ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

INFORMED CONSENT FOR NON-INVASIVE PRENATAL TESTING (NIPT PANORAMA®)

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

NIPT is a screening test that can be performed on women at or after 9 weeks 0 days gestation, and is primarily used to identify fetuses at risk for duplicate or deleted 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; trisomy 21 is on the milder end of the spectrum, while trisomy 18, trisomy 13, and triploidy are on the severe end of the spectrum, 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 Natera.

Disorder \ Test	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			✓

*Not available for egg donor/surrogate or dizygotic twin gestations (monozygotic twins are acceptable for testing)
†Not available for egg donor/surrogate or twin gestations

The following has been explained to me:

1. NIPT is a highly accurate screening test, but is not intended to replace diagnostic testing by chorionic villus sampling (CVS) or amniocentesis, both of which are available to me.
2. Participation in genetic testing is completely voluntary. Genetic counseling is available if you have questions about testing. See www.nsgc.org or www.acmg.net to find a medical genetic professional.
3. There are four 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, 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) Low risk: indicates 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 if there are other concerns about your fetus' health.
   c) No result: indicates the lab is unable to interpret the results of the screen. This may be due to not enough 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; 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) Unchanged: result 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.

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4. This test has the ability to identify fetal sex.
    a) Fetal sex *will* be reported unless I check "No" on the patient history form.
    b) If the fetus is at high risk for 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, abnormalities involving non-tested chromosomes, 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 results do occur. **Positive results should be confirmed by direct fetal testing (CVS or amniocentesis).**
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 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 above requested test by contacting the laboratory at Natera's customer service department (650) 249-9090. Refusal to permit the use of my sample for test validation or education will not affect my test result. For more information about Panorama NIPT, please refer to www.natera.com/panorama-test.

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.

Healthcare Provider Printed Name	Signature	Date

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