Prenatal Screening and Diagnosis of Down's Syndrome

There are two different types of tests that can be done to look for Down's syndrome during pregnancy - a screening test and a diagnostic test.

What is Down's syndrome?

Down's syndrome is a genetic chromosome problem that some people are born with. A person with Down's syndrome can usually be recognised by their typical features. It can also cause learning disability and there are certain medical problems that someone with Down's syndrome has an increased risk of developing. For example, a congenital heart defect (that is, a heart problem that you are born with), vision problems or growth problems. There is no cure for Down's syndrome. It will affect someone throughout their life.

Someone with Down's syndrome has an extra copy of chromosome 21 in the cells of their body. This is known as trisomy 21 (trisomy means there are three copies of a chromosome - in this case, chromosome 21). Because there is an extra chromosome 21, there is extra genetic material in the body. This causes the typical features of Down's syndrome. See separate leaflet called Down's Syndrome for more details.

What increases your risk of having a baby with Down's syndrome?

Anyone can have a baby with Down's syndrome but a woman's risk increases as she becomes older. Large studies have been carried out to look at the risk of Down's syndrome related to a mother's age and have shown that:

- A 20-year-old woman has a 1 in 1,500 risk of having a baby with Down's syndrome.
- A 30-year-old woman has a 1 in 800 risk.
- A 35-year-old woman has a 1 in 270 risk.
- A 40-year-old woman has a 1 in 100 risk.
- A 45-year-old woman has a 1 in 50 risk or greater.

However, it should also be remembered that the risk of conceiving a baby with Down's syndrome is actually higher than the above figures. This is because about three quarters of embryos or developing fetuses with Down's syndrome will never actually develop fully and so a miscarriage will occur before a baby is born.

If you have previously had a baby with Down's syndrome, your risk of having another affected baby is increased. About 1 in 100 women with a previous baby with Down's syndrome will have another.

Note: health professionals will either talk about higher risk or higher chance but they mean the same thing.

How is Down's syndrome diagnosed?

Down's syndrome can be diagnosed before birth (prenatally) or after birth (postnatally).

Down's syndrome may be suspected shortly after birth because of the typical features that a baby with Down's syndrome may have. Diagnosis is confirmed by genetic tests to look at the appearance and number of chromosomes of the baby. This is essentially their genetic make-up. It involves the baby having a blood test. The chromosomes in their blood can then be examined to look for an extra chromosome 21.

The rest of this leaflet concentrates on screening for Down's syndrome and the diagnosis of Down's syndrome during pregnancy, before the baby is born.

What is the difference between a screening test and a diagnostic test for Down's syndrome?

There are two different types of tests that can be done to look for Down's syndrome during pregnancy - a screening test and a diagnostic test.

As mentioned above, every woman has a risk of having a baby with Down's syndrome. A screening test is offered to all women in the UK during early pregnancy to look at the risk in this pregnancy of the baby being born with Down's syndrome.
It is important to understand that the screening test does not give a definite ‘yes’ or ‘no’ answer as to whether or not the baby does have Down's syndrome. If your screening test shows a high risk that the baby has Down’s syndrome (see below), you will usually be offered a diagnostic test. A diagnostic test is done before the baby is born (prenatally) to see if the developing baby actually does have Down's syndrome or not. There are two main prenatal diagnostic tests available: amniocentesis and chorionic villus sampling.

However, there is a small risk of complications if you have a diagnostic test. This includes miscarriage. This is why a diagnostic test for Down’s syndrome is not offered to all women. (A screening test for Down's syndrome does not increase your risk of having a miscarriage.)

**Screening tests for Down's syndrome**

All pregnant women should be offered a screening test for Down's syndrome. However, these tests are not compulsory. It is your choice as to whether you have the test or not.

**When will I be offered Down's syndrome screening?**

Screening can take place between 10 and 20 weeks of pregnancy. However, where possible, it is usually completed by 14 weeks and 2 days of pregnancy. This is so that you can have the combined test which is the recommended screening strategy in the first trimester of pregnancy.

**What screening tests are available?**

There are two basic methods of screening for Down's syndrome - the ultrasound scan and biochemical serum screening:

**Nuchal translucency ultrasound scan (also called the NT scan):** this is a special ultrasound scan that is done between 11 weeks and 2 days and 14 weeks and 1 day of pregnancy. It may be done at the same time as your early dating scan and is carried out in the same way. It measures the fluid collection under the skin at the back of the baby's neck (the nuchal translucency (NT)).

