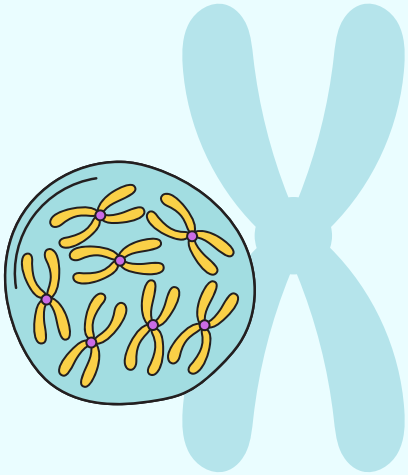
The National Maternity Hospital

Optional Screening for Chromosomal Anomalies in Pregnancy



Introduction

This leaflet is about the screening test for chromosomal anomalies (such as Down syndrome and Edward syndrome) you can consider during your pregnancy. This leaflet aims to provide you with information so that you can make an informed decision about whether you want to have this screening test.

Some people choose to have screening tests in pregnancy to assess the chance of their baby having a particular chromosomal problem.

As women age, the chance of having a child affected by a chromosomal abnormality increases, but all women can have a pregnancy affected by a chromosomal abnormality.

These screening tests are usually simple tests (for example, ultrasound scans or blood tests, or a combination of both). They do not provide a definite diagnosis, but help you and your antenatal team decide whether you need further tests to make that diagnosis.

Screening tests such as Non-Invasive Prenatal Screening (known as NIPS or NIPT) are currently not funded by the Health Service Executive (HSE), but you can get this test done through a private provider.

What are chromosomes?

The body is made up of cells. In the centre of most cells are thread-like structures known as chromosomes. Usually there are 46 chromosomes in each cell of the body, arranged in 23 pairs. Chromosome pairs numbered 1 to 22 are the same in males and females. The 23rd pair are called the sex chromosomes. Females tend to have two X chromosomes and males tend to have one X and one Y chromosome. DNA is the substance that our chromosomes are made of.

Chromosome abnormalities are changes to the structure or number of chromosomes that can lead to a birth defect or other health problems.

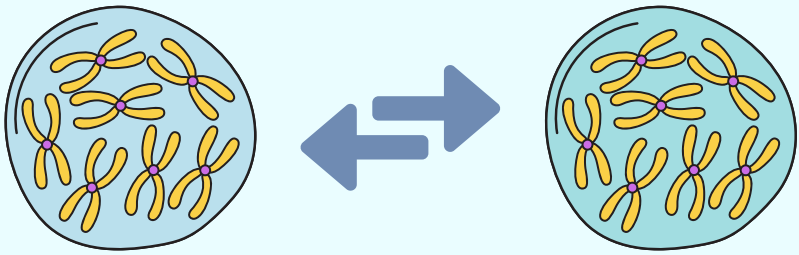
The most common chromosomal abnormalities are Down syndrome (Trisomy 21), Edward syndrome (Trisomy 18) and Patau syndrome (Trisomy 13), as well as sex chromosome abnormalities (e.g. Turner syndrome, Klinefelter syndrome, etc).

DNA

What screeening tests are available?

The most accurate method of chromosomal abnormalities screening is Non-Invasive Prenatal Screening/Testing (known as NIPS/NIPT).

It is important to be aware that different private clinics may offer different screening tests.



What is NIPS/NIPT?

The purpose of the NIPS/NIPT screening test is to identify pregnancies at increased risk of being affected by common chromosomes abnormities: Down syndrome, Edward syndrome and Patau syndrome.

Some of the baby's DNA from the placenta (afterbirth) circulates in the mother's blood during pregnancy. The test is performed on a sample of the mother's blood and then sent to the laboratory for testing from 10 weeks of pregnancy. Even after 10 weeks there is a chance that there will not be enough of baby's DNA in the sample to provide a result. This can happen in about 3% of cases and another blood sample may be required.

This test can be offered on singleton and twin pregnancies (including IVF pregnancies), but at present not for triplet pregnancies.

Is NIPS/NIPT suitable for everyone?

No. NIPS/NIPT is unsuitable for some women. Some reasons include:

- If there is evidence of a vanished twin pregnancy
- If the expectant mother has cancer
- If the expectant mother had a blood transfusion in the last four months
- If the expectant mother is a bone marrow or organ transplant recipient
- If the expectant mother has a chromosomal abnormality (please discuss this with your midwife who will contact the genetics team if required)
- If the expectant mother is a balanced translocation carrier
- If the expectant mother is on immunotherapy in the current pregnancy (excluding IVIg)
- If the expectant mother has had stem cell therapy

How accurate is NIPS/NIPT?

