NON-INVASIVE PRENATAL TEST REPORT

BIOGENIX LABS
By M42

PATIENT DETAILS		SAMPLE DETAILS		REFERRING FACILITY	
Patient Name:	XXXX	**Sample Collection Date:**	XXXX	**Referring Facility:**	XXXX
Patient EID/Passport:	XXXX	**Sample Received Date:**	XXXX	**Referring Clinician:**	XXXX
Date of Birth:	XXXX	**Sample Type:**	Peripheral Blood	**Assisting Clinician:**	-
Gestational Age:	XXXX	**No. of Fetuses:**	XXXX	**MRN:**	XXXX
EDC:	XXXX	**Report Date:**	XXXX		
Order ID:	XXXX				
Test Ordered:	6000050 - NIPT Comprehensive, with gender				
Test Indication:	-				

Test Results	**Positive**
Result Table	
Fetal Fraction (%)	10%
Fetal Sex	Consistent with male
Autosomal Anomaly	Detected
Sex chromosome Aneuploidy (SCA)	Not Detected
Anomaly Description	Detected: Trisomy 13

About the test: NIPT is a non-invasive prenatal screening test that analyzes circulating cell free DNA (cfDNA) from a maternal peripheral whole blood specimen. The NIPT screen test is indicated for use in pregnant women of at least 10 weeks gestation. The test offers two options for types of screening: Core and Comprehensive. The Core screening provides information on the aneuploidy status of chromosomes 21, 18, 13, X, and Y only. Comprehensive screenings provide partial duplications and deletions of at least 7 Mb for all autosomes and aneuploidy status for all chromosomes. Both screening types provide the option for sex chromosome aneuploidy (SCA) reporting with or without fetal sex reporting. Sex chromosome aneuploidy cannot be reported for twin samples, only presence or absence of Y chromosome will be reported (if fetal sex is requested).

Below are the test options and the results reportable based on test requested:

Test options	T21	T13	T18	SCA	Gender	Any Anomaly	RAA	Partial deletion/duplication
Singleton								
Core	✓	✓	✓	✓	✓	✗	✗	✗
Comprehensive	✓	✓	✓	✓	✓	✓	✓	✓
Twin								
Core	✓	✓	✓	✗	✓*	✗	✗	✗
Comprehensive	✓	✓	✓	✗	✓*	✓	✓	✓

*Reports as either “CHR Y PRESENT” or “NO CHR Y PRESENT”

Test Method: The test works by isolating the cfDNA (including both maternal and fetal DNA) from a maternal peripheral whole blood specimen and performing low-coverage whole genome sequencing using Next Generation Sequencing technology. Data is analyzed using bioinformatics algorithms, that generate quantitative scores to aid in the detection and differentiation of any fetal anomaly such as trisomies, monosomies, and partial deletions or duplications of at least 7 Mb or greater. The quantitative scores are log likelihood ratio scores associated with under-or-over representation of a target chromosome relative to an expectation for a diploid genome.

Test Performance: The information provided below is based on the study performed by the manufacturer. The performance for detecting trisomies 21, 18 and 13 was derived from outcomes in 2,307 singleton and twin pregnancies.

Sensitivity and Specificity:

	Trisomy 21	Trisomy 18	Trisomy 13	Any Anomaly	Rare Autosomal Aneuploidy	Partial Deletions and Duplications
Sensitivity	> 99.9% (130/130)	> 99.9% (41/41)	> 99.9% (26/26)	95.5% (318/333)	96.4% (27/28)	74.1% (20/27)
2-sided 95% CI	97.1%, 100%	91.4%, 100%	87.1%, 100%	92.7%, 97.3%	82.3%, 99.4%	55.3%, 86.8%
Specificity	99.90% (1982/1984)	99.90% (1995/1997)	99.90% (2000/2002)	99.34% (1954/1967)	99.80% (2001/2005)	99.80% (2000/2004)
2-sided 95% CI	99.63%, 99.97%	99.64%, 99.97%	99.64%, 99.97%	98.87%, 99.61%	99.49%, 99.92%	99.49%, 99.92%

Estimates for Trisomy 21, 18, and 13 in Simulated Population of Twin Pregnancies:

	Trisomy 21	Trisomy 18	Trisomy 13
Sensitivity	96.4%	95.7%	93.6%
2-sided 95% CI	(86.4%, 98.9%)	(68.3%, 99.4%)	(64.1%, 98.9%)
Specificity	99.9%	> 99.9%	> 99.9%
2-sided 95% CI	(99.8%, > 99.9%)	(99.9%, > 99.9%)	(99.9%, > 99.9%)

Percent Concordance for Fetal Sex Classification:

	Cytogenetic Results			
Fetal Sex Classification	XO	XXX	XXY	XYY
Percent Concordant	90.5%	100%	100%	91.7%

6000050 - NIPT Comprehensive, with gender	
PATIENT DETAILS	
Patient Name:	XXXX
Order ID:	XXXX

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Test Limitation: NIPT is a screening test, NOT a diagnostic test. The test does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis. A patient with a positive result should be referred for genetic counseling and recommended for invasive diagnostic tests for confirmation. A negative result does not ensure an unaffected pregnancy, nor does it exclude the possibility of other chromosomal abnormalities or birth defects which are not a part of these tests. Inaccurate test results or a failure to obtain test results may occur due to one or more of the following rare occurrences: courier/shipping delay; laboratory failure or error; biological factors such as but not limited to: insufficient sequencing coverage, noise or artifacts in the region, amplification or sequencing bias, or insufficient fetal fraction, sample contamination or degradation, mosaicism (a mixture of cells with normal and abnormal chromosomes) in the fetus, placenta or mother, recent maternal blood transfusion, prior maternal organ transplant, maternal neoplasms, or an unrecognized twin or vanishing pregnancy,; other circumstances beyond our control; or unforeseen problems that may arise. About 1 to 2% of all pregnancies have confined placental mosaicism, a situation in which the placenta has cells with a chromosome abnormality while the fetus has normal chromosomes or vice versa. This means that there is a chance that the chromosomes in the fetus may not match the chromosomes in the DNA screened. The assay is not intended to detect polyploidy, balanced chromosome rearrangements, and to identify pregnancies at risk for open neural tube defects. The test result is specific to the tested sample and should always be interpreted by a qualified professional in the context of clinical and familial data. The test result cannot be used as the sole basis for diagnosis or other pregnancy management decisions.

Disclaimer: The test was validated, and its performance characteristics determined by Biogenix Clinical Laboratory. This test is used for clinical purposes. It should not be regarded as investigational or research.

Reference:

• Bianchi D et al. N Engl J Med. 2014;370(9):799-808.
• Grati, et al. Genet Med. 2014;16: 620–624.
• ACOG Practice Bulletin No. 163. Obstet Gynecol. 2016;127(5):e123-137.
• ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

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