NON-INVASIVE PRENATAL TEST (NIPT)

ACCURATE, SAFE, RELIABLE

HOW DOES NIPT WORK?

In pregnancy, small amounts of DNA from the placenta of the foetus passes into the bloodstream of the mother. New technology allows us to analyse this DNA directly from the mother's blood, and screen for specific chromosome abnormalities.

Chromosome abnormalities can arise when a baby inherits three copies of a specific chromosome (trisomy) instead of the usual two copies. Similarly, a monosomy occurs when only one copy of a chromosome is inherited. Both trisomies and monosomies can significantly affect the progression of a pregnancy, either leading to miscarriage or a genetic condition.

WHAT DOES THE NIPT TEST FOR?

The NIPT test screens for an extra copy (trisomy) of chromosomes 13, 18 and 21. NIPT also has the option of identifying whether a foetus is male or female, and if requested, can report on abnormalities in the number of sex chromosomes, or of the other chromosomes.

WHO CAN HAVE THE TEST?

Testing can be requested in women with a singleton or twin pregnancy from 10 weeks onwards, including IVF pregnancies conceived using the patient's own or donor eggs. Twin pregnancies can only be tested for trisomy of chromosomes 13, 18 and 21.

WHO SHOULD NOT HAVE THE NIPT TEST?

- If you have a malignancy (cancer) or have undergone a bone marrow/organ transplant and/or recent blood transfusion it may lead to incorrect results and the test is therefore not recommended.
- Women carrying more than 2 babies (for example triplets).

IS THERE AN AGE LIMIT FOR THE NIPT TEST?

No. All pregnant women of any age and/or risk category can have the NIPT test. Although the frequency of chromosomal trisomies increases with maternal age, they can happen at any age and in any pregnancy. Your doctor will inform you about your risk level.

ADVANTAGES OF HAVING THE NIPT TEST

- Highly accurate: >99% detection rate for detecting trisomy 21, 18 and 13.
- Safe: Only a blood specimen is required from the mother with no risk to the baby.
- The test can be performed during any stage of the pregnancy from 10 weeks onwards.
- Results are available in 7 working days from the time of the blood specimen reaching the laboratory.

MY RESULTS AND WHAT THEY MEAN

Your NIPT result will be communicated directly to the doctor requesting the test, within 7 working days. Your doctor will explain the result to you and answer any questions you may have. Genetic counselling can also be arranged at Ampath, if required.

Results reported as "No anomaly detected" indicate that the chances of the baby having that specific chromosome abnormality is low. However, the NIPT test is a screening test and therefore does not completely eliminate the possibility of abnormalities of the tested chromosomes, or the possibility of other chromosome abnormalities or other genetic or birth defects that may be present.

Results reported as "Anomaly detected" indicate that there is an increased risk that the baby has the specified chromosome abnormality. "Anomaly detected" in a twin pregnancy indicates that at least one baby is at risk of having the specified chromosomal abnormality.

It is important to remember that NIPT is a screening test, and your doctor will offer additional diagnostic testing (for example an amniocentesis or placental biopsy) to confirm an "Anomaly detected" result. Your doctor may also refer you for genetic counselling to discuss the implications and choices available to you and your baby.

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LIMITATIONS OF THE NIPT TEST

- NIPT only reports on an extra copy (trisomy) of chromosomes 21, 18 or 13, with or without sex chromosome abnormalities, or other chromosomes, depending on which testing option is requested by your doctor.
- NIPT cannot rule out the possibility that other genetic abnormalities may be present, nor does it guarantee a healthy baby.
- Twin pregnancies:
  - ◊ Chromosome abnormalities can be detected by this test but cannot be attributed to a specific baby in a twin pregnancy.
  - ◊ If a Y chromosome is detected (male), at least one twin is male, but the gender of each individual twin cannot be determined by the test.
- There is a small possibility that the test results might not reflect the chromosomes of the baby, but instead chromosome changes in the placenta only, or in the mother.
- In the case of uncertain/inconclusive test results, the test result must be confirmed by invasive prenatal diagnosis (amniocentesis or placental biopsy).
- Although rare, there is a chance that a result will not be obtained due to the amount of the baby's DNA in the mother's blood stream being too low (low foetal fraction). Under these circumstances, an additional specimen may be requested.

MEDICAL AID COVER FOR THE TEST

Medical aid schemes may cover the cost of the test (partial or in full) if it is deemed medically necessary and meets specific criteria. This may require submitting a motivation with supporting documentation from your doctor as part of pre-authorisation. In some instances, the NIPT test may be funded from your medical savings or need to be paid for upfront. It is important that you contact your medical aid to find out whether the NIPT is covered as part of your maternity benefit.

MORE QUESTIONS?

Should you require additional information, please ask your doctor. You can also contact our experts at the NGS Laboratory at 012 678 0645 or email nipt@ampath.co.za.

AMPATH APP

Your test results are available to you through our easy-to-use Ampath App

*Certain test results require an explanation by your doctor first and will only be available on the App later.

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