MaterniT21® PLUS Lab Report

Sequenom Laboratories
3595 John Hopkins Court
San Diego, CA 92121
CLIA #: 05D2015356 CAP #: 7527138
Lab Director: Phillip Cacheris, MD, PhD
877.821.7266

Final Report

MaterniT® 21 PLUS (Core)
Singleton Gestation

Ordering Provider:	Last, First	Patient:	**Last, First**
Provider Location:	Sequenom SD	DOB:	mm/dd/year
Provider Phone:		Specimen:	1234567890
Date Ordered:	09/24/2019	Fetal Fraction:	7%
Date Collected:	09/24/2019	Gestational Age ≥ 9w:	Yes
Date Received:	09/24/2019	External Accession:	
Order ID:	xxx1234567	Referral Clinician:	
Patient ID:	1234567890	Date Reported:	**01/10/2020 10:51 AM PT**

Test Result

Negative

Lab Director Comments

This specimen showed an expected representation of chromosome 21, 18 and 13 material. Clinical correlation is suggested.

Result Table

Content	Result
FETAL SEX	**Male**
AUTOSOMAL ANEUPLOIDIES	
Trisomy 21 (Down syndrome)	**Negative**
Trisomy 18 (Edwards syndrome)	**Negative**
Trisomy 13 (Patau syndrome)	**Negative**

Negative Predictive Value

The Negative Predictive Value (NPV) for trisomy 21, 18, and 13 is greater than 99%. The NPV for SCA and ESS cannot be calculated as SCA and ESS are only reported when an abnormality is detected.

About the Test

The MaterniT® 21 PLUS laboratory-developed test (LDT) analyzes circulating cell-free DNA from a maternal blood sample. The test is indicated for use in pregnant women with increased risk for fetal chromosomal aneuploidy. Validation data on twin pregnancies is limited and the ability of this test to detect aneuploidy in a triplet pregnancy has not yet been validated.

Test Method

Circulating cell-free DNA was purified from the plasma component of maternal blood. The extracted DNA was then converted into a genomic DNA library for aneuploidy analysis of chromosomes 21, 18, and 13 via next generation sequencing.[1] Optional findings based on the test order include sex chromosome aneuploidy (SCA)[2], and enhanced sequencing series (ESS)[3], which will only be reported on as an additional finding when an abnormality is detected. SCA testing includes information on X and Y representation, while ESS testing includes deletions in selected regions (22q, 15q, 11q, 8q, 5p, 4p, 1p) and trisomy of chromosomes 16 and 22.

Performance

The performance characteristics of the MaterniT® 21 PLUS laboratory-developed test (LDT) have been determined in a clinical validation study with pregnant women at increased risk for fetal chromosomal aneuploidy. [1],[2],[3],[4]

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Y-Chromosome (Fetal Sex)	**Accuracy: 99.4%**	
Region (associated syndrome)	**Estimated Sensitivity****	**Estimated Specificity**
Trisomy 21 (Down Syndrome)	99.1%	99.9%
Trisomy 18 (Edwards Syndrome)	>99.9%	99.6%
Trisomy 13 (Patau Syndrome)	91.7%	99.7%
Sex Chromosome Aneuploidies (singleton gestation only)	96.2%	99.7%

* As reported in ISCA database nstd37 - http://dbsearch.clinicalgenome.org/search/

** Sensitivity estimated across the observed size distribution of each syndrome [per ISCA database nstd37] and across the range of fetal fractions observed in routine clinical NIPT. Actual sensitivity can also be influenced by other factors such as the size of the event, total sequence counts, amplification bias, or sequence bias.

Limitations of the Test

While the results of these tests are highly accurate, discordant results, including inaccurate fetal sex prediction, may occur due to placental, maternal, or fetal mosaicism or neoplasm; vanishing twin; prior maternal organ transplant; or other causes. Sex chromosomal aneuploidies are not reportable for known multiple gestations. These tests are screening tests and not diagnostic; they do not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis. A patient with a positive test result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results.[5] A negative result does not ensure an unaffected pregnancy nor does it exclude the possibility of other chromosomal abnormalities or birth defects which are not a part of these tests. An uninformative result may be reported, the causes of which may include, but are not limited to, insufficient sequencing coverage, noise or artifacts in the region, amplification or sequencing bias, or insufficient fetal fraction. These tests are not intended to identify pregnancies at risk for neural tube defects or ventral wall defects. Testing for whole chromosome abnormalities (including sex chromosomes) and for subchromosomal abnormalities could lead to the potential discovery of both fetal and maternal genomic abnormalities that could have major, minor, or no, clinical significance. Evaluating the significance of a positive or a non-reportable result may involve both invasive testing and additional studies on the mother. Such investigations may lead to a diagnosis of maternal chromosomal or subchromosomal abnormalities, which on occasion may be associated with benign or malignant maternal neoplasms. These tests may not accurately identify fetal triploidy, balanced rearrangements, or the precise location of subchromosomal duplications or deletions; these may be detected by prenatal diagnosis with CVS or amniocentesis. The ability to report results may be impacted by maternal BMI, maternal weight, maternal systemic lupus erythematosus (SLE) and/or by certain pharmaceutical agents such as low molecular weight heparin (for example: Lovenox®, Xaparin®, Clexane® and Fragmin®). The results of this testing, including the benefits and limitations, should be discussed with a qualified healthcare provider. Pregnancy management decisions, including termination of the pregnancy, should not be based on the results of these tests alone. The healthcare provider is responsible for the use of this information in the management of their patient.

Note

This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration. This laboratory is certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing and accredited by the College of American Pathologists (CAP).
All previously submitted samples were used in generating the MaterniT® 21 PLUS result. If there is future clinical need for adding on MaterniT® GENOME testing, a redraw will need to be submitted at that time.

References

1. Palomaki GE, et al. *Genet Med*. 2012;14(3):296-305.
2. Mazloom AR, et al. *Prenat Diag*. 2013;33(6):591-597.
3. Zhao C, et al. *Clin Chem*. 2015 Apr;61(4):608-616.
4. Palomaki GE, et al. *Genet Med*. 2011;13(11):913-920.
5. ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

Eyad Almasri, MD, PhD
Director, Sequenom Laboratories
01/10/2020

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Thank you for selecting MaterniT® 21 PLUS for your cfDNA/NIPT screening needs.

All previously submitted samples were used in generating the MaterniT® 21 PLUS result. If there is future clinical need for adding on MaterniT® GENOME testing, a redraw will need to be submitted at that time.

Please keep this page with the patient's file and use it as your test request form to order re-sequencing as needed. (Fax#: 858.202.9108)

RE-SEQUENCING PATHWAY FROM MaterniT® 21 PLUS FOR SUSPECTED HIGH RISK SPECIMENS (LCA Test Code: 452114)

☐ Fetal anomaly suspected. Re-sequence using MaterniT® GENOME.

Medical Indication: _____

Referring Clinician: _____ Fax: (_____) _____ - _____

MaterniT® GENOME assay is not validated for multifetal gestations; multifetal samples are excluded from the resequencing pathway.

Contact Integrated Genetics Client Services at 877.821.7266 for any questions regarding re-sequencing, or to place your order.

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