Ф.И.О.: **ИВАНОВА МАРЬЯ ИВАНОВНА**
Дата рождения: 24.03.1986 (32 г.) Пол: Ж
Регистрации биоматериала: 02.04.2018
Биоматериал: Кровь с ЭДТА;
Взятие биоматериала: 02.04.2018 13:15

Заявка №: 3300079160
Заказчик: **МО Тверская**
Беременность: 10 нед.

Неинвазивный пренатальный тест

РЕЗУЛЬТАТЫ ИССЛЕДОВАНИЯ

Результат	Пол плода	Фетальная фракция
Низкий риск	Мужской	8,3%

Тестируемое состояние[1]	Результат	Риск до теста[2]	Риск после теста[3]
Трисомия 21	Низкий риск	1/424	<1/10,000
Трисомия 18	Низкий риск	1/860	<1/10,000
Трисомия 13	Низкий риск	1/2,740	<1/10,000
Моносомия X	Низкий риск	1/255	<1/10,000
Триплоидия	Низкий риск		

*[1] За исключением случаев фетального/плацентарного мозаицизма.
[2] Основан на возрасте женщины, гестационном сроке и/или общепопуляционном риске. Ссылки доступны по запросу
[3] Для анеуплоидий включает результаты алгоритма Panorama (анализа циркулирующей ДНК плода в крови матери) и данные опубликованного исследования 17,885 женщин и сообщается как «высокий риск» (положительная прогностическая значимость, ППЗ) или «низкий риск» (отрицательная прогностическая значимость, ОПЗ). В расчете используется возраст матери, однако «риск после теста» может не отражать актуальную ППЗ для этого пациента, т.к. результаты другого скрининга, ультразвуковые исследования, личный/ семейный анамнез, не включены в оценку риска.

Дата выполнения исследования: **20.04.2018** Результаты одобрил: **Зайцева С. А.**

Patient Information

Patient Name:	Maria Ivanova
Date of Birth:	03/24/1986
Maternal Age at EDD:	32
Gestational Age:	10 weeks/1 days
Maternal Weight:	145.51 lbs
Collection Kit:	4603196-2-N
Reference ID:	5335116-2-N
Case File ID:	1750786

Test Information

Ordering Physician:	Serebrenikova
Clinic Information:	LLC Progen
Additional Reports:	N/A
Report Date:	04/14/2018
Samples Collected:	04/02/2018
Samples Received:	04/09/2018
	Mother Blood

panorama
natera prenatal screen

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.

FINAL RESULTS SUMMARY

Result	*Fetal Sex*	*Fetal Fraction*
LOW RISK	**Male**	**8.3%**

RESULT DETAILS: ANEUPLOIDIES

Condition tested[1]	*Result*	*Risk Before Test*[2]	*Risk After Test*[3]
Trisomy 21	Low Risk	1/424	<1/10,000
Trisomy 18	Low Risk	1/860	<1/10,000
Trisomy 13	Low Risk	1/2,740	<1/10,000
Monosomy X	Low Risk	1/255	<1/10,000
Triploidy	Low 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. Risk after test for aneuploidy incorporates results from the Panorama algorithm and data from a published study of 17,885 women [Dar et al. Am J Obstet Gynecol. 2014. Nov;211(5):527.e1-27.e17] and are reported as PPV (high risk) and NPV (low risk). Maternal age 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.

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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panorama® natera prenatal screen

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OVERALL TEST SPECIFICATIONS FOR PANORAMA

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]	90.0% (CI 55.5-99.7)	>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)	10%**	>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. Martin et al. Clin Genetics. 2017 Jul 11
9. Norvez A et al. The European Human Genetics Conference, ESHG. Copenhagen, Denmark. May 27–30, 2017.

* 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 and Angelman syndrome in published studies was 20% and 10% respectively when no ultrasound anomalies were seen and was up to 100% when ultrasound anomalies were seen prior to testing.

*** 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: ***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, 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. 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).