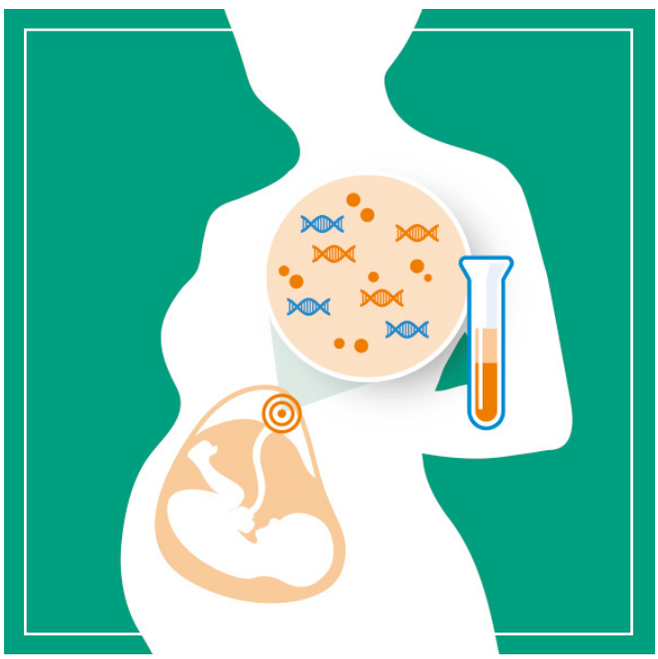
THE PATHCARE NEWS

Non-Invasive Prenatal Testing for Chromosome Abnormalities

Non-Invasive Prenatal Testing (NIPT) is a safe and accurate screening test for Down syndrome and other common chromosome abnormalities from maternal blood.



Placental DNA circulates freely in the maternal blood stream which can be sampled by venipuncture of the expecting mother. NIPT is able to test for extra chromosome 13, 18 or 21 material in foetal placental DNA. The foetal DNA originates from placental trophoblasts. NIPT has been shown to have significantly higher accuracy than other non-invasive screening tests such as nuchal translucency ultrasound (higher sensitivity and higher specificity). Using a screening test with a higher detection rate leads to fewer undetected cases while a higher specificity leads to fewer unnecessary invasive procedures.

American College of Medical Genetics and Genomics 2016 position statement:

"Clinical validation strongly suggested that NIPT can replace conventional screening for Patau (trisomy 13), Edward (trisomy 18) and Down (trisomy 21) syndromes. Objective measures of clinical utility support this. Test metrics support NIPT across the maternal age spectrum and continuum of gestational age beginning at 9–10 weeks as long as patients are not significantly obese."

NIPT remains a screening test and results indicating an aneuploidy should always be confirmed by a diagnostic test (Karyotyping, QF-PCR or FISH) by performing CVS or amniocentesis if termination of pregnancy is being considered. Pre- and/or post-test genetic counselling can be facilitated by PathCare. Please contact our helpdesk for details of a genetic counsellor in your area: Helpdesk 021 596 2130 or helpdesk@pathcare.co.za

NIPT is not indicated for:

- screen for single-gene disorders
- predict late pregnancy complications
- screen for open neural tube defects; therefore, maternal serum α-fetoprotein testing should still be offered at 15–20 weeks of gestation
- replace routine foetal anatomic screening using ultrasound

PathCare offers the following NIPT options performed by accredited laboratories:

- Harmony™ (UK) by Ariosa Diagnostics®
- TriScreen™ (UK) by Genesis Genetics®/NextBiosciences
- Panorama™ (USA) by Natera®

	Non-Invasive Prenatal Tests available via PathCare				Conventional First Trimester Screening
	Company	**Ariosa: TDL**	**NextBio: Genesis Genetics**	**Natera**	**Serum PAPP-A , B-hCG & NT**
	NIPT product	**Harmony**	**TriScreen**	**Panorama**	
Indications and limitations:	**Twin/multiple pregnancy * Donor egg/surrogate**	Yes	Yes	Yes	
	Detects Triploidy	No	No	Yes	
	Down syndrome (Trisomy 21) DR False Positive Rate	> 99% < 0,1%	> 99% < 0,1%	> 99% < 0,1%	85% [1] 5.4% [1]
	Edward syndrome (Trisomy 18) DR False Positive Rate	97.40% < 0.1%	97.40% < 0,1%	96% < 0.1%	
	Patau syndrome (Trisomy 13) DR False Positive Rate	93.80% < 0,1%	98.15% < 0,1%	99% < 0,1%	
	Turner syndrome (Monosomy X) DR False Positive Rate	96% 0.23% [2]	95% 1%	92% < 0.1%	
	Detection of other sex chromosome syndromes: XXY (Klinefelter syndrome), XYY (Jacobs syndrome), Triple X syndrome	Yes	Yes	Yes	
	Cost (standard rates, subject to change) standard testing	R 6 560.00	R 6 450.00	R 6 950.00	
	Detection of selected Microdeletions ** $ Routine screening for microdeletions should not be performed.	Yes	Yes	Yes	
	All chromosome screening $ (expanded autosomal trisomies) Indicated for women who decline an invasive procedure **in the presence of an abnormal ultrasound.**	No	Yes	No	
	Testing available from (GA weeks)	10	10	9	

** DR = Detection Rate*

The above information was obtained from each company's website and is subject to change.

1. Norton et al. N Engl J Med. 2015 Apr 23;372(17):1589-97.
2. Gil et al. Ultrasound Obstet Gynecol. 2015 Mar;45(3):249-66.

* Expanded panels (microdeletion panel and all chromosome screening) are not available for multiple pregnancies.

** See the following position statements:

The American College of Obstetrics and Gynaecologists Committee Opinion (Reaffirmed 2017):

"Screening for these microdeletions has not been validated in clinical studies, and the true sensitivity and specificity of this screening test is uncertain. **Routine cell-free DNA screening for microdeletion syndromes should not be performed."**

American College of Medical Genetics and Genomics 2016 position statement:

"Obstetric care providers should inform their patients of the **higher likelihood of false-positive and false-negative results for microdeletion conditions.** Obstetric care providers should inform their patients of the potential for results of conditions that, once confirmed, may have an uncertain prognosis."

$ Additional costs apply.

Turn around time: 10 - 15 working days

Please note that incomplete information/documentation cause delays and that accurate patient information is critical for analysis.

Sample: Product specific kits are available at selected PathCare depots, please confirm stock with the nearest depot before requesting a test.

Medical insurers may cover the cost of NIPT, patients are advised to request information from their insurer. Patients can contact the PathCare debtors call centre at 0860 100 442 to request an amended invoice for possible reimbursement (patients to submit their own claim to their medical aid) and to confirm pricing.