ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: Female

Specimen Collected: 23-May-22 08:41

Non-Invasive Prenatal Aneuploidy by NGS | **Received: 23-May-22 08:41** | **Report/Verified: 23-May-22 08:57**

Procedure	Result	Units	Reference Interval
Multiple Gestation	No		
Gestational Age at Draw	10 wks or over		
Report Fetal Sex	Yes		

Non-Invasive Prenatal Aneuploidy by NGS | **Received: 23-May-22 08:41** | **Report/Verified: 23-May-22 12:20**

Procedure	Result	Units	Reference Interval
Fetal Fraction	10.1	%	
Trisomy 21	**High Risk** *		
Trisomy 18	Low Risk		
Trisomy 13	Low Risk		
Sex Chromosome Trisomies and Monosomy X	Low Risk		
Fetus Sex	Female		
Result Summary	High Risk f1 i1		
EER Non-Invasive Prenatal NGS Aneu	See Note f2		

Result Footnote

f1: Result Summary
This pregnancy is classified as HIGH RISK for trisomy 21 (Down syndrome) by this screen. This result should be confirmed by a diagnostic test. The chance that a pregnancy classified as high risk will have the screened condition (the positive predictive value, or PPV) is affected by the pre-test risk of the screened condition. For women with no additional risk factors, 50-67% of pregnancies classified as high risk by NIPT are found to have trisomy 21. For women with a high pre-test risk of Down syndrome, 83-95% of pregnancies classified as high risk by NIPT are found to have trisomy 21. Online calculators are available to determine patient-specific PPV based on clinical context.

This is a screening test and is NOT diagnostic for the condition(s) listed in this report. Both false positive and false negative results may occur. Appropriate clinical follow-up such as genetic counseling, comprehensive ultrasound, and confirmatory diagnostic testing should be undertaken as recommended by the patient's healthcare provider. Irrevocable action such as pregnancy termination should not be taken based on the results of this screening test.

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

This result has been reviewed and approved by [redacted]

f2: EER Non-Invasive Prenatal NGS Aneu
Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

Test Information

i1: Result Summary
INTERPRETIVE INFORMATION: Non-Invasive Prenatal Aneuploidy

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Test Information

i1: Result Summary

Screen by cell-free DNA Sequencing

CHARACTERISTICS: This assay is a screening test that interrogates chromosomal abnormalities (i.e., aneuploidies) using cell free DNA (cfDNA) extracted from the blood plasma of any singleton pregnancy. Patient risk for trisomy 13, trisomy 18, trisomy 21, and sex chromosome aneuploidies is reported. Fetal fraction, in conjunction with other data quality metrics, must be met in order for each sample to yield a result. The assay is intended for use as a screen only and is not equivalent to prenatal genetic diagnostic testing.

METHODOLOGY: Next Generation Sequencing (NGS) (aka Massively Parallel Sequencing (MPS)) of fetal and maternal cfDNA present in the plasma.

ANALYTICAL VALIDATION ACCURACY: The analytical sensitivity was calculated using positive percent agreement compared to established methods to detect fetal aneuploidy. For samples with greater than 5 percent observed fetal fraction, the positive percent agreements (PPA) are as follows: T13 greater than 99.9 percent, T18 greater than 99.9 percent, and T21 is 96.1 percent. The combined PPA for all aneuploidies is 97.5 percent. For samples with less than or equal to 5 percent observed fetal fraction, the positive percent agreements (PPA) are as follows: T13 is 66.7 percent, T18 is 60 percent, and T21 is 87.5 percent. The combined PPA for all aneuploidies is 72.3 percent. The specificity, as calculated as negative percent agreement, is 99.5 percent across all observed fetal fraction values.

CLINICAL PERFORMANCE: Information on clinical performance for this assay can be found in the following reference: Borth H. Analysis of cell-free DNA in a consecutive series of 13,607 routine cases for the detection of fetal chromosomal aneuploidies in a single center in Germany. Arch Gynecol Obstet. 2021;303(6):1407-1414.

LIMITATIONS: This is a screening test and should not be considered in isolation from other clinical findings and diagnostic test results. High risk results must be confirmed by diagnostic testing (amniocentesis, CVS, or postnatal testing) before any clinical decisions are made based on the screening test result. The current iteration of this assay is limited to reporting the following on singleton pregnancies: fetal sex, fetal fraction, risk level for trisomy 13, 18, 21, and risk level for sex chromosome aneuploidies XO, XXX, XXY, and XYY. This assay is not meant to detect deletions or duplications within a chromosome, polyploidy, maternal abnormalities, balanced chromosome rearrangements, or chromosomal aneuploidies not listed above. Results may be confounded by the following: recent maternal blood transfusion, organ transplant, surgery, immunotherapy, malignancy, maternal mosaicism, placental mosaicism, fetal demise, disappearing twin, fetal partial aneuploidy, and/or fetal mosaicism. Samples with observed fetal fraction less than 5.0 percent have lower sensitivity to detect fetal aneuploidy, and the accuracy of

Test Information

i1: Result Summary

the fetal fraction estimate is significantly lower. Fetal demise/miscarriage is not assessed.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.