LABUPDATE no. 65

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August 2025

NON-INVASIVE PRENATAL TESTING: CONFIRMATORY TESTING UPDATE

Non-Invasive Prenatal Testing (NIPT) has become an invaluable screening tool in modern obstetric care. By analysing cell-free foetal DNA in maternal blood, NIPT offers a highly sensitive and specific method for detecting common foetal aneuploidies such as trisomy 21, 18, and 13, as well as sex chromosome abnormalities. Its non-invasive nature, early availability from 10 weeks' gestation, and superior performance compared with traditional biochemical screening make NIPT an attractive first-line screening option, particularly in high-risk pregnancies. Importantly, while NIPT significantly reduces the need for unnecessary invasive procedures, it remains a screening and not a diagnostic test.

As with any screening modality, NIPT findings that indicate a high probability of chromosomal abnormalities must be followed by confirmatory diagnostic testing. Chorionic villus sampling (CVS) or amniocentesis remains the gold standard for definitive diagnosis. It is crucial that patients receive accurate information to support informed decision-making, and this is best facilitated through comprehensive pre- and post-test genetic counselling. Such counselling ensures that patients understand the implications of their results, the limitations of the testing, and the possible outcomes and choices available to them.

Ampath Laboratories is proud to announce a new initiative to support patients and clinicians in navigating high-risk NIPT results. As part of our commitment to cost-effective and patient-centred care, we will now offer confirmatory testing (see table below) as well as genetic counselling at **no additional charge** for cases where a high-risk result is returned on any NIPT test performed through Ampath. This initiative aims to reduce financial barriers to diagnostic testing and to reinforce our dedication to providing comprehensive, ethical, and accessible prenatal care.

TABLE: HIGH-RISK NIPT CONFIRMATORY TESTING INFORMATION

	High-risk NIPT finding	Free confirmatory testing offered
Confirmatory testing offered based on specific high-risk NIPT results*	Trisomy 13	Aneuploidy PCR
	Trisomy 18	Aneuploidy PCR
	Trisomy 21	Aneuploidy PCR
	Monosomy X	Aneuploidy PCR and karyotype
	Any other sex chromosome aneuploidy	Aneuploidy PCR
	Other autosomal aneuploidy	Chromosomal microarray
Sample types accepted	CVS or amniotic fluid (15-20ml) Cord/postnatal blood will also be accepted in cases (such as sex chromosome aneuploidies), where invasive testing is declined despite a high-risk result.	
Process	All free confirmatory testing and counselling can be arranged by contacting **geneticsclinic@ampath.co.za** The general mnemonic CONNIPT will be used for all free confirmatory testing.	

*Note: These free services are only offered for high-risk NIPT results obtained through Ampath.