All babies have a collection of fluid here but babies with Down's syndrome tend to have more fluid in this area. The fluid measurement, your age, the size of the baby and other details such as your weight, ethnicity and smoking status are then put into a computer program to give the risk of the baby having Down's syndrome.

Sometimes it may be difficult to obtain an accurate measurement of the NT. This may be due to the position of the baby or because the pregnant woman is overweight.

**Blood tests (also called biochemical serum screening):** blood tests may be done to measure levels of various hormones and proteins in your blood. These hormones and proteins are produced by the placenta or the developing baby.

Examples include pregnancy-associated plasma protein A (PAPP-A), beta human chorionic gonadotrophin (beta-hCG) and alpha-fetoprotein (AFP). If the baby has Down's syndrome, the levels of these substances can be affected.

Again, a computer program is used to give the risk of the baby having Down's syndrome, based on the blood test results, your age, the stage that you are at in your pregnancy, your weight, ethnicity and smoking status. The blood test used in Down's syndrome screening is also sometimes called serum screening.

The two main screening tests used for Down's syndrome using the above methods are:

- **Combined screening test.** This combines the result of the NT scan with the result of blood tests for PAPP-A and beta-hCG to give the risk of the baby having Down's syndrome. The blood tests are often taken at the same time as the scan. This is the screening test that the NHS Fetal Anomaly Screening Programme recommends.

- **Quadruple screening test.** This is based on the results of a blood test carried out between 14 weeks and 2 days and 20 weeks of pregnancy. This test does not include an ultrasound scan. It is used for women who come for screening later in pregnancy, or in centres which do not offer the combined test.

**How will I receive the results of the screening test?**

You should ask your doctor or midwife how long the results will take and how you will receive them. The results can take up to two weeks.

**How do I interpret the results of the screening test?**

As mentioned above, the screening test will give the risk in this pregnancy of the baby being born with Down's syndrome. For example, the test may show that there is a 1 in 1,000 risk of having a baby with Down's syndrome. This means that for every 1,000 pregnant women, one will have a baby born with Down's syndrome and 999 will have a baby born without Down's syndrome. So, this would be quite a low risk.

In the UK, the National Screening Committee has suggested a cut-off level to differentiate between screening test results with a higher risk that the baby is born with Down's syndrome and those with a lower risk.

The cut off is 1 in 150. This means that if your screening test results show a risk of between 1 in 2 to 1 in 150 that the baby has Down's syndrome, this is classified as a higher risk result. If the results show a risk of 1 in 151 or more, this is classified as a lower risk result. The higher the second number gets, the lower the risk becomes (the less likely you are to have a baby with Down's syndrome).
So, when you are given your results, you will be told whether this is a lower risk or a higher risk result.

What happens if I have a lower risk result?
Most women who have a screening test for Down's syndrome will have a lower risk result. If this is the case, you can be reassured by this. This does not mean your baby definitely does not have Down's syndrome (although the likelihood that this will happen is very small). In some cases, the baby does have Down's syndrome (this is called a 'false negative' result).

What happens if I have a higher risk result?
If you have been given a higher risk result, this does not mean your baby definitely has Down's syndrome. If you are given a higher risk result, further tests are needed to confirm the diagnosis and give a definite 'yes' or 'no' answer (see below). In cases where the baby does not have Down's syndrome, this is called a 'false positive' result.

You can still choose whether or not you want to go ahead with the diagnostic test at this stage. You can still say no.

Diagnostic tests for Down's syndrome
The two main tests that are used to diagnose Down's syndrome before birth (prenatally) are amniocentesis and chorionic villus sampling (also called CVS). In both tests samples are taken from the inside of your womb with a needle usually passed through your tummy. There is a small risk of miscarriage in both tests. CVS can be done earlier in pregnancy than amniocentesis. Both tests are discussed further in separate leaflets. Read about amniocentesis and about chorionic villus sampling (CVS).

How long will the results of diagnostic testing take and how will I receive them?
You should ask your doctor or midwife how long the results will take and how you will receive them. For example, you may be given another appointment or sometimes results are given by telephone.

