**Patient Information** 

VICTORIA ROMO Patient Name:

06/11/1993 Date of Birth:

Maternal Age at EDD:

12 weeks/ 1 days Gestational Age:

127 lbs Maternal Weight: 35207654 Medical Record #: 38202418-2-N Collection Kit: 15403410 Case File ID:

**Test Information** 

**Brady Coad** Ordering Physician: Clinic Information:

The Everett Clinic

N/A

Additional Reports:

Report Date: Samples Collected: Samples Received:

OB/GYN

01/28/2025 01/21/2025

01/23/2025 Mother Blood



ABOUT THIS SCREEN: Panorama 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 genetic variants or conditions. 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.

#### FINAL RESULTS SUMMARY:

#### **ANEUPLOIDIES AND MICRODELETIONS**

Result **LOW RISK** 

Fetal Sex **Female** 

Fetal Fraction(s) 10.8%



#### **FETAL RHD**

Result

#### Fetal status not assessed



The pregnant patient is RHD positive by genotype and therefore, the fetal status is not assessed. Reasons for this result type include Rh positive blood type or Rh negative blood type with Weak D, Partial D (e.g. DVI), or other rare RHD genotype.

\*Clinical management should be based upon the pregnant patient's Rh blood type result by routine serology.\* A repeat specimen is not indicated.

## **RESULT DETAILS: ANEUPLOIDIES**

Condition Tested <sup>1</sup>	Result	Risk Before Test <sup>2</sup>	Risk After Test <sup>3</sup>
Trisomy 21	Low Risk	1/461	<1/10,000
Trisomy 18	Low Risk	1/1,072	<1/10,000
Trisomy 13	Low Risk	1/3,378	<1/10,000
Monosomy X	Low Risk	1/255	<1/10,000
Triploidy	Low Risk		

#### **RESULT DETAILS: MICRODELETIONS**

Condition Tested 1 Result Risk Before Test <sup>2</sup> Risk After Test 4 22q11.2 deletion syndrome Low Risk 1/12.000

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. Risk after test for aneuploidy incorporates results from the Panorama algorithm and data from a published population study of over 1 million women [DiNonno et al.J.Clin.Med.2019.Aug 26; 8(9):1311.doi:10.3390/jcm8091311] and are reported as PPVs (high risk) and NPVs (low risk). Maternal age and fetal fraction are utilized in this calculation; however, the "risk after test" may not reflect the actual PPVs for this patient, as additional risk factors, including but not limited to: results of other screening, ultrasound findings, and personal/family history, are not included in the risk assessment. 4. Risk after test for microdeletion(s) incorporates results from the Panorama algorithm and data from multiple studies [Dar P et al. Cell-free DNA screening for prenatal detection of 22q11.2 deletion syndrome, American Journal of Obstetrics and Gynecology (2022), https://doi.org/10.1016/j.ajog.2022.01.002; Martin et al. Clin Genetics. 2017 Jul 11, Wapner R J et al. Am J Obstst Gynecol. 2015 Mar;212 (3):332 .e1-9] and are reported as PPVs (high risk) and NPVs (low risk). Risks for microdeletions are independent of maternal age and fetal fraction is utilized in this calculation; however, the "risk after test" may not reflect the actual PPV for this patient, as additional risk factors, including but not limited to: results of other screening, ultrasound findings, personal/family history, are not included in the risk assessment.



**Patient Information** 

Patient Name: VICTORIA ROMO

Date of Birth: 06/11/1993

Maternal Age at EDD: 3

Gestational Age: 12 weeks/ 1 days

Maternal Weight: 127 lbs
Medical Record #: 35207654
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**Test Information** 

Ordering Physician: Brady Coad

Clinic Information: The Everett Clinic

OB/GYN N/A

Additional Reports:

 Report Date:
 01/28/2025

 Samples Collected:
 01/21/2025

 Samples Received:
 01/23/2025

Mother Blood



Panorama\*

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 genetic variants or conditions. 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.

