



Testing for Down's syndrome in pregnancy

Choosing whether to have the tests is an important decision, for you and for your baby.

This chapter gives you some information about Down's syndrome and about testing for it, so you can decide whether to have the tests.

- * **All pregnant women in England are now offered tests for Down's syndrome. This booklet gives you some information about Down's syndrome and about testing for it, so you can decide whether to have the tests.**
- * **Choosing whether or not to have these tests is an important decision for you and your baby. You need to make the decision that is right for you, so please read this booklet carefully.**
- * **Your midwife or your GP will talk to you about testing for Down's syndrome. They will be happy to answer your questions – so please do ask if there is anything you are not clear about.**

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.

If you do not want your personal information stored or used for monitoring please inform your midwife or doctor.

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.

How common is Down's syndrome?

People do not usually expect to have a baby with Down's syndrome. It does not usually run in families. Some people think that only older women can have a baby with Down's syndrome, but this is not true. Anyone can have a baby with Down's syndrome, but the risk does go up with age.

The older a mother is the more chance she has of having a baby with the condition. For example, the chance of having a baby with Down's syndrome is one in 1500 for women who are 20 years old, one in 900 for women who are 30 years old, and one in 100 for



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women who are 40 years old. Some people prefer to think of the risk as a percentage. A risk of one in 1500 is the same as a 0.07% risk. A risk of one in 900 is the same as a 0.1% risk. A risk of one in 100 is the same as a 1% risk.

What causes Down's syndrome?

Inside all of the cells of our bodies there 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 it is the reason why one of the medical names for Down's syndrome is Trisomy 21). The extra chromosome cannot be removed from the cells, so there is no cure for the condition.

If the chromosomes divide incorrectly, this happens by accident. It is not caused by anything parents have done or have not done.

Testing for Down's syndrome during pregnancy

Should I have the tests for Down's syndrome?

Only you can decide that. Some women want to find out if their unborn baby has Down's syndrome and some do not. Information about the tests and how they work can help you make up your mind. This booklet gives the main facts and tells you how you can get more information if you want to know more.

A screening test carries no risk of miscarriage.

Will the tests tell me for certain if my baby has Down's syndrome?

We do not offer all women a test that will tell them for certain. This section explains why.

There are tests which give definite information. These are called **diagnostic tests**. The problem is that having a diagnostic test increases your risk of miscarriage. This is why we do not offer diagnostic tests to all women. Instead, we offer tests in two stages. We begin by offering all women a test that carries no risk of miscarriage. This type of test is called a **screening test**.

Screening tests **do not** give a definite answer, but they do tell us which babies have a higher risk (increased risk) of having Down's syndrome. We then offer diagnostic tests to the women at higher risk (increased risk). (There is more information on diagnostic tests later in the booklet.) If your screening result shows that you are at higher risk (increased risk), then you will be offered a diagnostic test. It is very important to understand that screening tests cannot tell you whether your baby definitely does or definitely does not have Down's syndrome.

We only use screening tests, and the two-stage process (the screening tests first and then the diagnostic tests), because we do not have a risk-free diagnostic test to offer. If you decide to have a screening test, and we later offer you a diagnostic test, it is your choice whether or not to have that diagnostic test. We explain more about the two-stage process later in the booklet.

What information does a screening test give me?

All of the tests described later in this booklet give results in the form of 'one in ...', for example, 'one in 100' or 'one in 1500'. (The same results can also be given as percentages – see page 18.) These numbers tell us

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how likely it is that your baby has Down's syndrome. For example, the result 'one in 100' means that there is one chance in 100 that your baby has Down's syndrome. The result 'one in 1500' means that there is one chance in 1500 that your baby has the condition. **It is very important to understand that as the second number in the result gets bigger, the chance of your baby having Down's syndrome gets smaller.**

Next we explain how we use the results from the screening test to decide whether to offer you more tests.

