LifeLabs GENETICS™
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PATIENT INFORMATION

Patient Name:	TEST TEST
Date of Birth:	1991-11-15
Estimated Due Date:	2022-04-15
Maternal Age at EDD:	30 (25 at Egg Retrieval)
Gestational Age:	18 Weeks / 0 Days
Maternal Weight:	-
Health Card #:	-
Order ID:	AUC80
Sample ID:	M80
Accessioning ID:	11_PANO_SING_MD_HDOR

PROVIDER INFORMATION

Ordering Physician:	TEST TEST
Genetic Counsellor:	-
Additional Reports:	-
	-
Test Requested:	Panorama Prenatal Test Microdeletion Extended Panel
Report Date:	2024-10-15
Samples Collected:	2021-11-12
Samples Received:	2021-11-15
	Maternal 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.

FINAL REPORT SUMMARY

Result	*Fetal Sex*	*Fetal Fraction*
LOW RISK	**Female**	**7.0%**

RESULT DETAILS

Condition Tested[1]	*Result*	*Risk Before Test*[2]	*Risk After Test*[3]
Trisomy 21	Low Risk	4/1,000	<1/10,000
Trisomy 18	Low Risk	2/1,000	<1/10,000
Trisomy 13	Low Risk	5/10,000	<1/10,000
Monosomy X	Low Risk	4/1,000	<1/10,000
Triploidy/Vanishing Twin	Low Risk		

MICRODELETIONS

Condition Tested[1]	*Result*	*Risk Before Test*[2]	*Risk After Test*[4]
22q11.2 deletion syndrome	Low Risk	1/2,000	1/12,000
1p36 deletion syndrome	Low Risk	1/5,000	1/12,400
Angelman syndrome	Low Risk	1/12,000	1/16,600
Cri-du-chat syndrome	Low Risk	1/20,000	1/57,100
Prader-Willi syndrome	Low Risk	1/10,000	1/13,800

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 (2) and are reported as PPVs (high risk) and NPVs (low risk). Maternal age and fetal fraction are utilized in this calculation*. 4. Risk after test for microdeletion(s) incorporates results from the Panorama algorithm and data from multiple studies (7,8,9) and are reported as PPVs (high risk) and NPVs (low risk). Risks for microdeletions are independent of maternal age. Fetal fraction is utilized in this calculation*. *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.

TESTING METHODOLOGY: DNA 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 algorithms to determine the fetal copy number for chromosomes 13, 18, 21, X, and Y, thereby identifying whole chromosome abnormalities at these locations. The optional 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 of at least 2.8% to produce a result. Fetal fraction is the percentage of fetal (placental) DNA in the maternal plasma compared to the amount of maternal DNA. 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 positive and false negatives can occur. High risk test results need diagnostic confirmation by alternative testing methods, such as chorionic villus sampling (CVS) or amniocentesis. 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 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 for deletions of 0.5Mb or greater 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 the clinical and familial data with the availability of genetic counselling when appropriate. The Panorama prenatal test was developed by Natera, Inc., 201 Industrial Road Suite 410, San Carlos, CA 94070., 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).

Condition	Sensitivity (95% CI)	Specificity (95% CI)	Positive Predictive Value	Negative Predictive Value
Trisomy 21[1,2]	99.0% (CI 97.1-100)	>99% (CI 99.93-99.99)	95%	>99.99%*
Trisomy 18[1,2]	94.1% (CI 82.9-100)	>99% (CI 99.96-100)	91%	>99.99%*
Trisomy 13[1,2]	>99% (CI 73.5-100)	>99% (CI 99.6-100)	68%	>99.99%*
Monosomy X[2,3]	94.7% (CI 74.0-99.9)	>99% (CI 99.7-100)	78%	>99.99%*
Triploidy[4,5]	>99% (CI 66.4-100)	>99% (CI 99.5-100)	7.5%	>99.99%*
XXX, XXY, XYY[6] ****	73.1% (CI 61.0-85.1)	99.9% (CI 99.90-99.99)	86.4%	99.87%
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)		
22q11.2 deletion syndrome[7]	83.3% (CI 51.6-97.9)	>99% (CI 99.91-99.98)	53%**	99.9%***
1p36 deletion syndrome[8,9]	>99% (CI 2.5-100)	>99% (CI 99.1-100)	7-17%***	99.98-99.99%***
Angelman syndrome[8,9]	95.5% (CI 77.2-99.9)	>99% (CI 99.1-100)	10%**	>99.99%
Cri-du-chat syndrome[8,9]	>99% (CI 85.8-100)	>99% (CI 99.1-100)	2-5%***	>99.99%
Prader-Willi syndrome[8,9]	93.8% (CI 69.8-99.8)	>99% (CI 99.1-100)	5%	>99.99%

1. Dar P et al. Am J Obstet Gynecol. 2022. doi: https://doi.org/10.1016/j.ajog.2022.01.019
2. DiNonno W et al. J Clin Med. 2019. 26;8(9):1311. doi: https://doi.org/10.3390/jcm8091311
3. Martin et al. ISUOG World Congress 2022: September, 2022.
4. Nicolaides KH et al. Fetal Diagn Ther. 2014. 35;(3):212-7. doi: https://doi.org/10.1159/000355655
5. Kantor et al. Prenat Diagn. 2022. 42;(8): 994-999. doi: https://doi.org/10.1002/pd.6169
6. Martin K et al. ISPD 25th International Conference: June, 2021
7. Dar P et al. Am J Obstet Gynecol. 2022. doi: https://doi.org/10.1016/j.ajog.2022.01.002
8. Martin K et al. Clin Genet. 2018. 93(2):293-300. doi: https://doi.org/10.1111/cge.13098
9. Wapner RJ et al. Am J Obstet Gynecol. 2015. 212(3):332.e1-9. doi: https://doi.org/10.1016/j.ajog.2014.11.041

*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 53% 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%.
****Sex chromosome abnormalities are only reported when identified.
Test specifications above are applicable to singleton and monozygotic twin pregnancies only. For additional information, please visit: www.natera.com/panorama-test/test-specs

AUTHORIZED BY **R.F. Carter, PhD, FCCMG (Laboratory Director)**

IF THE ORDERING PROVIDER HAS QUESTIONS OR WISHES TO DISCUSS THE RESULTS, PLEASE CONTACT US AT 1-844-363-4357. Ask for the NIPT genetic counsellor on call. For more information, please visit www.lifelabsgenetics.com. LifeLabs Genetics is accredited by the Institute for Quality Management in Healthcare (IQMH) and College of American Pathologists (CAP) and is licensed by the Ontario Ministry of Health to operate as a clinical genetic laboratory: MOH license 5806.