MaterniT21 PLUS

見本

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# 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 | 1 | 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 | 陽性 (Positive) |
|---|---|
| この検体では、18トリソミー（エドワーズ症候群）の胎児に見られるような18番染色体の量の異常が認められました。 | |

胎児DNA率　14 %

## 結果一覧

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

【陰性的中率について】
この検査における13、18、21トリソミーの陰性的中率は99%以上です。

【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。

【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)
## 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**

| | | | |
|---|---|---|---|
| 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: | **14%** |
| Date Collected: | **MM/DD/YYYY** | Gestational Age ≥ 9w: | **Yes** |
| Date Received: | **MM/DD/YYYY** | External Accession: | **26395612SEQCA** |
| Order ID: | **ORD22334-02958** | Referral Clinician: | |
| Patient ID: | **23164206/12345** | Date Reported: | **MM/DD/YYYY 10:38 AM** |

## Test Result

# Positive

**Trisomy 18**

### Lab Director Comments

This specimen showed an increased representation of chromosome 18, suggestive of trisomy 18 (Edwards syndrome). Genetic counseling, confirmatory diagnostic testing, and clinical correlation are recommended.

### Result Table

| Content | Result |
|---|---|
| **FETAL SEX** | **Opt-Out** |
| **AUTOSOMAL ANEUPLOIDIES** | |
| **Trisomy 21 (Down syndrome)** | **Negative** |
| **Trisomy 18 (Edwards syndrome)** | **Positive T18 PPV*: 61.6%** |
| **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.

| *A Priori* Risk (1:X) | 10 | 20 | 30 | 40 | 50 | 100 | 200 | 300 | 400 | 500 | 1000 | 1500 | 2000 | 2500 | 3000 | 5000 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| PPV (%) TRISOMY 18 | 96.5 | 92.9 | 89.6 | 86.5 | 83.6 | 71.6 | 55.7 | 45.5 | 38.5 | 33.4 | 20.0 | 14.3 | 11.1 | 9.1 | 7.7 | 4.8 |

### 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]

| **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.

### 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 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. 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.

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*12/03/2022*