Non-Invasive Prenatal Testing for Fetal Aneuploidy with 22q11.2 Microdeletion

Patient: DOB: Age: 35 Gender: F **Patient Identifiers:** **Visit Number (FIN):**	Client: Physician:	ARUP Test Code: 2013142 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	Report Fetal Sex:	Yes
Estimated Due Date:	19 Jun 2018	Maternal Weight:	150 lbs
Gestational Age at Draw:	20 weeks 0 days	Maternal Height:	65 in
		Number of Fetus:	One

Results Summary

HIGH RISK 22q11.2 deletion syndrome

Fetal Fraction: **8.0%** Fetal Sex: **Male**

This pregnancy is classified as HIGH RISK by this screen for deletion/duplication at 22q11.2, which is associated with DiGeorge/Velocardiofacial syndrome (or 22q11.2 deletion syndrome). This result should be confirmed by a diagnostic test. On average, 20% of pregnancies classified as HIGH RISK are found to have 22q11.2 deletion syndrome when no ultrasound anomalies were seen, and 100% when ultrasound anomalies were seen prior to testing.

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, including SNP microarray, 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
Ph.D., FACMG
Electronic Signature

Conditions Screened

Trisomy 21:	Low risk	Monosomy X:	Low risk
Trisomy 18:	Low risk	Triploidy/Vanishing twin:	Low risk
Trisomy 13:	Low risk	22q11.2 deletion syndrome:	**HIGH RISK**

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JULIO C. DELGADO, MD, MS | Director of Laboratories

Chart continues on following page(s)

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

TEST INFORMATION: Non-Invasive Prenatal Testing for Fetal Aneuploidy with 22q11.2 Microdeletion (Powered by Constellation)

METHODOLOGY: DNA isolated from the maternal blood, which contains placental DNA, is amplified at >20,000 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. This screen also estimates the fetal copy number of the chromosomal region attributed to 22q11.2 deletion syndrome. Barring QC failures and fetal fractions below the performance limits of the algorithm, the minimum confidence threshold for aneuploidies is 0.98 for a High Risk call, and for the microdeletion the minimum confidence threshold is 0.95 for a High 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. Under specific circumstances, the algorithm may return a result of "unchanged" which is equivalent to the population frequency of the microdeletion.

SENSITIVITY AND SPECIFICITY: For combined autosomal aneuploidies, Turner's syndrome, and 22q11.2 microdeletion (DiGeorge 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: Risks for aneuploidy are calculated based on maternal age, gestational age and test results. Risks for microdeletions are based on population frequency and test results. This test will not identify all deletions associated with each disorder. Ability to detect deletions will be based on size and location. 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

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