

PANORAMA 無創產前測試篩檢

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.

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 2 ^{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 ^{1,2,2,4}	>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 et al. Prenat Diagn 2013 June 33/6:1575-9
 2. Pergament E et al Obstet Gynecol. 2014 Aug 124:12 Pt 11210-8
 3. Ryan A etw. Fetal Diagn Ther. 2016;40(3):219-223
 4. Dar Pat et al. Am J Obstet Gynecol. 2014 Nov 211:515-527.e1-527.e17
 5. Nicolaides et al. Fetal Diagn Ther. 2014;35(3):212-7.
 6. Curnow KJ et al. Am J Obstet Gynecol. 2015 Jan;212:1179 1-9
 7. Wapner R et al. Am J Obstet Gynecol. 2015 Mar;212(3):332.e1-9
 8. Martin et al. Clin Genetics. 2017 Jul 11
 9. Narvez 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 able 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 contain placental DNA sampled 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 the estimated fetal fraction is ≥2.8%, sequencing data is analyzed using a proprietary SNP-based algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X and Y [Ryan A et al. Am J Obstet Gynecol. 2014 Nov;211(5):527.e1-527.e17]. 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 a sample fails to meet the quality threshold, or the fetal fraction is insufficient an additional algorithm is utilized to determine whether there is an increased risk for triploidy, trisomy 18 and trisomy 13 [McKanna et al. The European Human Genetics Conference. Copenhagen, Denmark, May 27-30, 2017]. However, some samples will not produce a result due to failure to meet the necessary quality thresholds.

Disclaimer: 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).