Quest Diagnostics

Report Status: Final

TEST, QNATAL1

Patient Information	Specimen Information	Client Information
TEST, QNATAL1 **DOB: 03/28/1970 AGE: 46** Gender: F Phone: NG Patient ID: VE40272017	Specimen: VE40272017 Requisition: Lab Ref #: DB027070V Collected: 05/13/2016 / 02:00 PDT Received: 05/13/2016 / 10:55 PDT Reported: 05/16/2016 / 17:30 PDT	Client #: Not Given COLMENAR QUEST DIAGNOSTICS INCORPORATED Attn: REFERRAL TESTING 400 EGYPT RD NORRISTOWN, PA 19403

QNatal® Advanced

Interpretation Summary **Lab: EZ**

This specimen showed expected representation of chromosome 21, 18, and 13 material.

Chromosome Results

Chromosome Tested	Results
Trisomy 21 (T21)	Negative
Trisomy 18 (T18)	Negative
Trisomy 13 (T13)	Negative

Fetal Sex Result

Y Chromosomal material Not detected	**Consistent with a female fetus.** 

Pregnancy Data

Fetal Fraction	12%
Number of Fetuses	1
Gestational Age	
Weeks	32
Days	5

Additional Chromosome Results

Chromosome Tested	Results	Interpretation
Sex Chromosome	No aneuploidy	No apparent abnormality was detected. See "Limitations" below.
Microdeletion	Not detected	No apparent abnormality was detected. See "Limitations" below.

Laboratory Comments **Lab: EZ**

Laboratory results and submitted clinical information reviewed by Weimin Sun, Ph.D., ABMG, CGMB.

Clinician Provided High-Risk Indications **Lab: EZ**

Component	Response	Component	Response
Advanced Maternal Age	NO	Abnormal Ultrasound	NO
Abnormal Maternal Serum Screening	NO	Personal/Family History	NO

Healthcare providers, please contact your local Quest Diagnostics genetic counselor or call 866-GENEINFO (866-436-3463) for assistance with interpretation of these results.

Limitations

This test has been validated on women with a singleton pregnancy that is >=10 weeks gestational age. As such, the accuracy of this test for specimens drawn at less than 10 weeks gestation is unknown. In addition, there are limited data available for the performance of this test in multiple gestation pregnancies and for the detection of microdeletions. Specimens are analyzed for aneuploidies involving chromosomes 21, 18, 13, X, and Y, and microdeletions of the specified regions only. The Y chromosome is analyzed for the determination of fetal sex, and the sensitivity and specificity of this analysis may be less than that of the autosome analysis. Sex chromosome aneuploidy analysis is not performed for multiple gestation pregnancies. Aneuploidies involving chromosomes other than those specified above or abnormalities involving chromosomal regions other than those specified are not included in this testing. While the results of this test are highly accurate, not all of the chromosome abnormalities interrogated may be detected due to maternal, placental, or fetal mosaicism, or other unexplained causes. The accuracy of the test results may also be affected by the presence of chromosome abnormalities or copy number variations that are maternal in origin, or by vanishing twin syndrome in a multiple gestation pregnancy. Circulating cell-free fetal DNA screening does not replace the precision of diagnosis using chorionic villus sampling or amniocentesis. It does not assess the risk of fetal anomalies such as neural tube defects or ventral wall defects, and should not be considered in isolation from other clinical findings and laboratory test results. Management decisions, including pregnancy termination, should not be based solely on the results of cell-free DNA screening. A negative test result does not ensure an unaffected pregnancy. The healthcare provider is responsible for the use of this information in the management of his/her patient.

Health care providers, please contact your local Quest Diagnostics genetic counselor or call Genomics Client Services at 866-GENEINFO (866-436-3463) for assistance with interpretation of these results.

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Methodology

Circulating cell-free (cf) DNA was isolated from plasma. It was then detected on a massively parallel sequencing platform. Bioinformatic analysis was performed to determine the representation of fetal DNA in the specimen, especially fetal material from chromosomes 21, 18, and 13. The representation of other fetal material, including the sex chromosomes (X and Y) and select chromosomal regions (22q, 15q, 11q, 8q, 5p, 4p 1p), was also evaluated and will only be reported as "Additional Chromosome Results" when an abnormality is detected. This test was developed and its performance characteristics have been determined by Quest Diagnostics Nichols Institute, San Juan Capistrano. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. Performance characteristics refer to the analytical performance of the test. This test is performed pursuant to a license agreement with Sequenom Laboratories.

Test Specifications

Intended Use	Performance	
Trisomy 21	Sensitivity	>99.9%
	Specificity	>99.9%
Trisomy 18	Sensitivity	>99.9%
	Specificity	>99.9%
Trisomy 13	Sensitivity	>99.9%
	Specificity	>99.9%
Y Chromosome	Accuracy	>99.9%

Performance of the QNatal Advanced laboratory-developed test (LDT) has been determined based on internal analytical assessment.

PERFORMING SITE:

EZ QUEST DIAGNOSTICS/NICHOLS SJC, 33608 ORTEGA HWY, SAN JUAN CAPISTRANO, CA 92675-2042 Laboratory Director: JON NAKAMOTO, MD PHD, CLIA: 05D063352

This is supplemental to your standard report.

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