

Patient: [REDACTED]  
 DOB: [REDACTED] Age: [REDACTED] Sex: [REDACTED]  
 Patient Identifiers: [REDACTED]  
 Visit Number (FIN): [REDACTED]

Client: [REDACTED]  
 Physician: [REDACTED]

ARUP Test Code: 3003043  
 Collection Date: 06/20/2024  
 Received in lab: 06/20/2024  
 Completion Date: 07/16/2024

## Result Summary

### See Note

Fetus Sex	No Result
Fetal Fraction	10.1%
Trisomy 21 (Down Syndrome)	Low Risk
Trisomy 18 (Edwards Syndrome)	High Risk
Trisomy 13 (Patau Syndrome)	Low Risk
Monosomy X (Turner Syndrome)	No Result
Sex Chromosome Trisomies	No Result

This pregnancy is classified as HIGH RISK for trisomy 18 (Edwards syndrome) by this screen. This result should be confirmed by a diagnostic test. The chance that a pregnancy classified as high risk will have the screened condition (the positive predictive value, or PPV) is affected by the pre-test risk for the screened condition. For women with no additional risk factors, 23-33% of pregnancies classified as high risk by NIPT are found to have trisomy 18. For women with a high pre-test risk, 67-84% of pregnancies classified as high risk by NIPT are found to have trisomy 18. Online calculators are available to determine patient-specific PPV based on clinical context.

Testing of this specimen did not yield result(s) on the sex chromosome(s); neither the sex of the fetus nor the risk for sex chromosome abnormalities can be assessed. A repeat specimen will not provide additional information and is not indicated. Underlying causes may include, but are not limited to, maternal medical conditions and/or aneuploidy/mosaic aneuploidy, fetal chromosome abnormalities/mosaic chromosome abnormalities other than the targeted conditions.

This is a screening test and is NOT diagnostic for the condition(s) listed in this report. Both false positive and false negative results may occur. Appropriate clinical follow-up such as genetic counseling, comprehensive ultrasound, and confirmatory diagnostic testing should be undertaken as recommended by the patient's healthcare provider. Irrevocable action such as pregnancy termination should not be taken based on the results of this screening test.

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

### Test Specific Patient Information

Report Sex:	Yes
Gestational Age at Draw:	10 wks or over
Number of fetuses:	Singleton



Patient: [REDACTED] ARUP Accession: [REDACTED]

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

Patient: [REDACTED] | Date of Birth: [REDACTED] | Sex: [REDACTED] | Physician: [REDACTED]  
 Patient Identifiers: [REDACTED] | Visit Number (FIN): [REDACTED]

Trisomy 18*						
Maternal age at EDD	20 years	25 years	30 years	35 years	40 years	44 years
Gestational Age	Positive Predictive Value (PPV) %					
10 weeks	23.9	26.2	34.9	57.4	83.3	94.1
12 weeks	20.1	22.1	30.1	51.9	80.0	92.7
14 weeks	17.2	19.0	26.2	47.1	76.7	91.4
16 weeks	14.8	16.4	22.9	42.8	73.4	89.8
20 weeks	11.3	12.6	17.9	35.4	66.9	86.6
30 weeks	6.2	6.9	10.1	22.0	51.0	76.8

Prior risks in this table are based on gestational age and maternal age at estimated due date (EDD). Additional clinical information affecting prior risk – such as ultrasound abnormalities, family history, and prior screening results – is not included. Clinical decisions should take all relevant medical and family history into account.

PPV is the chance that a fetus with a high risk screening result has the condition. The PPV calculation is based on the sensitivity and specificity of testing along with the prevalence of the condition.

\*PPV values presented are calculated based on patient pretest risk, reported VeriSeq test performance,<sup>1</sup> and the incidence of aneuploidy at specific maternal/gestational age as previously reported.<sup>2,3</sup>

<sup>1</sup>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.

<sup>2</sup>Snijders RJ. Maternal age and gestational age-specific risk for chromosomal defects. *Fetal Diagn Ther.* 1995;10(6):356-67.

<sup>3</sup>Snijders RJ. Maternal age- and gestation-specific risk for trisomy 21. *Ultrasound Obstet Gynecol.* 1999;13(3):167-70.

## Background Information:

**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 compared to established methods to detect fetal aneuploidy. For samples with greater than 5 percent observed fetal fraction, the positive percent agreements (PPA) are as follows: T13 greater than 99.9 percent, T18 greater than 99.9 percent, and T21 is 96.1 percent. The combined PPA for all aneuploidies is 97.5 percent. For



Patient: [REDACTED] ARUP Accession [REDACTED]

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

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Patient Identifiers: [REDACTED] | Visit Number (FIN): [REDACTED]

samples with less than or equal to 5 percent observed fetal fraction, the positive percent agreements (PPA) are as follows: T13 is 66.7 percent, T18 is 60 percent, and T21 is 87.5 percent. The combined PPA for all aneuploidies is 72.3 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. High risk results must be confirmed by diagnostic testing (amniocentesis, CVS, or postnatal testing) before any clinical decisions are made based on the screening test result. 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 XO, XXX, XXY, and XYY. This assay is not meant to detect 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, maternal mosaicism, placental mosaicism, fetal demise, disappearing twin, fetal partial aneuploidy, and/or fetal mosaicism. Samples with observed fetal fraction less than 5.0 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 US 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.

