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AMPATHCHAT

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Non-invasive prenatal testing: Introducing VERACITY®

Our new NIPT test - continuing to deliver accurate, safe and reliable results

Introduction

Non-invasive prenatal testing (NIPT) is a prenatal screening test that calculates the risk of a fetus being affected with a chromosome abnormality. The test utilises cell-free DNA released from the placenta into the maternal bloodstream during pregnancy. NIPT is the most sensitive and specific screening test for the common fetal aneuploidies and reduces the need for invasive prenatal testing, with its associated risks.

VERACITY® overview

The VERACITY® NIPT was designed and developed by NIPD Genetics, a European biotechnology company, in 2015[1]. This new generation approach is based on proprietary target capture enrichment to overcome the limitations of current technology. The novel, targeted protocol measures fetal fraction accurately, providing increased sensitivity and specificity compared with other NIPT technologies. VERACITY® has been validated in two large multi-centre studies.[2,3] Ampath has concluded a technology transfer agreement with NIPD Genetics, and will be offering VERACITY® at Ampath's National Reference Laboratory.

VERACITY® scope and options

Three testing options (with different pricing/mnemonics) are available, and the selection should be made in the context of the clinical indication and pre-test discussion with the patient.

		VERACITY CORE	VERACITY PLUS	VERACITY PREMIUM
Mnemonic		VERACY	VERACYS	VERACYSM
Condition and prevalence				
TRISOMIES	Trisomy 21 (Down Syndrome) (1:700)	✔	✔	✔
	Trisomy 18 (Edwards Syndrome) (1:5 000)	✔	✔	✔
	Trisomy 13 (Patau Syndrome) (1:16 000)	✔	✔	✔
SEX	Presence of Y (Fetal sex)	✔*	✔	✔
SEX CHROMOSOME ANEUPLOIDIES	Turner Syndrome (1:2 000) Triple X Syndrome (1:1 000) Klinefelter Syndrome (XXY) (1:500–1 000) XYY, XXYY		✔**	✔**
MICRODELETIONS	Del (22q11.2) (Di George Syndrome) (1:1 000)			✔
	Del (1p36) (1p36 Deletion Syndrome) (1:5 000)			✔
	Del (17p11.2) (Smith-Magenis Syndrome) (1:15 000)			✔
	Del (4p) (Wolf-Hirschhorn Syndrome) (1:50 000)			✔

* Optional
** Sex chromosome aneuploidies cannot be determined for twins

AMPATH LABORATORIES

Clinical performance of VERACITY®

Based on a mixed risk population cohort, the specificity and negative predictive value of the VERACITY® test is >99.9%. The sensitivities and positive predictive values are shown in the table below:

	Sensitivity	Positive predictive value (PPV)
Trisomy 13,18,21	>98%	>97% (trisomy 21); >76% (trisomy 18); >60% (trisomy 13)
Sex chromosome aneuploidies	>75%	>82%
Microdeletions	>60%	>55%

How is VERACITY® performed?

From 10 weeks of pregnancy

Discussion, consent and option choice made with referring doctor

Samples collected at Ampath Depot
NB: 2 STRECK tubes are required

Sample analysed at Ampath

Results sent to referring doctor within 7–10 working days

VERACITY® reporting

Results are clearly reported for ease of interpretation, as illustrated in the example below:

VERACITY PRENATAL SCREENING TEST RESULTS		
	CONDITION	**REMARK**
Low-risk NIPT results	**Trisomy 21**	The results show low risk for Trisomy 21
	Trisomy 18	The results show low risk for Trisomy 18
	Trisomy 13	The results show low risk for Trisomy 13
	Trisomy X	The results show low risk for Trisomy X
	Monosomy X	The results show low risk for Monosomy X
	XXY Constitution	The results show low risk for XXY Constitution
FETAL FRACTION	**XYY Constitution**	The results show low risk for XYY Constitution
7.0%	**XXYY Constitution**	The results show low risk for XXYY Constitution
	Microdeletions: (DiGeorge, 1p36 Deletion Syndrome, Smith-Magenis, Wolf Hirschhorn)	The results show low risk for microdeletions (DiGeorge (22q11.2), 1p36 Deletion Syndrome, Smith-Magenis (17p11.2), Wolf Hirschhorn (4p16.3))
	Presence/absence of Y chromosome	The results show the presence of Y chromosome
INTERPRETATION		
The results show low risk for all tested conditions. The fetal fraction is 7.0%, which is sufficient for analysis. Please consult Test Method and Test Description overleaf for information on the method, performance and limitations of the test. The results should be communicated by the referring clinician with appropriate counselling.		

Eligible for the VERACITY® test

- Singleton pregnancies from 10 weeks gestation including pregnancies conceived using a donor egg or surrogate
- Twin or vanishing pregnancies (test mnemonic: Veracy or Veracysm)

Limitations of VERACITY®

- Testing for sex chromosome aneuploidies is not possible for twin or vanished twin pregnancies
- The test is not intended or validated for mosaicism, triploidy, partial trisomy or translocations
- Twin or vanishing twin IVF pregnancies conceived using a donor egg or surrogate are not eligible for the test
- Patients with malignancies, bone marrow or organ transplants, or recent blood transfusion are not eligible for the test

NIPT remains a screening test and positive results should be followed up by an invasive diagnostic test such as amniocentesis. Ampath offers prenatal diagnostic testing, including karyotyping and FISH, to confirm high risk NIPT results.

Genetic counselling, by an HPCSA registered genetic counsellor at Ampath, is also available on referral basis. This is to assist with the counselling and support of families around testing, results and decision-making. To make an appointment for these counselling services, please contact us on 012 678 0645. For any other NIPT queries please email: nipt@ampath.co.za

References: [1]PMID: 27117469, [2]PMID: 28158200, [3]PMID: 31338126