Women who are not offered more tests

If the screening test shows that the risk of your baby having Down's syndrome is lower than the recommended national cut-off, we will not offer you a diagnostic test.

Most screening test results (above 95%) fall into this category. This is known as having a 'low-risk' result. It is important to understand that a low-risk result means exactly that. It does not mean that there is no risk at all that your baby has Down's syndrome, just that it is unlikely.

There is still some risk of your baby having Down's syndrome because some babies with Down's syndrome are not detected by screening tests. This happens if the expected pattern is not seen in the blood test or in the scan measurements because of natural variations. Overall, about a quarter of babies with Down's syndrome are not detected by screening tests.

Women who are offered more tests

If the result of the screening test shows that the risk of your baby having Down's syndrome is greater than the recommended national cut-off we will offer you a diagnostic test. The screening result that leads us to

offer you more tests is sometimes known as the 'higher risk' or 'increased risk' result.

Overall about one in 30 (3%) of women screened have a higher (increased) risk result and are offered a diagnostic test. It is very important to remember how screening tests fit into the two-stage process. If you get a higher risk (increased risk) result from a screening test it means that we will offer you more tests. It does not mean that your baby definitely has Down's syndrome. As we explain later, most of the women who are offered further tests learn that their baby does not have Down's syndrome.

What screening tests will I be offered?

Screening for Down's syndrome is offered to pregnant women of all ages. The tests can provide information about the chance of a baby having Down's syndrome. These tests use blood samples taken from the mother, measurements taken from ultrasound scans or both to work out this chance. The tests you will be offered depend on how many weeks pregnant you are.

All tests offered must meet the national standards. To ensure this happens we audit the data (see page 16 for more information).

The combined test offered in early pregnancy

The combined test uses the results of a blood test and an ultrasound scan to calculate the risk (chance) of the unborn baby having Down's syndrome.

A blood sample taken from the mother between 10 weeks to 13 weeks and 6 days of pregnancy is used to measure the amount of some substances that

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are found naturally in the mother's blood. These substances are passed to the mother from the baby.

An ultrasound scan is carried out between 11 weeks and 0 days to 13 weeks and 6 days of pregnancy. This scan measures the amount of fluid lying under the skin at the back of the baby's neck. This is called the nuchal translucency (NT) measurement. A computer program then uses the results from the blood sample combined with the NT measurement to work out a risk (chance) figure. In addition to the results from the blood sample and the NT measurement, the program also uses the mother's age, weight, weeks of pregnancy, family origin and smoking details to work out this risk (chance) figure.

Screening later in pregnancy

If it has not been possible to have the combined test in early pregnancy, you will be offered a blood test between 15 weeks and 0 days to 20 weeks of pregnancy. This test looks at different substances to those measured in early pregnancy. Like the combined test, a computer program uses the results and the mother's details to work out a risk (chance) figure. This test is known as the quad (or quadruple) test.

What happens next if I have a higher risk (increased risk) result and I am offered more tests?

Your midwife or doctor will discuss the results with you and answer any questions that you have. You will be offered a diagnostic test which can tell you definitely whether your baby has Down's syndrome or not. There are two diagnostic tests available – chorionic villus sampling (CVS) and amniocentesis. CVS can be performed from weeks 10 to 22 of pregnancy although it is usually performed between weeks 11 and 13. Amniocentesis is usually carried out from week 15 of pregnancy.

If you do get a higher risk result from a screening test, your midwife or doctor will give you information and support. You will also have time to make up your mind about what to do next.

If you are in this position it is important to understand that you have a difficult decision to make.

You have two options. You can decide not to have a diagnostic test. This means spending the rest of your pregnancy knowing the screening result, which might be stressful.

The only other option is to have the diagnostic test, knowing that this will slightly increase the risk of miscarriage. You need to think carefully about what you would do if you found yourself in this position. Once you know the result of the screening test, you can't put the clock back. If you would not be happy with either of the above options, you need to consider very carefully whether it would be better for you not to have the screening test in the first place.

