Down's Syndrome Association
A Registered Charity No. 1061474

Screening, diagnosis and support

Date: November 2024

If you require information or support you can speak to one of our information officers by phoning our **Helpline on 0333 1212 300**.

The Down's Syndrome Association is a source of balanced, accurate information and advice. We are available for calls to our Helpline every weekday from 10 am - 4pm or you can contact us by sending an email to info@downs-syndrome.org.uk. Your call or enquiry will remain confidential. We talk to parents at any stage of pregnancy. We can answer your questions and guide you to relevant information.

Prenatal screening

Prenatal screening for Down's syndrome is offered to pregnant women in England and Wales and may be offered to women in Northern Ireland. Any offer of screening should be presented with up to date and accurate information about Down's syndrome as well as about screening tests at the woman's first antenatal appointment. It is the woman's decision whether to choose screening tests or not and the options must be presented in a non-directive manner.

If you are pregnant, you should make informed choices that are right for you. You may want to think about why you might choose screening tests, what the tests might tell you and what choices the tests may lead you to consider as a result.

Screening tests

Screening tests can provide information about the chance that a baby may have Down's syndrome. The tests use blood samples taken from the mother and measurements taken from ultrasound scans to work out this chance. The tests you will be offered depend on how many weeks pregnant you are.

Screening in early pregnancy: blood test and ultrasound scan

The combined test is offered in early pregnancy. It uses the results of a blood test and an ultrasound scan to calculate the chance that a baby may have Down's syndrome.

A blood sample taken from the mother between 10 weeks to 14 weeks + 1 day of pregnancy is used to measure the amount of some substances that are found naturally in the mother's blood. An ultrasound scan is carried out between 11 weeks + 2 days and 14 weeks + 1 day 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 chance figure. In addition to the results from the blood sample and the NT measurement, the program also uses the mother's age to work out this chance figure.

Screening in later pregnancy: quad or quadruple test

If it has not been possible to have the combined test in early pregnancy you will be offered a blood test between 14 weeks + 2 days to 20 weeks of pregnancy. The quad or quadruple 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 chance figure.

Higher and lower chance results

If the screening test shows that the chance of the baby having Down's syndrome is between 1 in 2 and 1 in 150 this is called a higher chance result.

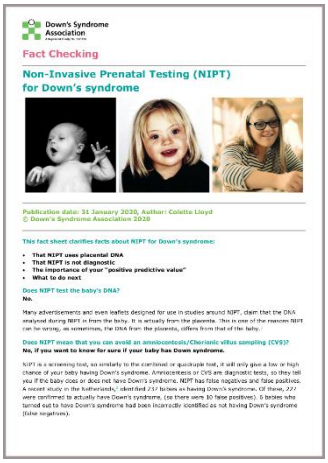
If the screening test shows that the chance of having a baby with Down's syndrome is lower than 1 in 150, this is a lower chance result. A lower chance result does not mean there is no chance at all of the baby having Down's syndrome.

Only a diagnostic test (amniocentesis or chorionic villus sampling) can tell you if your baby has Down's syndrome.

Non-invasive prenatal test

A non-invasive prenatal blood test (NIPT) is currently available as a supplementary (NHS) test to women who have had a higher chance result from a combined or quadruple test. NIPT can be carried out from 10 weeks of pregnancy.

The DSA NIPT 'Fact Checker' in our pregnancy and baby resources section clarifies facts for pregnant woman about NIPT for Down's syndrome.


Down's Syndrome Association

Fact Checking

Non-Invasive Prenatal Testing (NIPT) for Down's syndrome

Publication date: 31 January 2020, Author: Colette Lloyd
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This fact sheet clarifies facts about NIPT for Down's syndrome:

- That NIPT uses placental DNA
- That NIPT is not diagnostic
- The importance of your "positive predictive value"
- What to do next

Does NIPT test the baby's DNA?

No.

Many advertisements and even leaflets designed for use in studies around NIPT, claim that the DNA analysed during NIPT is from the baby. It is actually from the placenta. This is one of the reasons NIPT can be wrong, as sometimes, the DNA from the placenta, differs from that of the baby.

Does NIPT mean that you can avoid an amniocentesis/Chorionic villus sampling (CVS)?

No, if you want to know for sure if your baby has Down syndrome.

NIPT is a screening test, so similarly to the combined or quadruple test, it will only give a low or high chance of your baby having Down's syndrome. Amniocentesis or CVS are diagnostic tests, so they tell you if the baby does or does not have Down's syndrome. NIPT has false negatives and false positives. A recent study in the Netherlands,[1] identified 237 babies as having Down's syndrome. Of these, 227 were confirmed to actually have Down's syndrome, (so there were 10 false positives). 5 babies who turned out to have Down's syndrome had been incorrectly identified as not having Down's syndrome (false negatives).

