mercy diagnostics

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PATIENT HISTORY FORM FOR NON-INVASIVE PRENATAL TESTING (NIPT)

Patient Name_____Date of Birth_____

Physician/Genetic Counselor_____ Phone_____

FAX _____ Pager/Cell: _____

Draw Date: _____ Gestational Age at Draw: _____weeks _____ days
Fetal gender by ultrasound: [ ] Male [ ] Female [ ] Ambiguous [ ] Unknown
Patient's current weight _____ lbs (or) _____ kgs
Patient's race: [ ] Caucasian [ ] Black [ ] Hispanic [ ] Asian [ ] Other

Is the patient carrying more than one fetus, or is there a known twin demise? [ ] Yes* [ ] No [ ] Unknown
Is the patient the genetic mother of the fetus (i.e. was the fetus conceived using the patient's own egg)? [ ] Yes [ ] No*

***This test is NOT appropriate when there is more than one fetus/twin demise or when the patient is not the genetic mother of the fetus. If you have questions, please contact genetics at 800-242-2787 x2141 before drawing the patient.**

Indication for testing (check all that apply):

[ ] Advanced Maternal Age: 1st pregnancy (659.53) [ ] Advanced Maternal Age: not 1st pregnancy (659.63)
[ ] Abnormal Maternal Serum Screen positive for (796.5): [ ] T21 [ ] T18 [ ] T13 Risk: _____
[ ] Ultrasound Abnormality (please describe): _____
[ ] Family History (v19.8) Describe:_____
[ ] Personal History (v13.89) Describe:_____
[ ] Antenatal Screening (v28.89)
[ ] Other: (please describe)_____

A cheek swab sample from the father of the fetus will accompany the maternal blood sample: ☐ Yes ☐ No

If yes, father's Name _____Date of Birth: _____

I want to know the sex of the fetus (sex will be reported if nothing is checked) **[ ] Yes [ ] No**

Please Circle the test you intend to order

2007537 Non-Invasive Prenatal Testing for Fetal Aneuploidy (Panorama) Screening test for fetal aneuploidy involving chromosomes 13, 18, 21, X and Y

2010232 Non-Invasive Prenatal Testing for Fetal Aneuploidy with Microdeletions (Panorama XP) Screening 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

INFORMED CONSENT FOR NATERA'S PANORAMA AND PANORAMA XP TESTS

Non-Invasive Prenatal Testing (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.**

NIPT by the **Panorama test** will identify most fetuses with the following chromosomal conditions:

- Trisomy 21 (T21) – is commonly known as Down syndrome and is caused by an extra copy of chromosome 21
- Trisomy 18 (T18) - is caused by an extra copy of chromosome 18 and is sometimes referred to as "Edwards syndrome"
- Trisomy 13 (T13) - is occasionally called "Patau syndrome" and is caused by an extra copy of chromosome 13
- Turner syndrome (45,X) – is also known as monosomy X and is usually caused by a missing sex chromosome (either X or Y)
- Triploidy (69,XXX/69,XXY/69,XYY) - is caused by an extra copy of each chromosome

NIPT by the **Panorama Extended Panel (XP)** will detect most fetuses with the above conditions as well as most fetuses with:

- 22q11.2 deletion syndrome -- caused by the loss of a small piece of chromosome 22 that results in overlapping phenotypes known as DiGeorge and Velocardiofacial (VCFS) syndromes
- 1p36 deletion syndrome -- caused by loss of a small piece of the short arm of chromosome 1
- Angelman syndrome -- caused by a deletion of a small piece of the chromosome 15 inherited from the mother of the fetus
- Prader Willi syndrome -- caused by a deletion of a small piece of the chromosome 15 inherited from the father of the fetus
- 5p deletion syndrome (*cri-du-chat*) -- caused by the loss of a small piece of the short arm of chromosome 5

Other conditions which may be detected and reported with both tests include sex-chromosome aneuploidies such as 47,XXY (Klinefelter syndrome), 47,XYY and 47,XXX.
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.

There are four possible test results:

1. A “High Risk’ result indicates that the test has detected a significantly increased risk 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 specific risk will be listed on the report. Patients with a high risk NIPT result should be referred for genetic counseling and offered diagnostic testing.
2. A “Low Risk’ result means the test detected a very low chance (less than 1 in 100) for the fetus to have an abnormal number of any of the above chromosomes or a deletion at one of the specified genomic locations. The specific risk will be listed on the report. 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.
3. A “No Call” result occurs when the lab is unable to interpret the results of the test. This may happen if there is too little fetal DNA present in the maternal sample; there is mosaicism in the fetus, placenta or mother; the patient is not the genetic mother of the fetus; or 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.
4. A result of “Unchanged” is possible for microdeletions only. This indicates that the test 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.

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. This test has the ability to identify fetal gender.
   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, that result will be reported to me, even if I have elected not to have fetal sex disclosed.
3. A cheek swab sample from the father of the baby is optional, but may reduce the likelihood that the laboratory will need a second sample for a repeat test. It is highly recommended to include a sample from the father if a second sample is requested due to a low fetal fraction in the first sample.
4. 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, or triploidy)
   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
5. Limitations of NIPT include:
   a) **This is a screening test, not a diagnostic test. 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 and does not detect abnormalities of chromosomes other than those tested.
   c) Balanced chromosomal rearrangements such as translocations or inversions, other genetic disorders, birth defects, and other fetal or pregnancy complications will not be detected.
   d) 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 will be offered.
   e) Mosaic (the presence of both normal and abnormal cells) aneuploidy for the targeted chromosomes may not be detected.
   f) Testing cannot be performed using this method 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), if the patient has received an allogeneic bone marrow transplant, or if there is more than one fetus present.
   g) 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.
6. A ‘low risk’ result greatly reduces the chances that the fetus has an extra or missing copy of one of the tested chromosomes, or has a deletion of one of the targeted microdeletion sites, but false negative test results can occur.
7. Several sources of error are possible including, but not limited to: sample mishandling, sample misidentification, and sample contamination.
8. My DNA sample may be stored indefinitely to be used for test validation or education after personal identifiers are removed. Samples from New York clients will not be stored and 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.

This testing is performed by Natera. The performance characteristics of this test were validated by Natera Laboratories, Inc. The U.S. Food and Drug Administration (FDA) has not approved this test; however, FDA approval is currently not required for clinical use of this test. Natera is authorized under Clinical Laboratory Improvement Amendments (CLIA) and by all states to perform high-complexity testing. These results are not intended to be used as the sole means for clinical diagnosis or patient management decisions

NIPT is a fee-for-service test. I will be responsible for payment after the testing has begun, even if I decide not to receive results.
ARUP will provide a local referral for genetic counseling at my request.

PATIENT CONSENT STATEMENT

I have read or have had read to me the above informed consent information about the Panorama Non-Invasive Prenatal Test (NIPT). I have had the opportunity to ask questions of my health care provider regarding this test, the risks, and the alternatives prior to my informed consent. I request and authorize Natera to test my sample(s) for the fetal chromosome conditions listed above.

Patient/Guardian Signature _____ Date _____

PHYSICIAN/GENETIC COUNSELOR

I have explained NIPT and its limitations to the patient or legal guardian and answered all questions.

Printed Name of Provider: _____ Date _____

Signature_____ Phone Number _____