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How will I get the result from my screening test?

At the time of your test, ask your midwife or doctor how you will get the results. The test results should be available within two weeks.

What happens if I get a low-risk result, so I am not offered more tests?

Although your risk is low, you may want to discuss your results with your midwife or doctor.

Diagnostic tests for Down's syndrome

What can you tell me about diagnostic tests?

This booklet just gives some basic facts to help you decide whether you would want to have any screening or diagnostic tests you are offered. If you are actually facing a decision about diagnostic testing, your doctor or midwife will give you more detailed information.

What is chorionic villus sampling (CVS)?

CVS is a test carried out during pregnancy that involves removing a small piece of tissue from the placenta. It will generally be carried out between weeks 10 and 22 of pregnancy but is usually done between weeks 11 and 13. Sometimes it is carried out later. It is usually only offered in a specialist centre. The test itself takes around 10 minutes.

Immediately before the test, your abdomen is cleaned to make sure that the test can take place in the most sterile conditions possible. During CVS a

sonographer puts gel on your abdomen. You will then have an ultrasound scan to check the position of your baby. The sonographer or doctor will keep scanning you throughout the procedure. A fine needle is then inserted either through your vagina or through your abdomen and into your womb. A tiny sample of tissue is then removed from your placenta. Your placenta will usually contain tissue that is genetically identical to your baby.

The sample is analysed in the laboratory and the baby's chromosomes are counted. Very occasionally (about two in 100) CVS samples do not produce a result.

You may need to have a full bladder when you come for the appointment. Your doctor or midwife will let you know before you come. If you are not sure, you can contact them to ask.

What is amniocentesis?

Amniocentesis is a test carried out during pregnancy which involves using a fine needle to remove a small amount of the amniotic fluid around the unborn baby. It is a widely used procedure which usually takes about 10 minutes.

Amniocentesis is usually carried out between weeks 15 and 18 of pregnancy. However, the test can be done later in pregnancy.

Immediately before the test, your abdomen is cleaned to make sure that the test can take place in the most sterile conditions possible. During the amniocentesis, a sonographer puts gel on your abdomen. You will then have an ultrasound scan to check the position of your baby. The sonographer or doctor will keep scanning you throughout the procedure. A fine needle is then inserted through your skin, through your abdomen and into your womb. The needle is used to remove a small sample of the amniotic fluid surrounding your baby.

About one in 30 women screened are offered a diagnostic test.

This fluid contains cells from the baby, which will be examined at the laboratory and the baby's chromosomes counted. About one in every 100 samples does 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 these procedures safe?

These procedures are not completely safe, and this is why we don't offer them to everybody. The overall risk of you having a miscarriage after CVS is about 1 to 2%. In other words, about one or two in every 100 women who have CVS will miscarry. For amniocentesis, the rate is about one in 100. These figures vary slightly from hospital to hospital. If you would like to know the miscarriage rates after CVS or amniocentesis in your hospital, please ask your doctor or midwife.

Are the tests painful?

Many women find the procedures uncomfortable but they should not be painful. For a day or two afterwards you will be advised to take things easy. 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. Remember, you can only take a maximum of eight tablets in 24 hours. If you are worried about taking painkillers or have any questions, you should talk to your doctor or midwife.

How long does it take to get the results?

It can take up to 18 days to get the results of diagnostic tests. Some hospitals offer a 'molecular test' as part of the diagnostic test. This is usually known by its initials –

PCR. This test provides some information within two to three days.

Waiting for the results can be an anxious time. Do call your midwife or contact one of the support organisations listed on pages 29-31 if you need to talk.

How will I get my results?

It is important that you talk to your midwife about how you want to receive the results. The chances are that your results will show that your baby does not have Down's syndrome, but if the results show that your baby does have Down's syndrome, you need to think carefully about how you want to hear this information. Your midwife can give you the results at your home, at the antenatal clinic, over the phone or in a letter. Discuss what is appropriate for you with your midwife.

