just to get this news. They do not want to talk about tests and test results any more. Other women want to discuss the results with somebody. They want to know how the two tests they have had – the screening test and the diagnostic test – can seem to say different things.

We explained earlier that we use screening tests to decide who should be offered a diagnostic test.

When a woman has a diagnostic test and the result shows that one or both babies, if both are tested, do not have Down’s syndrome, the woman’s earlier screening test result is sometimes called a ‘false positive’ result. If this happens and you feel confused or upset about it, please talk to your midwife or doctor.

One baby has Down’s syndrome

A small number of women who have a diagnostic test will learn that one baby has Down’s syndrome
and the other is not affected. They then have the following options and it is entirely their decision which one to choose.

* Some people will decide to continue with the pregnancy, make plans and prepare for any challenges they might face, bringing up one twin with Down’s syndrome.

* Some people may feel unable to bring up the child themselves and consider adoption.

* Other people will decide they do not want to continue with the pregnancy and have a termination of the whole pregnancy.

* Others will decide that they want to consider the procedure to terminate the affected baby with the procedure called “selective termination (feticide)”, which can be discussed in detail with the fetal medicine specialist.

**Both babies have Down’s syndrome**

A very small number of women who have a diagnostic test will learn that both babies have Down’s syndrome. These women have the following options and it is up to them which they choose.

* Some people will decide to continue with the pregnancy, make plans and prepare for any challenges they might face bringing up two children with Down’s syndrome.

* Other people will decide they do not want to continue with the pregnancy and have a termination of the pregnancy.

* Others may feel unable to bring up both children themselves and consider adoption.

* Other chromosomal abnormalities may be detected by diagnostic test, and these will be discussed with you prior to the test being carried out, so you can decide if you wish to receive this information or not.

If you are faced with these results, you need to make sure you reach the right decision for you. You will be given information and support to help you make your decision, but it is up to you to decide what will be best for you. You will have the opportunity to discuss your options with health care professionals, and you will also be offered information and support from outside the health service. You will have time to decide what to do and will be supported by your GP, midwife and obstetrician in your decision. For further information please refer to the contact details at the back of this booklet.
You can get more information about screening from the following organisations.

* Antenatal Results and Choices (ARK)
  Website: www.arc-uk.org
  Helpline: 020 7631 0285
  Administration: 020 7631 0280

* National Down's syndrome Screening Programme
  Website: www.screening.nhs/downs/home.htm

You can get more information about multiple births from the following organisations:

* Multiple Births Foundation (MBF)
  Website: www.multiplesbirths.org.uk
  Email: mbf@hhnt.nhs.uk
  Telephone: 020 8383 3519

* Twins and Multiple Births Association (TAMBA)
  Website: www.tamba.org.uk
  Telephone: 0870 770 3305
  Twin line: 0800 138 0509
  (Everyday from 10am to 1pm, 7pm to 10pm)

You can get more information about Down's syndrome from the following organisations.

* Contact a Family
  Website: www.cafamily.org.uk
  Telephone: 020 7608 8700
  Helpline: 0808 808 3555
  This free helpline for parents and families is open from 10am to 4pm, Monday to Friday.

* Down's Syndrome Association
  Website: www.dsa-uk.com/
  Telephone: 0845 230 0372

* Down's Syndrome Medical Interest Group
  Website: www.dsmig.org.uk
  Telephone: 01159 627 658 extension: 45667
  An information service for healthcare professionals

Your midwife should have details of any local support groups.
Your notes

Your midwife's contact details
Name: 
Address: 
Phone number: 

Your doctor's contact details
Name: 
Address: 
Phone number: 

This booklet has been written by the National Down's Syndrome Screening Programme Multiple Pregnancy Information Group.

- Multiple Births Foundation
- Twins and Multiple Births Association
- Queen Charlotte's Hospital, London
- Addenbrookes Hospital, Cambridge
- St Mary's Hospital, London
- National Down's syndrome screening programme

References


National Down's Syndrome Cytogenetic Register
www.wolfson.qmul.ac.uk/ndscr


HTA (2003), First and second trimester antenatal screening for Down's syndrome, the results of the SURUSS study, Health Technology Assessment, Vol 7, No 11, (www.ncchta.org).

Data protection and guaranteeing quality in the Down's syndrome screening programme.

One of the aims of the screening programme is to make sure that it meets set quality standards and guidance. To do this, hospitals may need to use information about your screening choices to help improve the programme and to tell NHS planners about any extra funding they need. We will store personal information under the Data Protection Act 1999. By law all NHS staff must follow the act and keep your information confidential. We will only keep your personal information for as long as it is necessary for monitoring the screening programme. We will not give it to anyone outside the NHS. If you want to discuss any concerns you have about how we will keep the information, please ask your midwife.