



MaterniT® 21 PLUS (Core)
Singleton Gestation

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

Ordering Provider: Last, First
Provider Location: Sequenom SD
Provider Phone:
Date Ordered: mm/dd/year
Date Collected: mm/dd/year
Date Received: mm/dd/year
Order ID: xxx1234567
Patient ID: 1234567890
Patient: Last, First
DOB: mm/dd/year
Specimen: 1234567890
Fetal Fraction: xx%
Gestational Age ≥ 9w: Yes
External Accession:
Referral Clinician:
Date Reported: 04/01/2022 12:37 PM PT

Test Result

Positive
Trisomy 18

Lab Director Comments

This specimen showed an increased representation of chromosome 18, suggestive of high mosaic trisomy 18, which may affect the reported PPV (Rafalko et al., 2020). In placental testing, trisomy 18 is a common finding that is often confined to the placenta (CPM) (Grati, 2014). However, true fetal involvement is associated with phenotypic abnormality. Genetic counseling, confirmatory diagnostic testing, and clinical correlation are recommended.

Result Table

Table with 2 columns: Content, Result. Rows include FETAL SEX (Consistent with Male), AUTOSOMAL ANEUPLOIDIES, Trisomy 21 (Down syndrome) (Negative), Trisomy 18 (Edwards syndrome) (Positive T18 PPV\*: 74%), and Trisomy 13 (Patau syndrome) (Negative).

Positive Predictive Value

\* Positive Predictive Value (PPV) estimates the probability that a pregnancy with a positive test result is in fact an affected pregnancy. The PPV for this patient was calculated only using maternal age and gestational age[1], test performance[2] and the standard PPV formula.

For a more accurate and individualized PPV calculation, include additional clinical information from the patient's clinical history (which may include serum screen results, personal/family history, ultrasound findings, etc.), and refer to the table below.

Table with 17 columns: A Priori Risk (1:X) and PPV (%) TRISOMY 18. Rows show risk levels from 10 to 5000 and corresponding PPV percentages.

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. This test is used for screening purposes and not diagnostic. Clinical correlation is recommended. Validation data on twin pregnancies is limited and the ability of this test to detect aneuploidy in higher multiple gestations has not yet been validated.

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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 [3] Optional findings based on the test order include sex chromosome aneuploidy (SCA)[2], and enhanced sequencing series (ESS)[4], 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. [2-5]

Table with 3 columns: Region (associated syndrome), Accuracy, Estimated Sensitivity\*\*, and Estimated Specificity. Rows include Trisomy 21 (Down Syndrome), Trisomy 18 (Edwards Syndrome), Trisomy 13 (Patau Syndrome), and Sex Chromosome Aneuploidies (singleton gestation only).

\* As reported in ISCA database nstd37 [https://www.ncbi.nlm.nih.gov/dbvar/studies/nstd37/]

\*\* 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 reliable, 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. 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. [6] 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. Sex chromosome aneuploidies are not reportable for known multiple gestations. 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®, Clethane® and Fragmin®).

Note

Sequenom, Inc. is a subsidiary of Laboratory Corporation of America Holdings, using the brand Labcorp. 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).

References

- 1. Snijders RJ, et al. Fetal Diag. 1995;10(6):356-367.
2. Mazloom AR, et al. Prenat Diag. 2013;33(6):591-597.
3. Palomaki GE, et al. Genet Med. 2012;14(3):296-305.
4. Zhao C, et al. Clin Chem. 2015 Apr;61(4):608-616.
5. Palomaki GE, et al. Genet Med. 2011;13(11):913-920.
6. ACOG/SMFM Practice Bulletin No. 226, Oct 2020.

Latha Gowrishankar, MD, PhD
Director, Sequenom Laboratories
04/01/2022

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