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

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	4/3/1982
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

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

ARUP test code 2013142

Result Summary

HIGH RISK 22Q

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.

TEST INFORMATION: Non-Invasive Prenatal Testing for Fetal Aneuploidy (Powered by Constellation) with or without Microdeletions

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 and identify whole chromosome abnormalities for chromosomes 13, 18, 21, X, and Y 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. 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.

Microdeletions: An additional 6,600+ loci are amplified to estimate the fetal copy numbers of chromosomal regions attributed to 22q11.2, Prader-Willi, Angelman, Cri-du-chat, and 1p36 deletion syndromes. For any of the microdeletions, the minimum confidence threshold is 0.95 for a High Risk call. Under specific circumstances, the algorithm may return a result of "unchanged" which is equivalent to the population frequency of that microdeletion.

SENSITIVITY AND SPECIFICITY: For combined autosomal aneuploidies and Turner's syndrome, sensitivity and specificity are >99 percent. For combined microdeletions, sensitivity is >99 percent, specificity is >94 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 ABBREVIATIONS: T21 (trisomy 21); T18 (trisomy 18); T13 (trisomy 13); Trip (triploidy or vanishing twin); 45,X (monosomy X); XXX (trisomy X) ; XXY (Klinefelter syndrome); XYY (XYY syndrome); 22q (22q11.2 deletion syndrome); 1p36 (1p36 deletion syndrome); AS (Angelman syndrome); 5p- (Cri-du-chat syndrome); PWS (Prader-Willi syndrome)
Trisomy 21	Low risk
Trisomy 18	Low risk
Trisomy 13	Low risk
Monosomy X	Low risk
Triploidy/Vanishing Twin	Low risk
22q11.2 deletion syndrome	HIGH 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. 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.
Fetal Fraction	8.0

H=High, L=Low, *=Abnormal, C=Critical

Report Fetal Sex?	Yes
Fetal Sex	Male
Gestational Age at draw (Weeks)	20
Gestational Age at draw (Days)	0
Maternal Weight (Pounds)	150
Maternal Height (Inches)	65
Number of Fetuses	One
EER Fetal Aneuploidy w/22q11.2	See Note Access ARUP Enhanced Report using either link below: -Direct access: -Enter Username, Password: https://erpt.aruplab.com Username: Password: This result has been reviewed and approved by Reha Toydemir, MD, Ph.D., FACMG Electronic Signature

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Result Summary	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Trisomy 21	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Trisomy 18	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Trisomy 13	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Monosomy X	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Triploidy/Vanishing Twin	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
22q11.2 deletion syndrome	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Fetal Fraction	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Report Fetal Sex?	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 9 05:00 AM
Fetal Sex	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM
Gestational Age at draw (Weeks)	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 9 05:00 AM
Gestational Age at draw (Days)	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 9 05:00 AM
Maternal Weight (Pounds)	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 9 05:00 AM
Maternal Height (Inches)	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 9 05:00 AM
Number of Fetuses	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 9 05:00 AM
EER Fetal Aneuploidy w/22q11.2	18-030-101899	1/30/2018 8:14:00 AM	1/30/2018 8:19:47 AM	1/30/2018 10:41:00 AM

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical