ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

NONINVASIVE PRENATAL TESTING (NIPT) PATIENT HISTORY FORM

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

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

Ordering Provider: _____ **NPI #:** _____

Physician's Pager/Cell: _____ **Physician's Phone:** _____

Practice Specialty: _____ **Physician's Fax:** _____

Genetic Counselor: _____ **Counselor's Phone:** _____

Draw date: _____ Gestational age at draw: _____weeks _____days

Patient's current weight: _____ ☐ lb / ☐ kg Patient's height: _____ ☐ in / ☐ cm

Fetal sex by ultrasound: ..... ☐ Male ☐ Female ☐ Ambiguous ☐ Unknown

For twin or surrogate/egg donor pregnancies*, check all that apply

We do NOT accept vanished twin or higher order multiple gestation pregnancies, or twins conceived using a surrogate or egg donor.

☐ Ongoing **twin pregnancy:** ..... ☐ monozygotic ☐ dizygotic

☐ **IVF**-conceived pregnancy: Age of genetic parent at egg retrieval _____

☐ **Surrogate** or **egg donor** pregnancy

**Twin/egg donor samples will be forwarded to and run at Natera, but still reported by ARUP.*

Indication for testing (check all that apply)

☐ Advanced maternal age: ☐ Primigravida/1st trimester (O09.511) ☐ Primigravida/2nd trimester (O09.512)
☐ Multigravida/1st trimester (O09.521) ☐ Multigravida/2nd trimester (O09.522)

☐ Ultrasound abnormality (O28.3) (describe): _____

☐ Abnormal antenatal screening (birthing parent):

☐ Biochemical (O28.1)

☐ Chromosomal (O28.5)

☐ Serum screen positive for:
☐ T21 ☐ T18 ☐ T13
Risk based on MSS was 1 in _____

☐ Other (O28.8) (describe): _____

☐ Encounter for other screening for genetic and chromosomal anomalies (Z13.79)

☐ Family history (Z82.79) (describe): _____

☐ Personal history:

☐ Balanced translocation/inversion in normal individual (Q95.0): (Complete below)

☐ Translocation/inversion present in: ..... ☐ the patient ☐ in the FOB ☐ in a previous child/fetus

☐ Translocation/inversion involving chromosome(s): _____

☐ Other (describe): _____

I want to know the sex of the fetus (sex <u>will</u> be reported if nothing is checked) ..... ☐ Yes ☐ No

Check the test you intend to order.

☐ **3004764 Fetal Aneuploidy Screening**: Test for fetal aneuploidy involving chromosomes 13, 18, 21, X, and Y.

☐ **3004778 Fetal Aneuploidy Screening with 22q11.2 Microdeletion**: Test for fetal aneuploidy involving chromosomes 13, 18, 21, X, and Y, as well as for deletions causing DiGeorge/velocardiofacial syndrome. *NOT AVAILABLE for twin or egg donor/surrogate pregnancies (exception: monozygotic twin gestations)*

☐ **3004781 Fetal Aneuploidy Screening with Microdeletions**: Test for fetal aneuploidy involving chromosomes 13, 18, 21, X, and Y, as well as for deletions causing DiGeorge/velocardiofacial, 1p36, Angelman, cri-du-chat, and Prader-Willi syndromes.

NOT AVAILABLE for twin or egg donor/surrogate pregnancies

TPB Institutions Only: Front and back copies of insurance card required with specimen submission.

Master Label

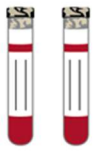
For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141

NONINVASIVE PRENATAL TESTING (NIPT) COLLECTION INSTRUCTIONS

Purpose of Proper Collection

The NIPT screen measures fetal DNA in maternal blood plasma. Each step below is important in ensuring that the maternal blood cells do not lyse and release extra maternal DNA in the plasma. If this happens, the fetal DNA fraction in the plasma becomes too small and is not able to be analyzed. In these cases, a redraw sample may be requested.

