panorama™ prenatal test

TEST REPORT

Report date: 4/4/2013
Case file ID: 20
Collection kit bar code

Patient Name: A Patient
Date of Birth: 01/01/1970
Maternal Age at EDD: 20
Gestational Age: 18 weeks/6 days

Ordering Physician: Dr. Pepper, MD
Additional Reports Sent to: N/A
Clinic Information: IVF, Inc.

Samples Collected: 1/1/2013
Samples Received: x Mother Blood
x Father Cheek

LOW RISK for Trisomies 21, 18, 13, and Monosomy X and Triploidy.

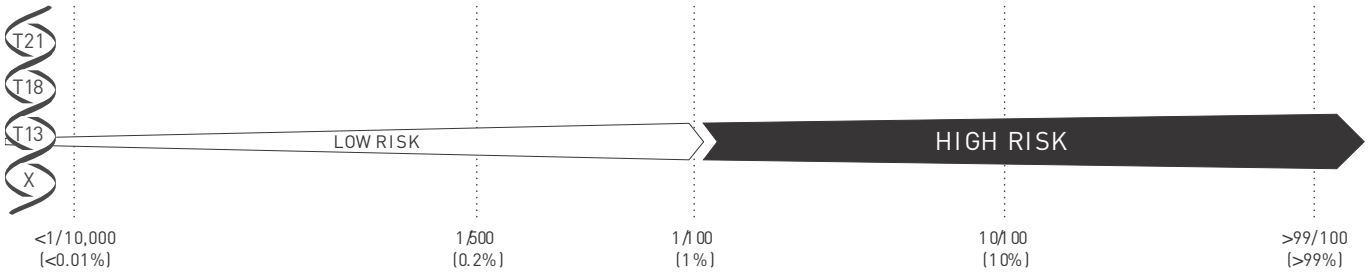
RESULTS [1]

SEX OF FETUS: Male

Fetal fraction: xx%

Condition tested	Age-based risk [2]	Panorama risk score [3]	Result	Comments
Trisomy 21	1/302 (0.33%)	<1/10,000 (<0.01%)	Low Risk	none
Trisomy 18	1/1,142 (0.09%)	<1/10,000 (<0.01%)	Low Risk	none
Trisomy 13	1/3,419 (0.03%)	<1/10,000 (<0.01%)	Low Risk	none
Monosomy X	1/568 (0.18%)	<1/10,000 (<0.01%)	Low Risk	none
Triploidy/Vanishing twins			Low Risk	none

[1]Excludes cases with evidence of fetal and/or placental mosaicism. [2]Based on maternal age and gestational age where applicable [3]Based on a priori risk and test results



Interpretation: Reasons for uninformative DNA pattern can include: higher than expected levels of homoygosity (i.e. haploblocks) that affect the test bioinformatics, clarity of maternal SNP profile, or use of egg donors and/or surrogates. Quality metrics include, but are not limited to total DNA amount (maternal and fetal combined), number of SNP reads, and other metrics that are in place to ensure good quality data for accurate, consistent results. Results indicating presence of vanishing twins/triploidy may require additional clinical evaluation such as ultrasound and/or review of medical records along with invasive testing to confirm presence of triploidy. Multiple gestations may complicate these results and are considered outside of test specifications. **Testing Methodology:** DNA isolated from the maternal blood, which contains fetal DNA, is amplified at 19,500 loci using a targeted PCR assay, and sequenced using an Illumina HiSeq high-throughput sequencer. Where available, paternal genomic DNA is amplified and sequenced using the same protocol. Sequencing data is analyzed using the NATUS algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X and Y, thereby identifying any whole chromosome abnormalities at those chromosomes. If a sample fails to meet the quality threshold, no result will be reported for that chromosome and a redraw may be requested. **Limitations:** This test has been validated on women with a singleton pregnancy, and of at least nine weeks gestational age. Tests run prior to 9 weeks have an increased no result rate. This test will not return results on pregnancies conceived with an egg donor or those which used a surrogate and cannot be performed on women who have received a bone marrow transplant. If a paternal sample is submitted and non-paternity is identified, it will not be reported and the paternal sample will not be used in the analysis. Samples are analyzed for aneuploidy of chromosomes 13, 18, 21, X and Y only. Abnormalities on other chromosomes or those involving only a portion of the chromosomes tested cannot be excluded. This test may not be able to identify abnormalities or may report a positive result in the presence of mosaicism (which may be confined to the placenta). Fetal sex will be reported as male or female based on presence or absence of a Y chromosome and does not confirm presence or absence of SRY. Gender will not be reported in cases consistent with triploidy/vanishing twins. Pregnancies involving multiples or abnormal ultrasound findings may be better served by other screening or testing options. There is a chance of detecting maternal sex chromosome abnormalities during this testing process (either in full or mosaic form), which, if present, may interfere with the accuracy of the results on the fetal sex chromosomes. Although this test has a high accuracy, the results are not diagnostic. These results should always be interpreted by a clinician in the context of clinical and familial data.