Non-invasive prenatal testing (NIPT) involves a blood sample being taken from the mother. The sample is sent to a laboratory where it is tested for the amount of cell-free fetal DNA (cffDNA) present, particularly chromosome 21.

Each chromosome has strings of DNA that are specific to that particular chromosome. So by analysing the DNA strings that link to each chromosome, the amount of chromosome 21 in the blood sample can be compared to the amount of the other chromosomes. The total amount of chromosome 21 in the blood sample is then used to calculate the likelihood of the baby having Down's syndrome. The cell-free fetal DNA analysed during NIPT is from the placenta, not the baby. This is one of the reasons why a NIPT result can be wrong sometimes.

NIPT is an enhanced screening test, not a diagnostic test. Further diagnostic tests would be necessary to diagnose Down's syndrome.

If you have a higher chance result

Your midwife or doctor will discuss the screening test results with you and answer any questions that you have. It is your choice whether you wish to have a diagnostic test. You may want to think carefully about this before you make your decision. You can discuss the options available with your midwife and you can call our Helpline to discuss any matters confidentially.

Prenatal diagnostic tests

A diagnostic test will tell you whether your baby does or does not have Down's syndrome.

If you are given a higher chance result from a screening test your midwife or doctor can give you information about diagnostic testing if you want this. It is your choice whether or not to take a diagnostic test.

You can call our Helpline to find out about Down's syndrome or talk with us.

There are two types of diagnostic prenatal test:

Chorionic Villus Sampling (CVS) is an invasive test. It can be performed from weeks 10 to 22 of pregnancy although it is usually performed between weeks 11 and 13.

The chance of 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.

Amniocentesis is an invasive test. It is usually carried out from week 15 of pregnancy.

The chance of having a miscarriage after an amniocentesis 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.

It is your choice to decide what to do after receiving a diagnosis of Down's syndrome.

If the test finds that your baby has Down's syndrome this knowledge may inform the management of your pregnancy and your birth plan in a helpful way.

Our position on prenatal testing

The Down's Syndrome Association would not want to see a world without people who have Down's syndrome. People who have Down's syndrome make an enormous contribution to their families, local communities and society in general in all sorts of different ways.

We acknowledge that prenatal testing is a routine part of antenatal care and that it is a matter of individual choice as to whether to have prenatal tests or not.

At the DSA, we are committed to ensuring that all potential parents and professionals are made aware of the joys and challenges of having a child who has Down's syndrome.

Before and during testing, easily understood and up-to-date information MUST be provided in an unbiased way by well trained professionals concerning:

- The accuracy of tests and associated risks of further tests;
- The life prospects of people who have Down's syndrome;
- The impact on families;
- The support available in the community;
- Broad and non-directive counselling services for those who may need it.

We expect respect and support for parents making choices about antenatal tests and their outcomes – whatever they decide to do.
downs-syndrome.org.uk/about-dsa/who-we-are/where-we-stand/

Postnatal diagnosis

A chromosome test will need to be done before a diagnosis of Down's syndrome can be made. The doctor will take a blood sample from the baby and send it for chromosome analysis. Until the results of a diagnostic test are returned, a healthcare professional cannot say whether or not the baby has Down's syndrome.

We advise that healthcare staff are open to answering questions from parents during this period because, overwhelmingly, parents tell us that what they value during this period is honesty. The giving of information at this stage is not detrimental and parents say they would rather this than feel that they are in a period of limbo, where professionals are reluctant to speak openly or answer questions.

Related links

- Public Health England provides information about the screening pathway for pregnant women in England: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/897412/STFYAYB_June_2020.pdf

- Government guidance for health professionals in England: gov.uk/government/publications/fetal-anomaly-screening-care-pathways/fetal-anomaly-screening-programme-screening-pathway-for-downs-syndrome-edwards-syndrome-and-pataus-syndrome

- Antenatal Screening Wales provides information about the screening pathway for pregnant women in Wales: antenatalscreening.wales.nhs.uk/public/home

- The HSC Public Health Agency provides information about screening in Northern Ireland: publichealth.hscni.net/directorate-public-health/service-development-and-screening/antenatal-screening

- NI Direct provides information about screening in Northern Ireland: nidirect.gov.uk/articles/antenatal-care

- NHS Inform provides information about screening in Scotland: nhsinform.scot/healthy-living/screening/pregnancy/pregnancy-screening

- NHS Direct Health A to Z - What is Down's syndrome? nhs.uk/conditions/downs-syndrome/

Contact us

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