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)		

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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
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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
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* 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***