

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: 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, Triploidy, Extended Panel

FINAL RESULTS SUMMARY

Result

HIGH RISK for Trisomy 21

Fetal Sex

Male

Fetal Fraction

8.3%

This is a screening test only. Genetic counseling and diagnostic testing should be offered to further evaluate these findings.

The Panorama risk score reflects analysis of DNA from the placenta. The placental DNA may not accurately reflect the status of the fetus; therefore, no irreversible decisions should be made based upon results of this screening test alone.

RESULTS DETAILS

Condition tested ¹	Result	Risk Before Test ²	Panorama Risk Score ³	Positive Predictive Values ⁴
Trisomy 21	High Risk	1/152	>99/100	T21: 91%
Trisomy 18	Low Risk	1/111	<1/10,000	T18: 93%
Trisomy 13	Low Risk	1/357	<1/10,000	T13: 38%
Monosomy X	Low Risk	1/256	<1/10,000	MX: 50%
Triploidy/Vanishing twin	Low Risk			
22q11.2 deletion syndrome	Low Risk	1/2,000 ⁴	1/13,300	
1p36 deletion syndrome	Low Risk	1/5,000 ⁴	1/12,400	
Angelman syndrome	Low Risk	1/12,000 ⁴	1/16,600	
Cri-du-chat syndrome	Low Risk	1/20,000 ⁴	1/57,100	
Prader-Willi syndrome	Low Risk	1/10,000 ⁴	1/13,800	

Positive Predictive Value (PPV) is the likelihood that diagnostic testing will confirm a High Risk result. PPV provided is NOT personalized for this patient, but calculated from a published study of 17,885 women. PPV for an individual specimen will vary based on prior risk.

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. 4. Dar P, et al. Am J Obstet Gynecol 2014;211:527.e1-17. Clinical experience and follow-up with large scale single-nucleotide polymorphism-based noninvasive prenatal aneuploidy testing.

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.

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).

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.