

Non-invasive Prenatal Test (NIPT) , BLOOD IN STRECK TUBE

PATIENT DETAILS:						
Pregnancy Status	Singleton	Gestational Age	17 Weeks			
Clinical Indication	-					
TEST RESULT SUMMARY						
Chromosomes	Screening Test Result					
Chromosome 13	Low Risk					
Chromosome 18	Low Risk					
Chromosome 21	Low Risk					
Sex Chromosomes	High Risk- Amniocentesis followed by Karyotyping recommended.					
Other Chromosomes	Low Risk					
INTERPRETATION						
Result	Interpretation					
Low Risk Aneuploidy	Low probability for the fetus to be affected with specific chromosomal aneuploidy					
High Risk Aneuploidy	High probability for the fetus to be affected with specific chromosomal aneuploidy. Confirmatory invasive diagnostic testing is recommended.					
Test Failed	Due to indefinite reasons result is not obtained with the given sample. Repeat sample testing is advised. Invasive testing is recommended if No Result of repeat sample.					
Disclaimer:						
1. This test does not report the gender of the fetus.						
2. Results are indicated for screening, NOT diagnosis. Results should be reviewed and discussed with your clinician. Pre and Post test Genetic counseling recommended.						
3. Clinical correlation with ultrasound findings, and other clinical data and tests is recommended. The test indicates the probability or provides the risk of the fetus being affected or unaffected, but does not it doesn't negate the possibility of chromosomal defect. Clinical decisions should not be solely based on the NIPT standalone test results. If definitive diagnosis is desired, amniocentesis is necessary.						
4. Although this test is highly accurate, there is still a small possibility for false positive or false negative results. This may be caused by technical and/or biological limitations, including but not limited to confined placental mosaicism (CPM) or other types of mosaicism, maternal constitutional or somatic chromosomal abnormalities, residual cfDNA from a vanished twin or other rare molecular events						
5. This test is not applicable in pregnant women with certain medical conditions like maternal aneuploidy, cancer, blood transfusion in past 12 months, transplant, history of stem cell therapy/immunotherapy						
6. The test does not identify mosaicism, balanced translocation, partial chromosome aneuploidy, polyploidy and neural tube defects.						
PCPNDT Registration Number: NMMC/PNDT/314						

If Values are marked with * , they are critical values.

*** End Of Report ***



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HOD – MOLECULAR

Patient Details			
Patient UHID	LDAA04225410	SIN No.	MO00077716
Gestational Age	17 weeks	Pregnancy	Singleton
Sample Information			
Fetal Fraction	4.45%		
Sample Information	Qubit Fluorometer (ng/ul):2.53; Volume (ul): 50; Total amount (ng): 126.5		
I. Screening results			
Chromosomes	Risk	Z score	Test Results
Chromosome 13		-2.71	Low Risk
Chromosome 18		-2.15	Low Risk
Chromosome 21		1.55	Low Risk
Sex Chromosomes	-5.64	High Risk - Further Investigation Recommended	
Other Chromosomes		Part II	Low Risk
			Part II

II. Other Chromosomes

Chromosome	Risk	Z score	Test Results	Reference interval
Chromosome 1		0.27	Low Risk	-6<Z score<6
Chromosome 2		-0.31	Low Risk	-6<Z score<6
Chromosome 3		1.21	Low Risk	-6<Z score<6
Chromosome 4		-0.97	Low Risk	-6<Z score<6
Chromosome 5		0.62	Low Risk	-6<Z score<6
Chromosome 6		0.07	Low Risk	-6<Z score<6
Chromosome 7		0.74	Low Risk	-6<Z score<6
Chromosome 8		-1.21	Low Risk	-6<Z score<6
Chromosome 9		-0.73	Low Risk	-6<Z score<6
Chromosome 10		0.12	Low Risk	-6<Z score<6
Chromosome 11		-0.75	Low Risk	-6<Z score<6
Chromosome 12		1.30	Low Risk	-6<Z score<6
Chromosome 14		-0.53	Low Risk	-6<Z score<6
Chromosome 15		-0.35	Low Risk	-6<Z score<6
Chromosome 16		-1.29	Low Risk	-6<Z score<6
Chromosome 17		-0.27	Low Risk	-6<Z score<6
Chromosome 19		0.86	Low Risk	-6<Z score<6
Chromosome 20		-1.16	Low Risk	-6<Z score<6
Chromosome 22		0.14	Low Risk	-6<Z score<6

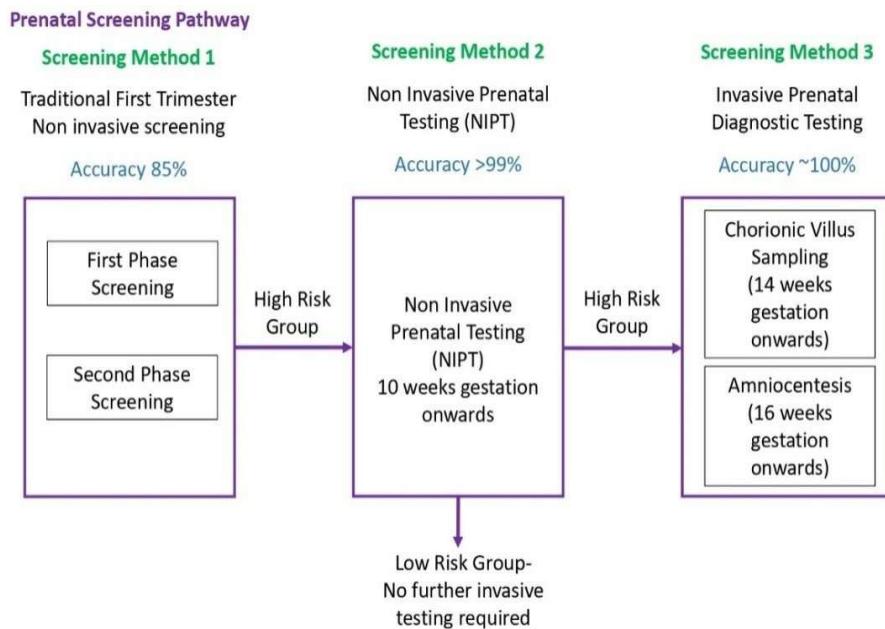
 Low Risk;  High Risk — Further Investigation Recommended

IV. Supplementary information

1. The NIPT test screens a maternal blood sample for chromosome aneuploidy in placental DNA using the following methodology:
 - (1) Extraction of cell-free placental DNA from the maternal blood sample
 - (2) High throughput sequencing of the extracted cell-free placental DNA
 - (3) Calculation of molecular mass of placental DNA in all chromosomes
2. The method is intended for use in pregnant women who are at least 10 weeks of pregnancy. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA.
3. Based on the scope, the NIPT test can detect the following:
Whole Genome - 23 pairs of human chromosomes
Sex chromosomal aneuploidies: XO, XXX, XXY/XYY
4. The test is capable of genome-wide aneuploidy detection over the whole fetal genome and gives the results for 23 pairs of chromosomes. This test confers an accuracy of up to 99% on the detection of fetal aneuploidy for chromosomes 13, 18 and 21. In a study of over 2000 samples, 6 samples were determined to be at high-risk of having an autosomal aneuploidy other than 13, 18 and 21. This is a prevalence rate of 0.3%, which is consistent with prevalence in published studies.

Results are indicated for screening, NOT diagnosis. – (Results should be reviewed and discussed with your clinician.)

This test does not report the gender of the fetus.



About NIPT prenatal screen

The NIPT prenatal screen is a new advanced non-invasive prenatal screening solution using the latest developments in DNA technology to detect placental DNA in maternal blood. It offers a menu-based chromosome analysis to estimate the risk of a fetus having Down's syndrome and other genetic disorders. Enabling pregnant women and their families fast, safe and reliable results and reducing the need for invasive tests and the associated risks, stress and anxiety. NIPT Prenatal screen is indicated for use in pregnant women who are at least 10-week pregnant. Chromosomal aneuploidy can then be detected using bioinformatics analyses, where the detection rate and sensitivity are over 99%.

Limitations

NIPT prenatal screen is a screening test and all high-risk results should be confirmed through further investigation which may include tests such as amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a high-risk result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. Pregnant women with a negative test result do not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes (micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy). The test is not reportable for known multiple gestations, or if the gestational age is less than 10 weeks.

Test method

A simple maternal blood sample is taken from the pregnant mother from 10-week gestation with no risk to the fetus. Circulating cell-free placental DNA was purified from the plasma component of anti-coagulated 10mL of maternal whole blood. It was then converted into a genomic DNA library for Next Generation Sequencing and then determination of chromosomal aneuploidy.

References:

1. Obstet Gynecol 2012;119:890-901.
2. BMJ 2011;342:c7401.
3. Prenat Diagn 2012;32:c7401.
4. ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

