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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

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

ARUP test code 3003043

Multiple Gestation	No
Gestational Age at Draw	10 wks or over
Report Fetal Sex	Yes
Fetal Fraction	0.6 %
EER Non-Invasive Prenatal NGS Aneu	See Note Authorized individuals can access the ARUP Enhanced Report with an ARUP Connect account using the following link. Your local lab can assist you in obtaining the patient report if you don't have a Connect account. [redacted]
Trisomy 21	Low Risk
Trisomy 18	Low Risk
Trisomy 13	Low Risk
Sex Chromosome Trisomies and Monosomy X	Low Risk
Fetus Sex	Male

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

Result Summary

Low Risk

The reported fetal fraction for this sample is less than 4.0 percent. Samples with observed fetal fraction less than 4.0 percent are associated with lower sensitivity to detect fetal aneuploidy (PMID: 32804883, PMID: 27467454). Clinical correlation is suggested.

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 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 performed. This may include genetic counseling, ultrasound, amniocentesis, CVS or other testing as recommended by the patient's healthcare provider.

A published meta-analysis determined negative likelihood ratios of 0.003 for trisomy 21, 0.022 for trisomy 18, and 0.010 for trisomy 13 (PMID: 28397325). Positive predictive values (PPV) and negative predictive values (NPV) are affected by an individual's pre-test risks for each of the screened conditions. Online calculators are available to calculate patient-specific PPV and NPV using age and clinical information.

Health care providers with questions may contact an ARUP genetic counselor at (800) 242-2787 ext. 2141.

This result has been reviewed and approved by [redacted]

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 (PPA) compared to established methods to detect fetal aneuploidy. For samples with greater than or equal to 4.0 percent fetal fraction, the PPAs are as follows: T13 is 86.7 percent, T18 greater than 99.9 percent, and T21 is 96.4 percent. The combined PPA for all aneuploidies is 95.5 percent. For samples with less than 4.0 percent fetal fraction, the sensitivity to detect fetal aneuploidy is significantly lower and the combined PPA is 60 percent. The specificity, as calculated as negative percent agreement, is 99.5 percent 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. Prior to any pregnancy management decisions, confirmation of high-risk results by diagnostic testing (amniocentesis, CVS, or postnatal testing) is recommended. 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 Turner Syndrome, XXX, XXY, and XYY. This assay does not assess 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, fetal demise, vanishing twin, fetal partial aneuploidy, and/or mosaic aneuploidy of the fetus, mother, and/or placenta. Samples with observed fetal fraction less than 4.0 percent have lower sensitivity to detect fetal aneuploidy, and the accuracy of the fetal fraction estimate is significantly lower. Fetal demise/miscarriage is not assessed.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Multiple Gestation	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Gestational Age at Draw	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Report Fetal Sex	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fetal Fraction	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Non-Invasive Prenatal NGS Aneu	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Trisomy 21	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Trisomy 18	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Trisomy 13	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Sex Chromosome Trisomies and Monosomy X	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fetus Sex	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Result Summary	25-009-102658	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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