Test Specifications [1]

	SENSITIVITY	SPECIFICITY
T21	>99% CI:[92.5%-100%]	>99% CI:[99.5%-100%]
T18	>99% CI:[78.2%-100%]	>99% CI:[99.5%-100%]
T13	>99% CI:[59.0%-100%]	>99% CI:[99.5%-100%]
MX	92% CI:[61.5%-99.8%]	>99% CI:[99.5%-100%]
Triploidy/ Vanishing Twins	>99% CI:[47.8%-100%]	N/A[2]

[1] Excludes cases with evidence of fetal and/or placental mosaicism
[2] Due to inability to distinguish between vanishing twins and triploidy, specificity cannot be calculated.

References: Snijders RJ, et al. Maternal age- and gestation-specific risk for trisomy 21. Ultrasound Obstet Gynecol 1999 Mar;13(3):167-70. Snijders etal. Maternal age and gestational age specific risk for chromosomal defects. Fetal Dign Ther 1995 Nov-Dec;10(6):356-67. Gravholt CH, et al. Prenatal and postnatal prevalence of Turner syndrome: a registry study. BMJ 1996; 312:16-21. Levy B, et al. Use of targeted sequencing of SNPs to achieve highly accurate non-invasive detection of fetal aneuploidy of chromosomes 13, 18, 21, and sex chromosomes in Society of Maternal Fetal Medicine(San Francisco 2013). Zimmerman B, et al. Noninvasive prenatal aneuploidy testing of chromosomes 13, 18, 21, X, and Y, using targeted sequencing of polymorphic loci. Prenat Diagn 2012 Dec; 32(13):1233-41.

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APPROVED BY: Greg M. Enns MB, CHB, FAAP

APPROVED BY: Susan Zneimer, Ph.D., FACMG, Laboratory Director

IF THE ORDERING PROVIDER HAS QUESTIONS OR WISHES TO DISCUSS THE RESULTS, PLEASE CONTACT US AT **650-249-9092** AND ASK FOR THE GENETIC COUNSELOR ON CALL.

CLIA ID#05D1082992
Natera, Inc., 1-855-866-NIPT (6478)
201 Industrial Road Suite 410, San Carlos, CA 94070

Report ID Customer

natera™

panorama™ prenatal test

TEST REPORT

Report date: 4/4/2013
Case file ID: 20
Collection kit bar code

Patient Name: A Patient
Date of Birth: 01/01/1970
Maternal Age at EDD: 20
Gestational Age: 18 weeks/6 days

Ordering Physician: Dr. Pepper, MD
Additional Reports Sent to: N/A
Clinic Information: IVF, Inc.

Samples Collected: 1/1/2013
Samples Received: x Mother Blood
x Father Cheek

Results are consistent with a possible triploid or vanishing twin pregnancy. Follow-up counseling and testing is recommended.

Although these results increase the risk for a triploid fetus, the possibility of a vanishing twin pregnancy (or unrecognized multiple gestation) cannot be excluded. Review of clinical history along with ultrasound findings and possible diagnostic prenatal testing is recommended to fully interpret results.

RESULTS [1]

SEX OF FETUS: N/A

Fetal fraction: xx%

Condition tested	Age-based risk [2]	Panorama risk score [3]	Result	Comments
Trisomy 21	1/229 (0.44%)	N/A	No result	none
Trisomy 18	1/465 (0.22%)	N/A	No result	none
Trisomy 13	1/1,481 (0.07%)	N/A	No result	none
Monosomy X	1/255 (0.39%)	N/A	No result	none
Triploidy/Vanishing twins			Increased risk	Follow up counseling and testing recommended

[1]Excludes cases with evidence of fetal and/or placental mosaicism. [2]Based on maternal age and gestational age where applicable [3]Based on a priori risk and test results

