TEST CHANGE

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

3003043, NIPT NGSAN

Specimen Requirements:	
Patient Preparation:	Specimen must be collected at 10 weeks gestation or greater. Testing will be canceled for specimens collected at less than 10 weeks of gestation. Number of fetuses must be provided. Testing will be canceled if number of fetuses is not provided.
Collect:	Black-and-tan top cell-free DNA BCT (Streck) Tube (ARUP Supply #56435) Available online through eSupply using ARUP Connect(TM) or contact ARUP Client Services at 800-522-2787.
Specimen Preparation:	Transport 10 mL maternal whole blood (Min: 7 mL) New York State Clients: Transport 20 mL maternal whole blood (Min: 16 mL)
Transport Temperature:	Refrigerated
Unacceptable Conditions:	Ambient and frozen specimens.
Remarks:	Patient History and Consent forms for the Non-Invasive Prenatal Aneuploidy Screening Test (NIPT/NIPS) are available on the ARUP Web site or by contacting Client Services at 800-522-2787.
Stability:	Ambient: Unacceptable; Refrigerated: 10 days; Frozen: Unacceptable. New York State Clients: Ambient: 5 days; Refrigerated: Unacceptable; Frozen: Unacceptable
Methodology:	Massively Parallel Sequencing
Performed:	Varies
Reported:	5-7 days
Note:	Results will not be reported without a gestational age greater than or equal to 10 weeks. Testing will not be performed without number of fetuses provided. ARUP only performs testing on singleton pregnancies. Multiple gestation samples will be sent to Integrated Genetics to perform the MaterniT21 PLUS Core (chr21,18,13) test.
CPT Codes:	81420

New York DOH Approval Status:	Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

Refer to report. ~~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.~~

Reference Interval:

N/A