ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

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INFORMED CONSENT FOR NON-INVASIVE PRENATAL TESTING (NIPT)

Patient Name _____ Date of Birth _____ Sex ☐ F ☐ M

NIPT is a screening test which can be performed on women at or after 9 weeks 0 days gestation, primarily to identify fetuses at risk to have extra or missing copies of chromosomes 13, 18, 21, X or Y. **This test is not intended to diagnose these conditions, and additional tests are recommended to confirm any positive NIPT results.**

Although the risk for having a pregnancy with an extra chromosome increases as women get older, every pregnancy has a small risk. NIPT identifies pregnancies at increased risk for common chromosome disorders such as trisomy 21 (Down syndrome), trisomy 18, trisomy 13 and triploidy. These disorders can cause a range of physical birth defects and intellectual disability, with trisomy 21 on the milder end of the spectrum, and trisomies 18 and 13, and triploidy on the severe end with most affected babies not surviving to birth. NIPT may also suggest an increased risk for an extra or missing sex chromosome which may be associated with learning disabilities, fertility issues and birth defects. Lastly, depending on the test selected, NIPT may also suggest an increased risk for the following rare microdeletion syndromes: deletion 1p36, cri-du-chat, Angelman or Prader-Willi, and DiGeorge. These are typically associated with intellectual disability and physical birth defects. Please note that under very few circumstances is a woman at increased risk to have a baby with a microdeletion. Therefore, all women should be considered *low risk* to have a child with a microdeletion, and pretest genetic counseling should be considered to help women fully understand the benefits and limitations of microdeletion screening.

The table below indicates the conditions detected by the various non-invasive prenatal tests offered by ARUP Laboratories.

Test / Disorder	Non-Invasive Prenatal Testing for Fetal Aneuploidy	Non-Invasive Prenatal Testing for Fetal Aneuploidy with 22q11.2 Microdeletion	Non-Invasive Prenatal Testing for Fetal Aneuploidy with Microdeletions
Trisomy 21	✓	✓	✓
Trisomy 18	✓	✓	✓
Trisomy 13	✓	✓	✓
Monosomy X	✓	✓	✓
Sex chromosome trisomies	✓	✓	✓
Triploidy	✓	✓	✓
22q11.2 deletion		✓	✓
1p36 deletion			✓
Angelman syndrome			✓
Prader-Willi syndrome			✓
5p deletion			✓
Twin, Egg donor, or surrogate*	✓	✓†	

*Testing ordered on twins, egg donor, or surrogate pregnancies will be performed at Natera.
†Available for monozygotic twins only

The following has been explained to me:

1. NIPT is a highly accurate screening test, but is not intended to replace diagnostic testing by CVS or amniocentesis. These tests are available to me.
2. Participation in genetic testing is completely voluntary. Genetic counseling is available if you have questions regarding testing. See www.nsgc.org or www.acmg.net to find a medical genetics professional.
3. There are four possible test results:
   a) A "high risk" result indicates that the screen 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, or a deletion at one of the specified genomic locations. The positive predictive value (chance the fetus is affected) for the specific disorder will be included in the report. Patients with a high-risk NIPT result should be referred for genetic counseling and offered diagnostic testing.
   b) A "low risk" result means there is less than 1 in 100 chance for one of the screened conditions. However, your healthcare provider may still recommend a fetal karyotype or other testing if your fetus is found to have ultrasound anomalies or there are other concerns about your fetus' health.
   c) A "no result" occurs when the lab is unable to interpret the results of the screen. This may be due to too little fetal DNA (low fetal- fraction); a low level of total cell-free DNA present in the maternal sample; mosaicism in the fetus, placenta, or mother; if the patient is not the genetic mother of the fetus; or may also occur if the mother and the father of the fetus are related by blood (e.g., cousins). Under some circumstances, the laboratory may request a second sample (at no charge) to clarify the test results.

   d) A result of "unchanged" is possible for microdeletions only. This indicates that the screen was unable to determine if your risk to have a child with the deletion was either increased or decreased. The population risk will be reported in these cases. A repeat screen is not indicated.

4. This test has the ability to identify fetal sex.
   a) Fetal sex *will* be reported unless the "No" box is checked on the patient history form.
   b) If the fetus is at high risk to have Turner syndrome, XXX, XXY, or XYY, that result will be reported to me, even if I have elected not to have fetal sex disclosed.
5. NIPT may:
   a) indicate that my fetus is at increased risk to have one or more specific chromosome abnormalities (Down syndrome, trisomy 18, trisomy 13, Turner syndrome, triploidy, or a sex-chromosome trisomy);
   b) be indeterminate due to biological or technical limitations;
   c) suggest a biological relationship between the mother and father of the fetus;
   d) identify a chromosomal abnormality in the mother of the fetus.
6. Limitations of NIPT include:
   a) This is a screening test, not a diagnostic test. False positive and false negative results may occur. Positive results should be confirmed by direct fetal testing.
   b) Testing is limited to the chromosomes and conditions listed above. This test will not identify other abnormalities of the tested chromosomes such as deletions or duplications, and does not detect other genetic disorders or birth defects.
   c) Results may not be interpretable if there is too little fetal DNA present in the sample (low fetal fraction). In these cases, a repeat test at no extra laboratory charge may be offered.
      - High maternal BMI is a common reason for low fetal fraction. In the case of maternal obesity, performing testing after 14 weeks gestation, and waiting a minimum of two weeks before having a repeat sample drawn, may increase the likelihood of obtaining results.
   d) Mosaicism for the targeted chromosomes may not be detected.
   e) Aneuploidy screening can be performed using this method in twin gestations, or if the patient whose blood is being tested is not the genetic mother of the fetus (i.e., if the fetus was conceived using another woman's egg). However, testing cannot be performed if more than one of these conditions is true (i.e., cannot be performed if the patient used an egg donor AND is carrying twins).
   f) Triploidy cannot be distinguished from a vanishing or existing twin gestation. Ultrasound and/or direct fetal testing may be necessary to distinguish between these two possibilities. Triploidy will not be reported in stated twin or egg-donor pregnancies.
7. A "high risk" result greatly increases the chances that the fetus has an extra copy of any of the tested chromosomes, or has a deletion of one of the targeted microdeletion sites, but false positive test results do occur. Thus, **positive results should be confirmed by direct fetal testing.**
8. A "low risk" result greatly reduces the chances that the fetus has an extra copy of any of the tested chromosomes, or has a deletion of one of the targeted microdeletion sites, but false negative test results can occur. If clinical results contradict test results, then diagnostic fetal testing (CVS or amniocentesis) should be considered.
9. Although genetic test results are usually accurate, several sources of error are possible including, but not limited to: sample mishandling, misidentification, and contamination.
10. Residual DNA samples may be stored indefinitely to be used for test validation or education after personal identifiers are removed. Samples from New York clients, however, will be disposed of 60 days after testing is complete. No clinical tests other than those authorized will be performed. I may request disposal of my blood and DNA sample following completion of the test requested above by contacting the laboratory at (800) 242-2787, ext. 3301. Refusal to permit the use of my sample for test validation or education will not affect my test result. For more information about ARUP, please refer to www.aruplab.com.

Patient/Legal Guardian: My signature below constitutes my desire to undergo NIPT testing and my acknowledgment that the benefits, risks, and limitations of NIPT have been explained to my satisfaction by a qualified health professional.

Patient/Guardian Printed Name _____ Signature _____ Date _____

Ordering Healthcare Provider: I have explained NIPT, its limitations and alternatives to the patient or legal guardian and answered all of their questions.

Physician/Genetic Counselor Printed Name _____ Signature _____ Date _____

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