Interpretation: Reasons for uninformative DNA pattern can include: higher than expected levels of homoygosity (i.e. haploblocks) that affect the test bioinformatics, clarity of maternal SNP profile, or use of egg donors and/or surrogates. Quality metrics include, but are not limited to total DNA amount (maternal and fetal combined), number of SNP reads, and other metrics that are in place to ensure good quality data for accurate, consistent results. Results indicating presence of vanishing twins/triploidy may require additional clinical evaluation such as ultrasound and/or review of medical records along with invasive testing to confirm presence of triploidy. Multiple gestations may complicate these results and are considered outside of test specifications. **Testing Methodology:** DNA isolated from the maternal blood, which contains fetal DNA, is amplified at 19,500 loci using a targeted PCR assay, and sequenced using an Illumina HiSeq high-throughput sequencer. Where available, paternal genomic DNA is amplified and sequenced using the same protocol. Sequencing data is analyzed using the NATUS algorithm to determine the fetal copy number for chromosomes 13, 18, 21, X and Y, thereby identifying any whole chromosome abnormalities at those chromosomes. If a sample fails to meet the quality threshold, no result will be reported for that chromosome and a redraw may be requested. **Limitations:** This test has been validated on women with a singleton pregnancy, and of at least nine weeks gestational age. Tests run prior to 9 weeks have an increased no result rate. This test will not return results on pregnancies conceived with an egg donor or those which used a surrogate and cannot be performed on women who have received a bone marrow transplant. If a paternal sample is submitted and non-paternity is identified, it will not be reported and the paternal sample will not be used in the analysis. Samples are analyzed for aneuploidy of chromosomes 13, 18, 21, X and Y only. Abnormalities on other chromosomes or those involving only a portion of the chromosomes tested cannot be excluded. This test may not be able to identify abnormalities or may report a positive result in the presence of mosaicism (which may be confined to the placenta). Fetal sex will be reported as male or female based on presence or absence of a Y chromosome and does not confirm presence or absence of SRY. Gender will not be reported in cases consistent with triploidy/vanishing twins. Pregnancies involving multiples or abnormal ultrasound findings may be better served by other screening or testing options. There is a chance of detecting maternal sex chromosome abnormalities during this testing process (either in full or mosaic form), which, if present, may interfere with the accuracy of the results on the fetal sex chromosomes. Although this test has a high accuracy, the results are not diagnostic. These results should always be interpreted by a clinician in the context of clinical and familial data.

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TEST REPORT

Report date: 4/4/2013
Case file ID: 20
Collection kit bar code

Patient Name: A Patient
Date of Birth: 01/01/1970
Maternal Age at EDD: 20
Gestational Age: 18 weeks/6 days

Ordering Physician: Dr. Pepper, MD
Additional Reports Sent to: N/A
Clinic Information: IVF, Inc.

Samples Collected: 1/1/2013
Samples Received: x Mother Blood
x Father Cheek

LOW RISK for Trisomies 21, 18, and 13, and Monosomy X.

RESULTS [1]

SEX OF FETUS: Male
Fetal fraction: xx%

Condition tested	Age-based risk[2]	Panorama risk score[3]	Result	Comments
Trisomy 21	1/302 (0.33%)	<1/10,000 (<0.01%)	Low Risk	none
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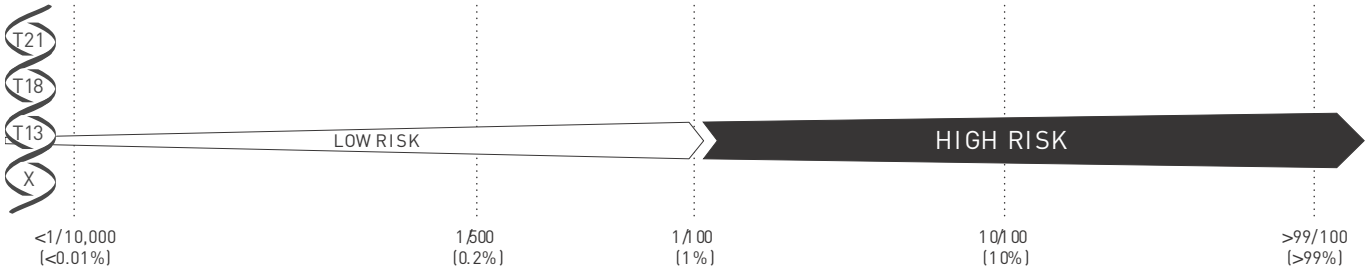

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TEST REPORT

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Patient Name: A Patient
Date of Birth: 01/01/1970
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Gestational Age: 18 weeks/6 days

Ordering Physician: Dr. Pepper, MD
Additional Reports Sent to: N/A
Clinic Information: IVF, Inc.

Samples Collected: 1/1/2013
Samples Received: x Mother Blood
x Father Cheek

Unable to report due to uninformative DNA pattern. Redraw is not recommended.

Reasons for uninformative DNA pattern can include: higher than expected levels of homozygosity (i.e.haplob-locks) that affect the test bioinformatics, multiple gestations, vanishing twins, triploidy, clarity of maternal SNP profile, or use of egg donors and/or surrogates

RESULTS [1]

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Fetal fraction: xx%

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Samples Collected: 1/1/2013
Samples Received: x Mother Blood
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Unable to report due to low fetal fraction. Submission of a new sample is required for testing.

RESULTS [1]

SEX OF FETUS: N/A

Fetal fraction: N/A

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APPROVED BY: Greg M. Enns MB, CHB, FAAP

APPROVED BY: Susan Zneimer, Ph.D., FACMG, Laboratory Director

IF THE ORDERING PROVIDER HAS QUESTIONS OR WISHES TO DISCUSS THE RESULTS, PLEASE CONTACT US AT **650-249-9092** AND ASK FOR THE GENETIC COUNSELOR ON CALL.