Non-Invasive Prenatal Testing for Fetal Aneuploidy

ARUP LABORATORIES

Patient: DOB: Age: 35 Gender: F **Patient Identifiers:** **Visit Number (FIN):**	Client: Physician:	ARUP Test Code: 2007537 Collection Date: 01/30/2018 Received in lab: 01/30/2018 Completion Date: 01/30/2018

Patient Information Used in Risk Calculations

Maternal Age at Delivery: 36 years
Estimated Due Date: 19 Jun 2018
Gestational Age at Draw: 20 weeks 0 days
Maternal Weight: 150 lbs
Report Fetal Sex: Yes

Results Summary

HIGH RISK Trisomy 21

Fetal Fraction: **8.0%** Fetal Sex: **Female**

This pregnancy is classified as HIGH RISK for trisomy 21 (Down syndrome) by this screen. This result should be confirmed by a diagnostic test. On average, 91% of pregnancies classified as HIGH RISK are found to have trisomy 21 based on a published study of 17,885 women (PMID 25111587).

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. Genetic counseling and confirmatory fetal diagnostic testing is recommended. Irrevocable action such as pregnancy termination should not be taken based on the results of this test alone.

This result has been reviewed and approved by
Electronic Signature

Conditions Screened

Trisomy 21:	**HIGH RISK**
Trisomy 18:	Low risk
Trisomy 13:	Low risk
Monosomy X:	Low risk
Triploidy/Vanishing twin:	Low risk

Patient: | **Date of Birth:** | **Gender:** F | **Physician:**
Patient Identifiers: | **Visit Number (FIN):**

TEST INFORMATION: Non-Invasive Prenatal Testing for Fetal Aneuploidy (Powered by Constellation)

METHODOLOGY: DNA isolated from the maternal blood, which contains placental DNA, is amplified at >13,300 loci using a targeted PCR assay and sequenced using a high-throughput sequencer. Sequence data are analyzed using Natera's Constellation software to estimate the fetal copy number for chromosomes 13, 18, 21, X, and Y, thereby identifying whole chromosome abnormalities at those chromosomes as well as fetal sex. Barring QC failures and fetal fractions below the performance limits of the algorithm, the minimum confidence threshold is 0.98 for a High Risk call, and 0.85 for a Low Risk call. For both Low Risk and High Risk calls, the majority of specimens will have a confidence of >0.99 across all regions tested. If a sample fails to meet the quality threshold, no result will be reported for one or more chromosomes.

SENSITIVITY AND SPECIFICITY: For combined autosomal aneuploidies and Turner's syndrome, sensitivity and specificity are >99 percent. Fetal sex has a sensitivity and specificity of >99 percent. Sex chromosome trisomies, if identified, will be reported with a specificity of 98 percent.

DISCLAIMER: Risk scores are calculated based on maternal age, gestational age, and test results. Findings of unknown significance will not be reported. Cases with evidence of fetal and/or placental mosaicism will not be reported. As this assay is a screening test and not diagnostic, false positive and false negatives can occur. Positive test results need diagnostic confirmation by alternative testing methods. Negative results do not fully exclude the diagnosis of any of the above syndromes. False positive and false negative results may be due to placental, fetal or maternal mosaicism, small imbalances, point mutations, gene inactivation, haploblocks, or other genetic/epigenetic mechanisms. Other potential sources of error include, but are not limited to, DNA sample contamination or degradation, limitations of current diagnostic techniques, misidentification of samples, or other factors that may interfere with correct interpretation of the analysis. This test has the potential to uncover consanguinity in the family. This test is not intended to identify pregnancies at risk for open neural tube defects. This test was developed and its performance characteristics determined by ARUP Laboratories. The U.S. Food and Drug Administration has not approved or cleared this test; however, FDA clearance or approval is not currently required for clinical use. The results are not intended to be used as the sole means for clinical diagnosis or patient management decisions.

Software version: Constellation 2.2

ARUP LABORATORIES | 500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787 | toll free: (800) 522-2787 | **www.aruplab.com**
JULIO C. DELGADO, MD, MS | Director of Laboratories

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