Testing Methodology: DNA isolated from maternal blood, which contains placental DNA, is amplified at specific loci using a targeted PCR assay and is sequenced using a high-throughput sequencer. Fetal fraction is determined using a proprietary algorithm incorporating data from single nucleotide polymorphism-based (SNP-based) next-generation sequencing [Pergament E et al. Obstet Gynecol. 2014 Aug;124(2 Pt 1):210-8]. If there is sufficient fetal fraction, sequencing data is analyzed using a proprietary SNP-based algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X and Y. If ordered, specific microdeletions will be evaluated using similar methodology [Wapner RJ et al. Am J Obstet Gynecol. 2015 Mar;212(3):332.e1-9]. If the fetal fraction is insufficient, an additional algorithm to determine whether there is an increased risk for triploidy, trisomy 18, and trisomy 13 may be utilized, known as fetal fraction based risk assessment (FFBR) [McKanna et al. Ultrasound Obstet Gynecol 2019; 53:73-79]. If ordered, and pregnant patient is RhD negative by genotype, fetal RhD status on SNP-based prenatal cell-free DNA screening. In: American Society of Human Genetics; Nov 1-5, 2023; Washington, D.C.] However, some samples will not produce a result due to failure to meet the necessary quality thresholds.

This test has been validated on women with a singleton, twin or egg donor pregnancy of at least nine weeks gestation. A result will not be available for higher order multiples and multiple gestation pregnancies with an egg donor or surrogate, or bone marrow transplant recipients. Complete test panel is not available for twin gestations and pregnancies achieved with an egg donor or surrogate. For twin pregnancies with a fetal fraction value below the threshold for analysis, a sum of the fetal fractions for both twins will be reported. As this assay is a screening test and not diagnostic, false positives and false negatives can occur.

High risk aneuploidy and microdeletion 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 phlebotomy labeling errors. This test will not identify all deletions associated with each microdeletion syndrome. This test has been validated for deletions ≥0.5 Mb within the 22q11.2 A-D region. This test has been validated on full region deletions only for 1p36 deletion syndrome, Cri-du-chat syndrome, Prader Willi syndrome and Angelman syndrome and may be unable to detect smaller deletions. Microdeletion risk score may be 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.

Fetal RhD non-invasive prenatal test does not replace the pregnant patient's serology result. False positive results may occur due to the presence of rare genotypes that include but are not limited to Weak D, Partial D, or RhD Pseudogene. False negative results, while rare, can also occur. Additional potential sources of inaccurate results include, but are not limited to, phlebotomy labeling errors, low fetal fraction, sample contamination, low DNA quantity, or low number of sequencing reads. Fetal RhD status will not be assessed for dizygotic twin pregnancies or in the context of certain maternal genetic variants. Test results should always be interpreted by a clinician in the context of clinical and familial data.

Disclaimers: The extraction, library preparation, and sequencing of this test were performed by NSTX, Inc., 13011 McCallen Pass Building A Suite 100, Austin, TX 78753 (CLIA ID 45D2093704). The data analysis and reporting of this test were performed by Natera, Inc., 201 Industrial Rd. Suite 410, San Carlos, CA 94070 (CLIA ID 05D1082992). The performance characteristics of this test were developed by NSTX, Inc.(CLIA ID 45D2093704). This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). These laboratories are regulated under CLIA as qualified to perform high-complexity testing. © 2023 Natera, Inc. All Rights Reserved.

Reviewed By: Wenbo Xu, M.D., Ph.D., FACMG, Senior Laboratory Director

CLIA Laboratory Director: J. Dianne Keen-Kim, Ph.D., FACMG

CLIA ID #05D1082992; RPT-10012 Rev. 06

Natera, Inc. 1-855-866-NIPT (6478)

IF THE ORDERING PROVIDER HAS QUESTIONS OR WISHES TO DISCUSS THE RESULTS, PLEASE CONTACT US AT 844-778-4700, option 2. Ask for the NIPT genetic counselor on call.

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#### **OVERALL TEST SPECIFICATIONS FOR PANORAMA**

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

15403410

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 (PPV) 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 95% chance that the fetus is affected by Trisomy 21. In other words, 5% of the time, you may get a high-risk result when the fetus is not affected by Trisomy 21.

