Non-Invasive Prenatal Aneuploidy Screen by cell-free DNA Sequencing

Patient: LOW RISK, NIPT NGSAN
DOB: Not Provided Age: N/A Sex: F
Patient Identifiers: 38830

Visit Number (FIN): 39152

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108

Physician: TEST

ARUP Test Code: 3003043

Collection Date: 05/23/2022
Received in lab: 05/23/2022
Completion Date: 05/23/2022

Result Summary

Low Risk

Fetus Sex	Not Reported
Fetal Fraction	10.1%
Trisomy 21 (Down Syndrome)	Low Risk
Trisomy 18 (Edwards Syndrome)	Low Risk
Trisomy 13 (Patau Syndrome)	Low Risk
Monosomy X (Turner Syndrome)	Low Risk
Sex Chromosome Trisomies	Low Risk

This is a screening test and is NOT diagnostic for the conditions listed in this report. Both false positive and false negative results may occur. Based on the analysis of the circulating cell-free DNA, the risk that this fetus has any of the conditions listed in this report is LOW. If clinical findings conflict with these test results, appropriate clinical follow-up should be performed. This may include genetic counseling, ultrasound, amniocentesis, CVS or other testing as recommended by the patient's healthcare provider.

A published meta-analysis determined negative likelihood ratios of 0.003 for trisomy 21, 0.022 for trisomy 18, and 0.010 for trisomy 13 (PMID: 28397325). Positive predictive values (PPV) and negative predictive values (NPV) are affected by an individual's pre-test risks for each of the screened conditions. Online calculators are available to calculate patient-specific PPV and NPV using age and clinical information.

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]

Test Specific Patient Information

Report Sex:	No
Gestational Age at Draw:	10 wks or over
Number of fetuses:	Singleton

Background Information:

INTERPRETIVE INFORMATION: Non-Invasive Prenatal Aneuploidy 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

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

This test was developed and its performance characteristics

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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.

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