Non-Invasive Prenatal Test

(Illumina VeriSeq NIPT Solution v2 – performed in the UK)

Name	EXAMPLE-REPORT	**Account**	TEST
Date of Birth (dd/mm/yyyy)	09/10/1983	**Cinic Name**	TEST
Lab Number	98G100155	**Referring Clinician**	TEST CLIN NAME
Gestational Age (weeks + days)	13+2	**Blood Collection date** (dd/mm/yyyy)	18/08/2023
Sample type	CF-BCT	**Sample Received date** (dd/mm/yyyy)	18/08/2023
Number of fetuses	1	**Report Date** (dd/mm/yyyy)	22/08/2023
Indication	Maternal Request.		

NIPT Result

Fetal cfDNA Percentage: 5%

Test	Result	Recommendation
Trisomy 21 (T21)	**HIGH PROBABILITY**	**Genetic counselling and further testing**
Trisomy 18 (T18)	Low Probability	Review results with patient
Trisomy 13 (T13)	Low Probability	Review results with patient

Interpretation

Testing of cell-free DNA shows an increased probability of **TRISOMY 21**. As NIPT is a screening test only, this result should be confirmed by fetal chromosome studies (CVS or amniocentesis) prior to any irreversible management decision about this pregnancy.
Genetic counselling is recommended to discuss this result.

Test Description and Limitations

Limitations - Non-invasive prenatal testing (NIPT) measures the relative abundance of cell-free DNA (cfDNA) from various chromosomes in a sample of maternal blood collected at 10 or more weeks gestation. The test screens for selected chromosome disorders and (optionally) sex in the fetus and a panel of sex chromosome aneuploidies. NIPT is a screening test for an uncommon event. The chance that a report of "low probability" in the fetus is correct (i.e. true negative) will vary with the pre-test probability of that disorder. The accuracy of this test is dependent on the provided gestational age and parity (as shown above); please contact the laboratory immediately if this information is incorrect. A non-invasive prenatal test (NIPT) based on a cell-free DNA analysis of the maternal blood is not a diagnostic test, but a screening test. The test results cannot therefore be used as the only basis for a diagnosis. Extensive further tests are necessary to verify the results in order to avoid irreversible decisions regarding pregnancy. This is true both in the case that the report sent to your doctor reveals an unremarkable result as well as if the report shows a high risk. Your doctor will discuss with you which tests can provide an unambiguous result. Biological factors which may affect the interpretation of results include but are not limited to; presence of an unknown demised / vanishing twin (patient is ineligible for testing if known), incorrect number of fetuses stated, maternal malignancy, maternal organ transplant or recent blood transfusion and maternal chromosome aneuploidy or mosaicism.

Method: This NIPT is performed using the Illumina VeriSeq Solution v2 test performed using an Illumina NextSeq Next Generation Sequencer at TDL Genetics, London UK. The bioinformatics pipeline used is Veriseq NIPT CE-IVD.

NIPT performance

	T21	T18	T13
Sensitivity	>99.9% (130/130)	>99.9% (41/41)	>99.9% (26/26)
2-sided 95% CI	97.1%, 100%	91.4%, 100%	87.1%, 100%
Specificity	99.90% (1982/1984)	99.90% (1995/1997)	99.90% (2000/2002)
2-sided 95% CI	99.63%, 99.97%	99.64%, 99.97%	99.64%, 99.97%

Further information regarding assay performance and PPV is available on our website www.tdlpathology.com/specialties/genetics/non-invasive-prenatal-testing-nipt/

Results are authorised by a Clinical Scientist (HCPC) and checked by a Molecular Biologist (these data are stored electronically).
Report produced by HSL Analytics (LLP).

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