What are the possible results from diagnostic tests?

The baby does 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. Others may 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. If you want more information, we give a very brief explanation on page 28. If you really don't want to read any more details, just remember, the diagnostic test gives the definite result.

We explained earlier that we use screening tests to decide who should be offered a diagnostic test. What the screening tests do is tell us if there is a certain sort of pattern in your blood test or ultrasound results. This pattern is one that can sometimes be seen when a baby

For every 100 women who have amniocentesis one will miscarry.

has Down's syndrome. The problem is that the same pattern can also be seen in many normal pregnancies.

Women's blood test and scan results vary for all sorts of normal reasons. Screening tests just detect the pattern, they don't tell us the reason for the pattern. Only diagnostic tests can tell us if the reason for the pattern is that the baby has Down's syndrome. When a woman has a diagnostic test and the result shows her baby does not have Down's syndrome, that 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.

The baby does not have Down's syndrome, but the tests have shown some other problem

The main purpose of a CVS or amniocentesis is to find out whether a baby has Down's syndrome. But when the baby's chromosomes are examined, very occasionally other chromosome variations are identified. Some of these chromosome variations can be serious, and others will have only a minor effect, or no effect, on the baby. If the tests show there is a problem, you will be referred to a genetic counsellor for specialist information and support.

The baby has Down's syndrome

A small number of women who have a diagnostic test will learn that their baby has Down's syndrome. They then have three options, and it is entirely the parents' decision which they choose. Some people will decide to continue with the pregnancy, make plans and prepare for any extra challenges they might face bringing up a child with Down's syndrome. Some people may feel unable to bring up their child themselves, and decide on adoption. Some people

will decide they do not want to continue with the pregnancy, and will choose to have a termination. If you are faced with this decision, you need to make sure you reach the right decision for you. We will give you 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 healthcare professionals, and you will also be offered information and support from outside the health service. You will have time to decide what you are going to do and your GP and midwife will support you in your decision.

The diagnostic test gives the definite answer.

You can get more information about screening from the following organisations:

Antenatal Results and Choices (ARC)

www.arc-uk.org
73 Charlotte Street
London
W1T 4PN
Helpline: 0207 631 0285
Email: info@arc-uk.org

Antenatal Results and Choices (ARC) provides impartial information and individual support to parents whether they are going through antenatal screening or whose unborn baby has been diagnosed with an abnormality.

Most women who have a diagnostic test, learn that their baby does not have Down's syndrome.

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

Contact a Family (CAFAMILY)

www.cafamily.org.uk

209-211 City Road

London

EC1V 1JN

Helpline: 0808 808 3555

Email: info@cafamily.org.uk

Contact a Family is a charity which provides support, advice and information for families with disabled children, no matter what their condition or disability.

Down's Syndrome Association

www.downs-syndrome.org.uk

Langdon Down Centre

2a Langdon Park

Teddington

TW11 9PS

Helpline: 0845 230 0372

Email: info@downs-syndrome.org.uk

The Down's Syndrome Association is a support group for parents and carers of people with Down's syndrome.

Down's Syndrome Medical Interest Group

www.dsmig.org.uk

Children's Centre

City Hospital Campus

Hucknall Road

Nottingham

NG5 1PB

Telephone: 01159 627658 extension: 45667

This website is aimed at health professionals but includes information that parents may find helpful.

NHS Fetal Anomaly Screening Programme

www.fetalanomaly.screening.nhs.uk

Innovation Centre

Rennes Drive

University of Exeter,

Devon,

EX4 4RN

Telephone: 0845 527 7910

Email: enquiries@ansnsc.co.uk

The NHS Fetal Anomaly Screening Programme is responsible for both the NHS Fetal Anomaly Ultrasound Screening Programme and the Down's syndrome Screening Programme for England.

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