LABUPDATE no. 37

February 2023

NON-INVASIVE PRENATAL TESTING: AN UPDATE

INTRODUCTION

Non-invasive prenatal testing (NIPT) is a prenatal screening test that is performed using maternal peripheral whole blood samples. The test calculates the risk of a foetus being affected with certain chromosomal abnormalities. From February 2023, Ampath will be offering the CentoNIPT® test, performed by CENTOGENE in Rostock, Germany. CentoNIPT® uses the VeriSeq™ NIPT Solution v2 from Illumina based on next-generation sequencing technology and is CE-IVD approved. Cell-free DNA (cfDNA) fragments in maternal blood are sequenced and analysed to report over-representations (trisomies) of chromosomes 21, 18 and 13. Foetal gender and sex chromosome aneuploidies (monosomy X, trisomy X, XXY and XYY) can also be detected.

ELIGIBILITY

- Both singleton and twin pregnancies, including pregnancies conceived using IVF and self/donor eggs are eligible from 10 weeks of gestation onwards.
- NIPT is not recommended in cases of recent maternal blood transfusion, maternal malignancy, organ transplants and stem cell therapy as this could lead to false negative or false positive results.

TEST OPTIONS

Singleton/twin pregnancy	Mnemonic	Price*	Specimen type and TAT*
Chromosomes 21, 18, 13 with/without foetal gender	NIPT1	R5 350	1 X Full streck tube TAT: 10 working days
Chromosomes 21, 18, 13 AND sex chromosome aneuploidies with/ without foetal gender	NIPT2	R6 000	

* Price: For upfront payment. For medical aid reimbursement, scheme rates apply.
* TAT: Turnaround time from being received in the NRL NGS laboratory, Centurion.

Please note: Testing for microdeletions will only be performed on specific request.

SENSITIVITY AND SPECIFICITY FOR THE DETECTION OF TRISOMY 21, 18 AND 13*

	Trisomy 21	Trisomy 18	Trisomy 13
Sensitivity	>99.9%	>99.9%	>99.9%
Specificity	99.9%	99.9%	99.9%

*Reference: VeriSeq NIPT Solution v2 package insert, p 45

INTERPRETATION OF RESULTS

Results are reported as **ANOMALY DETECTED** or **NO ANOMALY DETECTED**:

No anomaly detected There is a low risk that the foetus has the genetic condition tested for.	**Limitations** • This result does not eliminate the possibility of chromosomal abnormalities of the tested chromosomes (false negative). • It also does not eliminate the possibility that the pregnancy has other chromosomal abnormalities (for example microdeletions), genetic conditions or other birth defects.
Anomaly detected There is a high risk that the foetus has a genetic condition (specified) tested for.	**Limitations** • NIPT is a screening test. A high-risk test result must therefore be confirmed by performing diagnostic testing on a foetal sample (such as a chorionic villus sample (CVS) or amniocentesis sample). • False positive: The results might not reflect in the chromosomes of the foetus, but instead reflect chromosomal changes to the placenta (confined placental mosaicism), or the mother (chromosomal mosaicism). • Chromosomal aneuploidies are detectable in twin gestations, but cannot differentiate between individual foetuses. Sensitivity and specificity are limited in twin gestations.
Foetal gender Result reported: Male/female	**Limitations** • In twin gestations, the presence of a Y chromosome indicates that at least one twin is male. • In case of organ transplantation from a male donor to the mother, the sex chromosome status for the foetus cannot be determined.

For more information or assistance, contact the NGS Laboratory at **012 678 0645** or email **nipt@ampath.co.za**.