ARUP LABORATORIES

*A nonprofit enterprise of the University of Utah and its Department of Pathology*

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# NONINVASIVE PRENATAL ANEUPLOIDY SCREEN BY CELL-FREE DNA SEQUENCING

# CONSENT FORM

**Patient Name:** _____ **Date of Birth:** _____

**Sex Assigned at Birth:** ☐ Female ☐ Male ☐ Intersex **Gender Identity (optional):** ☐ Female ☐ Male ☐ _____

Prenatal cell-free DNA (cfDNA) screening (also referred to as noninvasive prenatal screening [NIPS]) is a screening test that identifies pregnancies at increased risk for specific chromosome aneuploidy: trisomy 21 (Down syndrome), trisomy 18, and trisomy 13. These disorders cause a range of physical birth defects and intellectual disability. Prenatal cfDNA screening may also suggest an increased risk for an extra or missing sex chromosome. High-risk prenatal screening using cfDNA technologies should be confirmed by diagnostic tests; irreversible clinical decisions should never be based solely on a screening test result.

The following has been explained to me:

1. Prenatal cfDNA screening is a screening test, not a diagnostic test. False-positive and false-negative results may occur.
2. Participation in genetic testing is completely voluntary. Genetic counseling is available. See nsgc.org or acmg.net to find a medical genetic professional.
3. ARUP prohibits the use of these results to facilitate any form of discrimination or violation of ethical or legal guidelines outlined by national and international standards.
4. Patients with a high-risk result, or no result, by prenatal cfDNA screening should be referred for genetic counseling, comprehensive ultrasound, and offered diagnostic testing through chorionic villus sampling [CVS] or amniocentesis.
5. There are three possible test results:
    a. High risk: indicates screening has detected a significantly increased chance for the fetus to have an abnormal number of one of the following chromosomes: 13, 18, 21, X, or Y.
    b. Low risk: indicates there is less than 1 in 100 chances for one of the screened conditions. However, healthcare providers may still recommend a fetal karyotype or other testing. If clinical results contradict test results, diagnostic testing should be considered.
    c. No result: indicates the lab is unable to interpret the results of the screen. Prenatal cfDNA screening may be indeterminate due to biological or technical limitations. There may be too little fetal DNA present in the sample (low fetal fraction); mosaicism for a chromosome abnormality in the fetus, placenta, or mother; and other maternal and fetal factors. Note that no result due to low fetal fraction is more common at early gestational ages and with high maternal BMI.
6. This test can identify fetal sex. Fetal sex will be reported unless “No” is marked on the patient history form. If the fetus is at increased risk for Turner syndrome, XXX, XXY, or XYY, this result will be reported, even if opting out of fetal sex reporting was chosen. In rare instances, incorrect sex results are reported.
7. This screening test is limited to trisomy 13, trisomy 18, trisomy 21, and sex chromosome aneuploidies. This screening test does not assess triploidy, microdeletions, other abnormalities of the tested chromosomes, or abnormalities involving nontested chromosomes. This test does not screen other genetic disorders or birth defects.
8. The cfDNA analyzed is both fetal and maternal. Prenatal cfDNA screening may occasionally indicate that a chromosomal abnormality, or malignancy, is present in the maternal DNA portion of the prenatal sample. ARUP’s prenatal cfDNA screening test was not designed or validated to report such findings.
9. Prenatal cfDNA screening cannot be interpreted accurately in pregnancies with a fetal demise/nonviable twin. Prenatal cfDNA screening in pregnancies with an unrecognized/unreported twin demise are more likely to have a false-positive result.
10. ARUP only performs testing on singleton pregnancies confirmed by the client upon order. Two or more (multiple) gestation samples will be sent out to LabCorp to perform the MaterniT21 PLUS Core test (test code 451927). Testing order will be canceled if the client fails to provide the number of fetuses.
11. Prenatal cfDNA testing cannot be interpreted accurately in

pregnancies less than 10 weeks gestation. Patients must confirm that gestational age is 10 weeks or greater. Testing will be canceled for patients with a gestational age <10 weeks or for patients for which this information is not provided.

12. Although genetic test results are usually accurate, several sources of error are possible including twin pregnancies disclosed as singleton pregnancies or errors which occurred during the specimen handling processes.

**Patient, Legal Guardian, Power of Attorney (POA):** I authorize ARUP Laboratories to perform prenatal cfDNA screening. The benefits, risks, and limitations of this testing have been explained to my satisfaction by a qualified health professional.

| Patient/Guardian/POA Printed Name | Signature | Date |
|---|---|---|

**Ordering Healthcare Provider, Genetic Counselor:** I have explained this genetic test and its risks, benefits, and alternatives to the patient or legal guardian and addressed all of their questions.

| Provider/Genetic Counselor Printed Name | Signature | Date |
|---|---|---|

| Specialty | Phone Number | Fax |
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