HOTLINE: Effective May 23, 2022

New Test **3003043** **Non-Invasive Prenatal Aneuploidy Screen by cell-free DNA Sequencing** **NIPT NGSAN**

Available Now

Non-Invasive Prenatal Aneuploidy Screening (NIPT/NIPS) Patient History Form

Methodology: Massively Parallel Sequencing
Performed: Varies
Reported: 5-7 days

Specimen Required: Patient Prep: Specimen must be collected at 10 weeks gestation or greater. Testing will be canceled for specimens collected at less than 10 weeks of gestation.
Collect: Black-and-tan top cell-free DNA BCT (Streck) Tube (ARUP Supply #56435) Available online through eSupply using ARUP Connect™ or contact ARUP Client Services at (800) 522-2787.
Specimen Preparation: Transport 10 mL maternal whole blood (Min: 7 mL)
Storage/Transport Temperature: Refrigerated
Remarks: Patient History and Consent Form for Non-Invasive Prenatal Aneuploidy Screening Test (NIPT/NIPS) form is available on the ARUP Web site or by contacting Client Services at (800) 522-2787.
Unacceptable Conditions: Ambient and frozen specimens.
Stability (collection to initiation of testing): Ambient: Unacceptable; Refrigerated: 10 days; Frozen: Unacceptable.

Reference Interval: N/A

Interpretive Data:
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 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% observed fetal fraction, the positive percent agreements (PPA) are as follows: T13 greater than 99.9%, T18 greater than 99.9%, and T21 is 96.1%. The combined PPA for all aneuploidies is 97.5%. For samples with less than or equal to 5% observed fetal fraction, the positive percent agreements (PPA) are as follows: T13 is 66.7%, T18 is 60%, and T21 is 87.5%. The combined PPA for all aneuploidies is 72.3%. The specificity, as calculated as negative percent agreement, is 99.5% 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% 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 determined by ARUP Laboratories. It has not been cleared or approved by the U.S. 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.

Note: Results will not be reported without a gestational age greater than or equal to 10 weeks. ARUP only performs testing on singleton pregnancies. Multiple pregnancies will be sent out to Integrated Genetics to perform the MaterniT21 PLUS Core test..

CPT Code(s): 81420

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.