There are two main tests that can be done to look at the baby's chromosomes after amniocentesis or CVS. The first is called a rapid test. This usually gives results within three days after CVS. This test just looks for the chromosomal disorders Down's syndrome, Edwards' syndrome and Patau's syndrome. Sometimes sex chromosome disorders such as Turner syndrome can also be detected on a rapid test.

The second test is a chromosomal microarray that looks at all of the baby's chromosomes in detail. It can show up other chromosomal disorders and abnormal genes. This takes longer to get the results - usually, two to three weeks. A chromosomal microarray test may be suggested by your doctor if other genetic conditions are suspected. Sometimes only a rapid test is carried out. Your doctor or midwife will discuss with you which tests are best in your situation.

What are my options if the results of the diagnostic test are abnormal?
Before you leave the hospital, someone will ask you how you would like to receive the results of your CVS or amniocentesis. You can usually choose whether to receive the results by phone or come into the hospital again and receive your results face-to-face. In some cases, a community midwife may be able to come to your home to give you the results. You will also receive written confirmation of your results.

For most women, the laboratory test will give a definite 'yes' or 'no' answer. These results will let you know whether your baby has the chromosomal abnormality the test was looking for.

Most women who have CVS or amniocentesis will have a 'normal' result (in other words, their baby won't have the chromosomal abnormality the test was looking for).

Some women will be told their baby has the chromosomal abnormality the test was looking for. If your baby is diagnosed with a chromosomal abnormality, your doctor or midwife will give you information on the condition your baby has and they will give you the opportunity to discuss this with a specialist.

Very occasionally, women who have CVS or amniocentesis to detect Down's syndrome find out their baby has a different chromosomal abnormality. If this happens to you, your doctor or midwife will give you information on the condition your baby has and they will give you the opportunity to discuss this with a specialist.

A normal CVS or amniocentesis result does not guarantee your baby will not have any abnormalities. Not all abnormalities can be detected by the CVS or amniocentesis test. Also occasionally the result will not be clear, and further tests may be suggested.

If the results show your baby has a chromosomal abnormality, your doctor or midwife will talk to you in detail about this. When you are deciding what to do, you need to consider what is best for you and your family.

These decisions are often very difficult and you might want to talk about your feelings with a midwife, doctor or a support organisation for parents, such as Antenatal Results and Choices (ARC).

You might choose to:

- Continue with your pregnancy and use the information you have gained from the test results to help prepare for the birth and care of your baby; or
• Continue the pregnancy and consider adoption.
• End the pregnancy (have a termination).

So, should I have screening for Down's syndrome during my pregnancy?

It can be difficult to make a decision about having screening tests and diagnostic tests for Down's syndrome. You may find it helpful to talk to your midwife, your doctor and your friends and family. You should remember that only you can decide whether or not you want to have this test. Considering the following questions might be helpful:

• If your screening test shows that there is a higher risk that the baby could be born with Down's syndrome, what would you do? Would you want to go ahead with a diagnostic test knowing that there is a small risk of miscarriage or other complications?
• If your screening test has a higher risk result and you choose not to have a diagnostic test, how will you feel spending the rest of your pregnancy knowing that you had this higher risk result?

There are some women who feel that it is better for them not to have a Down's syndrome screening test in the first place. They know that they would not go through with a diagnostic test if they had a higher risk screening result because of the small risk of miscarriage and the possibility that the baby that is miscarried might be healthy. Other women know that they would not consider a termination of pregnancy as an option if a diagnostic test showed that the baby did have Down's syndrome.

Equally, other women decide that, on balance, they are willing to accept the small risk of miscarriage or other complications of a diagnostic test in order that they have the definite information about whether or not the baby has Down's syndrome. They are then able to consider their options before the baby is born.

Further reading & references

• Antenatal care for uncomplicated pregnancies; NICE Clinical Guideline (March 2008, updated 2018)
• Fetal anomaly screening: programme overview; GOV.UK
• Population Screening Programmes (England)
• Antenatal Screening Wales
• Antenatal Screening; Health and Social Care Northern Ireland
• The UK NSC recommendation on Down's syndrome screening in pregnancy
• Amniocentesis and Chorionic Villus Sampling; Royal College of Obstetricians and Gynaecologists (June 2010)
• Routine antenatal anti-D prophylaxis for women who are rhesus D negative; NICE Technology Appraisal Guidance, August 2008

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