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

Condition	Sensitivity (95% CI)	Specificity (95% CI)	PPV	NPV
Trisomy 21 <sup>1,2</sup>	99.0% (CI 97.1-100)	>99% (CI 99.93-99.99)	95%	>99.99% *
Trisomy 18 <sup>1,2</sup>	94.1% (CI 82.9-100)	>99% (CI 99.96-100)	91%	>99.99% <sup>*</sup>
Trisomy 13 <sup>1,2</sup>	>99% (CI 73.5-100)	>99% (CI 99.6-100)	68%	>99.99% <sup>*</sup>
Monosomy X <sup>2,3</sup>	94.7% (CI 74.0-99.9)	>99% (CI 99.7-100)	78%	>99.99% <sup>*</sup>
Triploidy <sup>4,5</sup>	>99% (CI 66.4-100)	>99% (CI 99.5-100)	7.5%	>99.99% <sup>*</sup>
XXX, XXY, XYY 6**	73.1% (CI 61.0-85.1)	99.9% (CI 99.90-99.99)	83%	99.87%
22q11.2 deletion syndrome <sup>7</sup>	83.3% (CI 51.6-97.9)	>99% (CI 99.91-99.98)	53%	99.9% (CI 99.9-100) ***
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)		
Fetal RhD+ 8	>99.9% (CI 98.9 - 100)	99.3% (CI 97.6 - 99.8)	99.4%	>99.99%

- Dar P et al. Am J Obstet Gynecol. 2022. doi: https://doi.org/10.1016/j.ajog.2022.01.019 DiNonno W et al. J Clin Med. 2019. 26;8(9):1311. doi: https://doi.org/10.3390/jcm8091311
- Martin et al. ISUOG World Congress 2022: September, 2022 Nicolaides KH et al. Fetal Diagn Ther. 2014. 35(3):212-7. doi: https://doi.org/10.1159/000355655 Kantor et al. Prenat Diagn. 2022. 42;(8): 994-999. Doi: 10.1002/pd.6169 Martin K et al. Genet in Med. 2023. doi: https://doi.org/10.1016/j.gim.2023.100879
- Dar P et al. Am J Obstet Gynecol. 2022. doi: https://doi.org/10.1016/j.ajog.2022.01.002
- Natera internal validation data. 2024

- 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
- Sex chromosome trisomies are only reported when clearly identified. At lower fetal fractions, identification of sex chromosome trisomies
- may not be possible.

  Dependent upon fetal fraction. For 22q11.2 deletion syndrome, only the paternal allele is evaluated at FF <6.5%. For 1p36 deletion syndrome and Cri-du-chat syndrome, only the paternal allele is evaluated at FF <7%. For Angelman syndrome, no risk assessment is reported at FF<7%. For Prader-Willi syndrome, no risk assessment is reported at FF≤2.8%.

Test specifications above are applicable to singleton and monozygotic twin pregnancies only. For additional information, please visit:



# **Understanding Your Results**

## Low risk



## What do my results mean?

Your results show that there is a low risk to your baby for the chromosome conditions listed on the report. These results cannot tell with certainty that your baby does not have these conditions. The specific chance that your baby has each condition can be found on page 1 of your test report under "Risk after test." Most people with low risk results do not choose to have further testing for these chromosome conditions.<sup>1</sup>



#### What should I do next?

You should talk to your healthcare provider about these results and continue with the prenatal care recommended for you. Although the chance that your baby has these chromosome conditions is low, you have the option of doing further testing during pregnancy to find out for sure if your baby has these conditions. These tests are called CVS (chorionic villus sampling) and amniocentesis, and both have a small risk of miscarriage. Please talk to your healthcare provider if you have questions about further testing.



NEVA\* is always available to help you learn about your results. You can connect with Natera's Educational Virtual Assistant (NEVA) by logging into the patient portal at my.natera.com.







If you would like to discuss your results with a Natera genetic counselor, you can schedule a free information session at <u>naterasession.com</u>, by texting SESSION to 636363\*, or by calling +1.877.476.4743. Please select Panorama Non-Invasive Prenatal Chromosome Screening Post-Test as the appointment type.

You can find a local genetic counselor through the National Society of Genetic Counselors at findageneticcounselor.nsgc.org.
\*Text scheduling is available only in the United States.