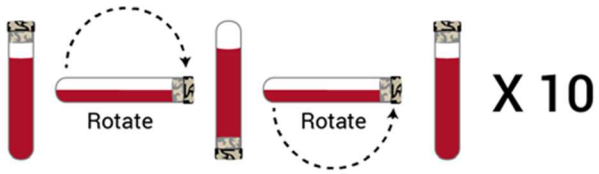
1 COLLECT MOTHER'S BLOOD



10 mL of blood in each of two Streck Cell-Free DNA BCT tubes
20–21 gauge straight needles
No butterflies

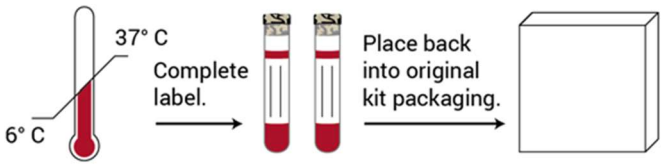
Blood cell breakage is minimized when the correct needle is used and extra tubing (i.e., "butterfly needles") is avoided. The proper technique is the shortest distance from the vein to the collection tube with a straight 20–21 gauge needle.

2 GENTLY MIX SAMPLE—DO NOT SHAKE



A preservative in the tube protects the cells during transport. This preservative must be gently mixed with the entire blood volume by completely inverting the tube 10 times. If the sample is not completely and thoroughly mixed, a fraction of the cells may lyse and release additional maternal DNA into the plasma, requiring a second sample.

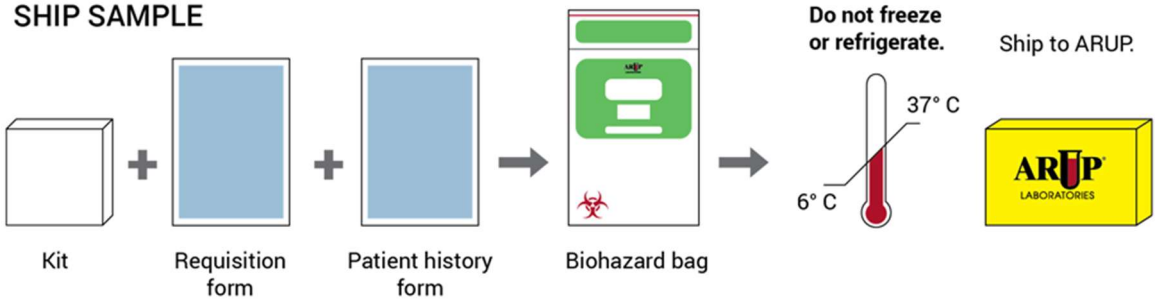
3 PRE-PACK SAMPLE



High or low temperatures may cause the cells to lyse and release additional maternal DNA into the plasma, thus diluting the fetal component and requiring a second sample.

Tubes are made of glass. To avoid breakage during transport, please place them back into original kit packaging.

4 SHIP SAMPLE



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NONINVASIVE PRENATAL TESTING (NIPT PANORAMA) INFORMED CONSENT FORM

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

NIPT is a screening test that can be performed on individuals 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 with age, 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 cognitive 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 fetuses 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 cognitive disability and physical birth defects. Please note that under very few circumstances is an individual at increased risk to have a baby with a microdeletion.

Therefore, all pregnant individuals should be considered *low risk* to have a child with a microdeletion, and pretest genetic counseling should be considered to help pregnant individuals fully understand the benefits and limitations of microdeletion screening.

The table below indicates the conditions detected by the various noninvasive 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:

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.
2. Participation in genetic testing is completely voluntary. Genetic counseling is available if you have questions about testing. See nsgc.org or acmg.net to find a medical genetic professional.
3. There are four possible test results:
   a) High risk: This indicates that 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: This indicates that 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: This indicates that 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 genetic parent; or may also occur if the genetic parents 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: This result 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.

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NON-INVASIVE PRENATAL TESTING (NIPT) INFORMED CONSENT FORM

4. This test has the ability to identify fetal sex.
   a) Fetal sex *will* be reported unless you 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 you, even if you have elected not to have the fetal sex disclosed.
5. NIPT may:
   a) Indicate that the 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 genetic parents of the fetus
   d) Identify a chromosomal abnormality in the birthing parent 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 nontested 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 parent of the fetus (i.e., if the fetus was conceived using another individual'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. You may request disposal of your 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 your sample for test validation or education will not affect your test result. For more information about Panorama NIPT, please refer to 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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