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

Physician: Doctor, Example

Patient: Patient, Example

DOB	4/3/1984
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

Low Risk

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

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. Based on maternal age, gestational age, and the analysis of the circulating cell-free DNA, the risk that this fetus has any of the conditions listed in this report is LOW. If clinical findings conflict with these test results, appropriate clinical follow-up should be undertaken. This may include genetic counseling, ultrasound, amniocentesis, CVS or other testing as recommended by the patient's healthcare provider.

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

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

	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	Low risk

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Fetal Fraction	9.0
Report Fetal Sex?	Yes
Fetal Sex	Female
Gestational Age at draw (Weeks)	12
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 Elaine Lyon, Ph.D. Electronic Signature

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Result Summary	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Trisomy 21	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Trisomy 18	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Trisomy 13	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Monosomy X	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Triploidy/Vanishing Twin	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
22q11.2 deletion syndrome	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Fetal Fraction	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Report Fetal Sex?	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8 22:00 AM
Fetal Sex	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM
Gestational Age at draw (Weeks)	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8 22:00 AM
Gestational Age at draw (Days)	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8 22:00 AM
Maternal Weight (Pounds)	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8 22:00 AM
Maternal Height (Inches)	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8 22:00 AM
Number of Fetuses	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8 22:00 AM
EER Fetal Aneuploidy w/22q11.2	18-080-101700	3/21/2018 8:09:00 AM	3/21/2018 8:16:00 AM	3/21/2018 8:28:00 AM

END OF CHART

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