1. van Schendel RV, et al; Dutch NIPT Consortium. Women's Experience with Non-Invasive Prenatal Testing and Emotional Well-being and Satisfaction after Test-Results. J Genet Couns. 2017 Dec;26(6):1348-1356.

doi: 10.1007/s10897-017-0118-3. Epub 2017 Jun 30. PMID: 28667567; PMCID: PMC5672853. 13011 McCallen Pass, Building A Suite 100 | Austin, TX 78753 | natera.com



## **Understanding Your Results**

## Baby's predicted RhD factor not determined



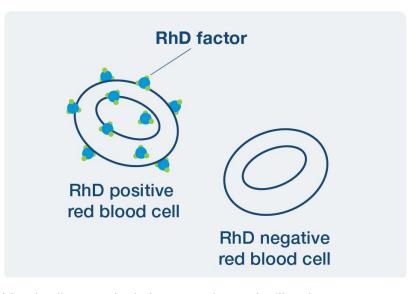
#### What does this result mean?

This result means that your baby's RhD factor was not determined. There are two main reasons that a baby's RhD factor is not determined by this test. First, your own RhD factor could mean that it is not necessary to determine your baby's RhD factor. Second, sometimes the lab is not able to perform the test due to technical limitations.



## What is RhD testing and why is it important in pregnancy?

RhD factor is a protein that can be present or absent on the surface of a person's red blood cells. RhD positive means that someone's red blood cells have the RhD protein on them. People who are RhD negative have red blood cells without this protein. The RhD factor can cause problems in a pregnancy if an RhD negative pregnant person's blood is mixed with RhD positive blood from the baby. When this happens, the RhD negative person's body will recognize



the RhD factor on the RhD positive blood cells from the baby as foreign and will make antibodies to fight the RhD factor. When a pregnant person has RhD factor antibodies, they are said to be sensitized. If a sensitized pregnant person is carrying an RhD positive baby, the antibodies can cross the placenta and attack the baby's red blood cells. The antibodies can cause a loss of red blood cells, which can lead to serious health problems for the baby. When a pregnant person and their baby have different RhD factors, it is called RhD incompatibility.



## Can I repeat this test to get an answer?

It depends on the reason that you did not get a result. Your lab report could say one of two things.

- 1. Fetal status not assessed. If your lab report says this, you should not repeat this test. You can get this result for one of two reasons.
  - "Fetal status not assessed" could mean that you are RhD positive. RhD incompatibility is not a risk to your baby if you are RhD positive.
  - Rarely, a result of "fetal status not assessed" could mean that you are RhD negative due to having what is called the *RHD* pseudogene. When a pregnant person has the *RHD* pseudogene, the lab does not determine the baby's RhD factor.

If your lab report says, "fetal status not assessed," please talk to your healthcare provider about looking at the results of other blood tests you had during pregnancy to determine what to do next.

2. No results for fetal RHD. If your lab report says this, the lab was not able to get a result. There are a few different reasons the lab would not be able to get a result. The reason you did not get a result is listed on your report. Sometimes this problem will not be solved by sending another blood sample. Many times the lab will be able to get a result on another sample. If another sample can be sent, your lab report will say so.



## What should I do next?

Talk to your healthcare provider about next steps. Looking at the results of other blood tests you have had during pregnancy can sometimes help to explain this result. Sometimes your healthcare provider will recommend different blood tests, or it can sometimes be helpful to repeat this test. If you are RhD negative and it is important to know the baby's RhD factor, you could have the option of a test called CVS (chorionic villus sampling) or amniocentesis. These tests can tell you your baby's RhD factor. Both tests have a small risk of miscarriage.



Please talk to your healthcare provider about your result. If you would also like to discuss your result with a Natera genetic counselor, you can schedule a free information session at <a href="mailto:naterasession.com">naterasession.com</a>, by texting\* SESSION to 636363, or by calling 1.877.467.4743. Please select Panorama non-invasive prenatal chromosome screening, post-test as the appointment type.

You can find a local genetic counselor through the National Society of Genetic Counselors at <u>findageneticcounselor.nsgc.org</u>.



\*Text scheduling is available only in the United States

