

Patient Information

Patient Name: Jane Doe
 Date of Birth: 11/08/1975
 Maternal Age at EDD: 37
 Gestational Age: 11 weeks/0 days
 Maternal Weight: N/A
 Patient ID: P99457
 Medical Record #: M84555
 Collection Kit: 123233-2-N
 Reference ID: 254233-2-N
 Accessioning ID: C47695
 Case File ID: 159466

Test Information

Ordering Physician: Dr. Matthew Goodbirth, M.D. (G123456)
 Clinic Information: Natera, Inc.
 Additional Reports: N/A
 Report Date: 02/01/2013
 Samples Collected: 01/31/2013
 Samples Received: 02/01/2013
 Mother Blood

ABOUT THIS SCREEN: Panorama™ is a screening test, not diagnostic. It evaluates genetic information in the maternal blood, which is a mixture of maternal and placental DNA, to determine the chance for specific chromosome abnormalities. The test does NOT tell with certainty if a fetus is affected, and only tests for the conditions ordered by the healthcare provider. A low risk result does not guarantee an unaffected fetus.

TEST SELECTED: Sex of Fetus, 22q11.2 Deletion

FINAL RESULTS SUMMARY
Result
LOW RISK

Fetal Sex
Male

Fetal Fraction
8.3%


Notes by the clinical reviewer, if any, will be shown here.

RESULTS DETAILS: ANEUPLOIDIES

Condition tested ¹	Result	Risk Before Test ²	Panorama Risk Score ³
Trisomy 21	Low Risk	1/152	<1/10,000
Trisomy 18	Low Risk	1/111	<1/10,000
Trisomy 13	Low Risk	1/357	<1/10,000
Monosomy X	Low Risk	1/256	<1/10,000
Triploidy	Low Risk		

RESULTS DETAILS: MICRODELETIONS

Condition tested ¹	Result	Risk Before Test ²	Risk After Test
22q11.2 deletion syndrome	Low Risk	1/2,000	1/13,300

1. Excludes cases with evidence of fetal and/or placental mosaicism. 2. Based on maternal age, gestational age, and/or general population, as applicable. References available upon request. 3. Based on a priori risk and results of analysis of circulating placental DNA.

Approved By:  Gregory M. Enns, MB, ChB, FAAP

Approved By:  Susan Zneimer, Ph.D., FACMGG, Laboratory Director

IF THE ORDERING PROVIDER HAS QUESTIONS OR WISHES TO DISCUSS THE RESULTS, PLEASE CONTACT US AT 650-249-9090 #3. Ask for the NIPT genetic counselor on call.

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OVERALL TEST SPECIFICATIONS FOR PANORAMA

The Panorama risk score shown on page 1 reflects the confidence of the algorithm for the result reported for an individual sample.

The information in the table below relates to the general performance of the test.

Sensitivity is the ability to correctly identify a truly high risk case as high risk. For example, in a group of Trisomy 21 cases, Panorama will correctly identify more than 99% of those cases.

Specificity is the ability to correctly identify an unaffected case as low risk.

Positive Predictive Value is the likelihood the result says high-risk and the fetus is actually affected. For example, when Panorama shows a high-risk result for Trisomy 21, there is a 91% chance that the fetus is affected by Trisomy 21. In other words, 9% of the time, you may get a high-risk result when the fetus is not affected by Trisomy 21.

Negative Predictive Value is the likelihood the result says low-risk and the fetus is truly not affected.

Condition	Sensitivity (95% CI)	Specificity (95% CI)	Positive Predictive Value	Negative Predictive Value
Trisomy 21 ^{1,2,3,4}	>99% (CI 97.8-99.9)	>99 (CI 99.7-100)	91%	>99.99%*
Trisomy 18 ^{1,2,3,4}	98.2% (CI 90.4-99.9)	>99% (CI 99.7-100)	93%	>99.99%*
Trisomy 13 ^{1,2,3,4}	>99% (CI 87.2-100)	>99% (CI 99.8-100)	38%	>99.99%*
Monosomy X ^{1,2,3,4}	94.7% (CI 74.0-99.9)	>99% (CI 99.7-100)	50%	>99.99%*
Triploidy ^{5,6}	>99% (CI 66.4-100)	>99% (CI 99.5-100)	5.3%	>99.99%*
XXX, XXY, XYY ⁴	N/A-Reported when identified	N/A-Reported when identified	89%	N/A-Reported when identified
22q11.2 deletion syndrome ^{7,8,9}	95.7% (CI 85.5-99.5)	>99 (CI 98.6-99.9)	20%**	99.97-99.99%***
1p36 deletion syndrome ^{7,8}	>99% (CI 2.5-100)	>99% (CI 99.1-100)	7-17%***	99.98-99.99%***
Angelman syndrome ^{7,8}	95.5% (CI 77.2-99.9)	>99% (CI 99.1-100)	4%	>99.99%
Cri-du-chat syndrome ^{7,8}	>99% (CI 85.8-100)	>99% (CI 99.1-100)	2-5%***	>99.99%
Prader-Willi syndrome ^{7,8}	93.8% (CI 69.8-99.8)	>99% (CI 99.1-100)	5%	>99.99%
Female	>99.9% (CI 99.4-100)	>99.9% (CI 99.5-100)		
Male	>99.9% (CI 99.5-100)	>99.9% (CI 99.4-100)		

1. Nicolaides KH et al. Prenat Diagn. 2013 June;33(6):575-9
 2. Pergament E et al. Obstet Gynecol. 2014 Aug;124(2 Pt 1):210-8
 3. Ryan A et al. Fetal Diagn Ther. 2016;40(3):219-223
 4. Dar P et al. Am J Obstet Gynecol. 2014 Nov;211(5):527.e1-527.e17
 5. Nicolaides KH et al. Fetal Diagn Ther. 2014;35(3):212-7.
 6. Curnow KJ et al. Am J Obstet Gynecol. 2015 Jan;212(1):79.e1-9
 7. Wapner RJ et al. Am J Obstet Gynecol. 2015 Mar;212(3):332.e1-9
 8. Ryan A et al. EUR J Hum Genet. 2016 May;24:E-supplement 1:53
 9. Natera. Panorama: SNP-Based Non-Invasive Prenatal Screening Test, white paper V3. 2017 Feb.

* Ongoing clinical follow-up is performed to ensure the NPV does not fall below the quoted value but follow up is not obtained for all low risk calls.

** PPV for 22q11.2 deletion syndrome in published studies was 20% when no ultrasound anomalies were seen and was up to 100% when ultrasound anomalies were seen prior to testing.

*** Dependent upon fetal fraction, see Panorama Risk score on report for accurate PPV/NPV for a specific patient.

For additional information, please visit: www.natera.com/panorama-test/test-specs

Testing Methodology: DNA isolated from the maternal blood, which contains placental DNA, is amplified at specific loci using a targeted PCR assay, and sequenced using a high-throughput sequencer. Sequencing data is analyzed using Natera's proprietary algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X, and Y, thereby identifying whole chromosome abnormalities at these locations, and if ordered, the microdeletion panel will identify microdeletions at the specified loci only. If a sample fails to meet the quality threshold, no result will be reported for the specified chromosome(s). The test requires sufficient fetal fraction to produce a result. Fetal fraction is determined using a proprietary algorithm incorporating data from single nucleotide polymorphism-based next-generation sequencing. Estimates of fetal fraction may differ when measured by different laboratories and/or methodologies.

Disclaimers: This test has been validated on women with a singleton pregnancy and of at least nine weeks gestation. A result will not be available where the maternal blood cells and oocytes are not of the same genetic lineage, as in the case of an egg donor, surrogate, or bone marrow transplant recipient. Findings of unknown significance will not be reported. As this assay is a screening test and not diagnostic, false positives and false negatives can occur. High risk test results need diagnostic confirmation by alternative testing methods. Low risk results do not fully exclude the diagnosis of any of the syndromes nor do they exclude the possibility of other chromosomal abnormalities or birth defects, which are not a part of this test. Potential sources of inaccurate results include, but are not limited to, mosaicism, low fetal fraction, limitations of current diagnostic techniques, or misidentification of samples. This test will not identify all deletions associated with each microdeletion syndrome. This test has been validated on full region deletions only and may be unable to detect smaller deletions. Microdeletion risk score is dependent upon fetal fraction, as deletions on the maternally inherited copy are difficult to identify at lower fetal fractions. Test results should always be interpreted by a clinician in the context of clinical and familial data with the availability of genetic counseling when appropriate. The Panorama prenatal test was developed by Natera, Inc., a laboratory certified under the Clinical Laboratory Improvement Amendments (CLIA). This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA).