NIPT Report

PATIENT DETAILS	
Patient Name:	**Internal Code No.:**
Medical Record No.:	**Date of Birth:**

REFERRER DETAILS	
Referring Clinic Name:	
Referring Clinician Name:	**Clinic Postcode:**

SAMPLE AND TEST DETAILS	
Indication for Testing:	**Primary Sample Type:**
Gestation:	**Pregnancy Type:**
Blood Draw Date:	**Repeat Sample:**
Requested Test: PRENATALSAFE 5	**Acceptance Date:**
Test Start Date:	**Test End Date:**

TEST METHODOLOGY
Non-invasive prenatal testing (NIPT) using VeriSeq Solution v2 (Illumina) for the detection of trisomy 13, 18 , 21 and sex chromosome abnormalities.

RESULT AND INTERPRETATION		
	ANOMALY	**TEST FINDINGS**
RESULT	TRISOMY 21	not detected
	TRISOMY 18	not detected
	TRISOMY 13	not detected
	XO/XYY/XXY/XXX	not detected
Fetal Sex:		**Fetal Fraction:**

Interpretation: Low chance of trisomy 13, trisomy 18, trisomy 21, Triple X, Monosomy X, XXY, or XYY.

RESULT AUTHORISATION	
Report Release Date:	
Signed by:	**Authorised by:**
Job Title:	**Job Title:**

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TECHNICAL SUMMARY

Test Details:

VeriSeq NIPT Solution v2 is a CE IVD marked in vitro diagnostic test intended for use as a screening test for the detection of foetal genetic anomalies from maternal peripheral whole blood specimens in pregnant women of at least 10 weeks gestation. This test is a screening test which does not replace diagnostic tests, e.g. foetal karyotyping, and uses whole genome sequencing to detect aneuploidy status of chromosomes. A negative result does not fully exclude the possibility for the foetus to be affected. The Limit of Detection (LOD) of the method is at a foetal fraction greater than or equal to 2% (Pertile *et al.*, 2021 PMID: 34077512). If the foetal fraction is not sufficient or the data obtained do not allow a univocal interpretation, a new sample will be requested to repeat the analysis. A low-chance result does not completely exclude the presence of one of the chromosomal abnormalities investigated. The assay might detect abnormalities in other chromosomes which will not be reported. The test result may not reflect the real state of the foetus, as it cannot detect chromosomal abnormalities arising from confined placental mosaicisms, vanishing twin or maternal condition.

Test Limitation:

The VeriSeq NIPT Solution v2 is a screening test and should not be considered in isolation from other clinical findings and test results. The test carries a residual risk of false positive and false negative results (0.1%) and is neither intended nor validated for diagnosis. Conclusions about the foetal condition and pregnancy management decisions should not be based on the results of the NIPT screening alone. This test is not validated for pregnancies with more than two foetuses and is not designed to detect chromosomal mosaicisms or polyploidy, such as triploidy. The result of this test does not exclude the possibility that the chromosomes harbour abnormalities other than those included in the test, and it does not detect abnormalities of untested chromosomes, genetic disorders, birth defects or congenital complications of other origin. The test result may not reflect the real state of the foetus, as it can detect chromosomal abnormalities arising from confined placental mosaicisms, vanishing twin or maternal condition. The results of the test can be confounded by certain maternal and foetal factors including but not limited to: recent maternal blood transfusion; maternal organ transplant; maternal surgical procedure; maternal immunotherapy or stem cell therapy; maternal malignancy; maternal mosaicism; foetal placental mosaicism; foetal demise; nonviable twin.

Sensitivity, specificity, and limitations: Pertile *et al.*, 2021. Concordance of the test for trisomy 13, trisomy 18 and trisomy 21 is 99.9% for singleton pregnancies (Pertile *et al.*, 2021). For foetal sex, concordance was 100% for both ‘female’ and ‘male’ (based on newborn physical exam) and for XX and XY (based on cytogenetic results). «Limitation» Sequencing data was generated using on Illumina NextSeq 550Dx platform and bioinformatic analysis and data analysis was performed using VeriSeq NIPT Assay Software v2.0 (Illumina).

CONTACT INFORMATION

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