MaterniT21 PLUS

見本

labcorp

NIPT結果報告書

被験者名	Patient, Name 様	医療機関名	Hospital Name
生年月日	YYYY/MM/DD	弊社検体番号	7777777
胎児数	2	担当医師名	Physician,Name
採血日	YYYY/MM/DD	登録施設番号	111111
病院被験者番号	12345		

検査結果

判定保留（Not Reportable）

技術的な問題、もしくは検体に関する問題により、検査データが精度管理基準を満たしませんでした。
再検査のために、再採血を行ってください。

胎児DNA率 N/A %

結果一覧

対象疾患	検査結果
21トリソミー （ダウン症候群）	判定保留
18トリソミー （エドワーズ症候群）	判定保留
13トリソミー （パトー症候群）	判定保留

【本検査について】
本検査では、母体血中に循環する胎児由来 Cell-free DNA の量を測定しています。胎児DNA率が3%未満の場合は原則として判定保留となります。

【判定保留（Not-Reportable）について】
結果の意義を確認するために、絨毛や羊水を用いた確定的検査や、母体の精査が必要になる場合があります。
また、母体の精査の結果として、母体自身の染色体変化、良性腫瘍や悪性腫瘍の診断につながる可能性があります。

【検査の限界と注意点】
本検査は正確な検査ではありますが、確定的検査に取って代わるものではありません。
「陽性」の検査結果が出た場合は、遺伝カウンセリングを受診し、検査結果を確認するための絨毛や羊水を用いた確定的検査を受けることを検討する必要があります。「陰性」の検査結果は、胎児が対象疾患に罹患していないことを保証するものではありません。また対象疾患以外の染色体異常（13/18/21番染色体の部分欠失・部分重複、13/18/21番以外の染色体の数的異常や部分欠失・部分重複など）や、染色体異常以外の原因による先天異常の可能性を否定するものでもありません。妊娠管理の方針は本検査結果だけではなく、その他の臨床情報を踏まえて総合的にご検討ください。

Hany Magharyous
Director, Sequenom Laboratories

病院使用欄

上記の米国人氏名は米国ラボコープ社の検査所 Sequenom Laboratoriesの検査責任者名です。当責任者の下、確かに検査が終了したことを示しています。
当書面は米国ラボコープ社での検査結果を元にラボコープ・ジャパンが作成しています。

ラボコープ・ジャパン 〒104-0045 東京都中央区築地 2-11-9 RBM 築地駅前ビル 2 階 TEL:03-6226-0900 / FAX:03-6226-0913

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FINAL REPORT

MaterniT® 21 PLUS (Core)
Twin Gestation

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

877.821.7266

Ordering Provider:	**Physician, Name**	Patient:	**Patient, Name**
Provider Location:	**Hospital Name**	DOB:	**MM/DD/YYYY**
Provider Phone:	**5555555555**	Specimen:	**2222901066**
Date Ordered:	**MM/DD/YYYY**	Fetal Fraction:	**N/A**
Date Collected:	**MM/DD/YYYY**	Gestational Age ≥ 9w:	**Yes**
Date Received:	**MM/DD/YYYY**	External Accession:	**26395612SEQCA**
Order ID:	**ORD22229-02065**	Referral Clinician:	
Patient ID:	**23164206/12345**	Date Reported:	**MM/DD/YYYY 10:38 AM**

Test Result	**Not Reportable**

Lab Director Comments

Testing for this sample was performed. Due to technical or sample-related issues, data failed to meet quality standards for interpretation.

Please submit another specimen for testing.

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.

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. [1-4]

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 [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.

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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 .[5] 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 chromosomal 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®, Clexane® 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. 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 Practice Bulletin No. 226, Oct 2020.

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