PATHOLOGY TESTS EXPLAINED

Information about pathology tests to help everyone take control of their health and make the right decisions about their care.

WHAT YOU SHOULD KNOW ABOUT YOUR **NON-INVASIVE PRENATAL TEST (NIPT)**

When you are pregnant, your blood contains fragments of your baby's DNA. A non-invasive prenatal test (NIPT), also known as cell free DNA testing, analyses this DNA in a sample of your blood to assess if your baby has an increased chance of having a chromosomal disorder such as Down syndrome or one of the less common conditions.

Chromosomal disorders

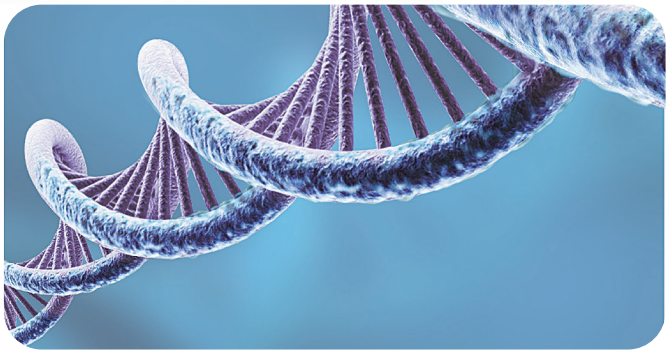
Chromosomes are spiralling strands of DNA - packaging for our genes. They are found in most cells in our bodies and can only be seen through a high powered microscope.

Chromosomes come in pairs. We inherit one of each pair from our mother (via the egg) and the other from our father (via the sperm).

We normally have 23 pairs (46 in total) including sex chromosomes – two Xs for a female and an X and a Y for a male.

Most chromosome abnormalities occur as an accident in the egg or sperm. In these cases, the abnormality is present in every cell of the baby's body.

Some abnormalities happen after conception. When this happens, some cells have the abnormality and some do not. NIPT does not test for these.



There are many different types of chromosome disorders. Some occur when there is an extra copy of a chromosome (called trisomy), others because one is missing (monosomy).

Some disorders are caused when a segment of a chromosome is missing or else there is an extra copy of a segment. Sometimes a segment of one chromosome can be moved to another. Sometimes two chromosomes can be fused together.

The most common chromosome condition is Down syndrome (trisomy 21), which is usually a sporadic disorder, meaning that there is usually no prior history in the family, and it is not inherited from a parent.

Other chromosome conditions that are commonly screened for by NIPT include Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13). Sex conditions such as Turners syndrome (monosomy X) are also screened for.

What can your results tell you?

NIPT measures the amount of DNA from your baby circulating in your blood stream. More specifically, it measures the DNA coming from the specific chromosomes known to be linked to certain disorders.

NIPT has a sensitivity of around 99.5 per cent for detecting Down syndrome – less than one case in 100 will be missed by the test. It also has a very high specificity but positive results do require confirmatory testing.

Very infrequently the test may not give a result as there may be insufficient DNA from the baby in the mother's blood sample. This is more likely if the test is conducted in very early pregnancy.

What happens next?

If your test result is positive and shows that your baby is at increased risk of a chromosome disorder, your result needs to be confirmed by a diagnostic test such as amniocentesis or chorionic villus sampling. These can provide a definitive answer on the chromosomal makeup of the baby's cells. These procedures are invasive and carry a very small risk of miscarriage.

First trimester ultrasound remains an important screening tool for all abnormalities that may, or may not have a chromosomal cause.

NIPT is available primarily through private laboratories. It is not covered by Medicare or private health insurance.

If your test is positive, it is important that you are put in touch with a genetic counsellor who can explain what the risk means in your personal situation. They can help you understand the information you've been given and provide support if you need it.

5 questions to ask your doctor

What is the risk of my baby having a chromosome disorder?

What do I need to think about before getting tested?

What will happen if the test is positive?

What if I test negative – can there still be a problem?

What will happen next, after the test?

For more detailed information on these and many other tests go to **pathologytestsexplained.org.au**

Please use this QR code to access more information

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www.pathologytestsexplained.org.au

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Pathology Tests Explained is managed by a consortium of medical and scientific organisations representing pathology practice in Australia. More details at: **www.pathologytestsexplained.org.au/about**

When you have pathology tests you can have your results sent directly to your My Health Record.

You'll find a direct link to the Pathology Tests Explained website embedded in the pathology results pages of your record.

Click on the link to find information about what your tests are investigating or measuring and what your results can tell your doctor.