NIPS/NIPT has a high sensitivity (the ability to correctly identify a truly high risk case as high risk) and specificity (the ability to identify an unaffected case as low risk) for Down syndrome. However, it has lower sensitivities and specificities when screening for the other chromosomes (18 and 13).

Although sensitivity and specificity are important measures, positive predictive value (PPV) and negative predictive value (NPV) are more clinically relevant after results have been returned to a woman/couple. PPV is the likelihood that the result of NIPS/NIPT says the pregnancy is high risk and the fetus is actually affected by the chromosomal abnormality. NPV is the likelihood the result says low risk and the baby is not affected.

It is important to note that maternal age is important in deciding PPV, so the statistics are based on age-related risk and the sequencing that occurs on the fetal DNA.

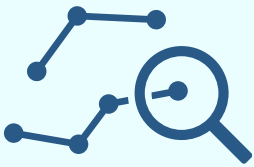
What are the limitations of NIPS/NIPT?

No test is accurate all of the time – the test will not detect all cases of a chromosomal abnormality (referred to as a false-negative result), and may sometimes indicate a problem even when none is present (referred to as a false-positive result).

NIPS/NIPT is not designed to detect small changes on chromosomes or non-chromosome disorders like single gene conditions like cystic fibrosis.

If there are anomalies detected on scan, NIPS/NIPT will be deferred and you will be offered an invasive test to investigate the cause of the abnormalities. In a small number of situations, NIPS/NIPT might be considered.

If you have a personal or family history of a genetic condition, please speak to your doctor or midwife at your booking visit. Alternatively, you might contact the Clinical Genetics team who performed the original testing.



What are the possible outcomes of NIPS/NIPT?

NIPS/NIPT does not give a yes/no answer; it gives a probability.

- If the result is reported as ‘low chance’ or ‘very unlikely to be affected’, it is unlikely that the baby has one of these chromosomal abnormalities. You should continue to have your routine antenatal care.

- If the test result given is ‘high chance’ or ‘likely to be affected’, there is a possibility that the baby has a chromosomal abnormality and further investigations would need to be carried out to give an accurate diagnostic result. This includes detailed ultrasound scans and an invasive test. It is likely that you will be seen in a fetal medicine unit for this.

What happens next after a ‘high chance’ NIPS/NIPT result?

You will be referred for a detailed scan in the Fetal Medicine Unit. If you have a high-chance result, you can decide to have an invasive diagnostic test, such as chorionic villus sampling (CVS) or an amniocentesis performed by Fetal Medicine Specialists.

A CVS uses a needle to obtain a small piece of tissue from the placenta from 11 weeks of pregnancy. In an amniocentesis a sample of amniotic fluid is taken from 15 weeks of pregnancy. There is a small risk of miscarriage associated with these procedures (0.5-1%). The Fetal Medicine Specialists will advise you regarding which test is best for your situation.

Once the invasive procedure is performed, the result of the invasive diagnostic test will take some time. Unfortunately, we cannot speed this up.

Some people choose not to have invasive testing and to continue with the pregnancy with the uncertainty from the NIPS/NIPT. In this situation, you will receive scans in fetal medicine where appropriate and you might be seen by the Clinical Genetics team to discuss genetic testing after the baby is delivered.

Do I have to have NIPS/NIPT?

No – having a screening test is a personal choice that only you can make. Before having screening, it is worth thinking about how you would feel if this test came back as ‘high chance’ of a chromosome abnormality, and if you would consider having an invasive test to clarify the result.

If you decide not to have screening in pregnancy, the rest of your antenatal care will not be affected. You will still have a detailed anatomy scan of your baby between 20-22 weeks of pregnancy as part of your routine antenatal care. Detailed scanning does not detect all structural abnormalities.

The National Maternity Hospital

If you wish to have more information on any of the above tests, please speak to a doctor or midwife at your booking visit.

Resources

Antenatal Results and Choices (ARC)
Non-directive information and support before, during and after antenatal screening
www.arc-uk.org

Irish Family Planning Association (IFPA)
Confidential and non-directive specialist pregnancy counselling for women and girls, pregnant people and their partner.
www.ifpa.ie

Leanbh Mo Chroi
Offer support and encouragement to those receiving a fatal or severe diagnosis during pregnancy.
www.lmcsupport.ie

Contact information

The National Maternity Hospital
Holles Street, Dublin 2, D02 YH21

Phone: 01 637 3100
Website: **www.nmh.ie**