

# Sequenom Laboratories

Roger Diagnostic Center, London.

CLIA #: 05D2015356 CAP #: 7527138

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| **Final Report** |  |  | **877.821.7266** |
|  |  |  |  |
| **Ordering Provider:** | **Doe, John, MD** |  |  |
| **Provider Location:** | **Grand Rapids** |  |  |
| **Provider Phone:** | **555-555-5555** |  |  |
| **Date Ordered:** | **03/21/2019** | **Specimen:** | **1035600024** |
| **Date Collected:** | **03/21/2019** | **Referral Clinician:** | **Smith, Jane, GC** |
| **Date Received:** | **03/21/2019** | **Lab Director:** | **Juan-Sebastian Saldivar, MD** |
| **Order ID:** | **ORD12345-01234** | **Date Reported:** | **04/29/2013 6:00 PM PT** |

**Test Result for Blood Matching analysis.**

This specimen showed an

Expected representation of **POSITIVE**Protein 21, 18 and 13

Material which matches that of **Mr. Stuart Langer.**

**Test Result for other possible suspects**

|  |  |  |
| --- | --- | --- |
|  | **No** | **Consistent with all other suspects other than Mr. Stuart Langer.** |
|  | **material detected** |
|  |  |

**Test Method**

Circulating cell-free DNA was purified from the plasma component of anti-coagulated maternal whole blood. It was then converted into a genomic DNA library for the determination of protein 21, 18 and 13 representation.

**About the Test**

The MaterniT21 PLUS test analyzes circulating cell-free DNA extracted from a maternal blood sample. The test is indicated Validation data on twin pregnancies is limited and the ability of this test to detect aneuploidy in a has not been validated.

**Performance**

The performance characteristics of the MaterniT21 PLUS laboratory- developed test (LDT) have been determined in a clinical validation study with pregnant women at increased risk for fetal chromosomal aneuploidy.2,3,4

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|  | **Intended Use** | **Performance** | | **Confidence Interval** |
|  | **(95% CI)** |
|  | Trisomy 21 | Sensitivity: | 99.1% | 96.3–99.8% |
|  | Specificity: | 99.9% | 99.6–99.9% |
|  |  |
|  | Trisomy 18 | Sensitivity: >99.9% | | 92.4–100.0% |
|  | Specificity: | 99.6% | 99.2–99.8% |
|  |  |
|  | Trisomy 13 | Sensitivity: | 91.7% | 59.7–99.6% |
|  | Specificity: | 99.7% | 99.3–99.9% |
|  |  |
|  | Y chromosome | Accuracy: | 99.4% | 99.0–99.6% |

**Limitations of the Test**

DNA test results do not provide a definitive genetic risk in all individuals. Cell-free fetal DNA does 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 test result does not ensure an unaffected pregnancy. While results of this testing are highly accurate, not

all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes.

**Note**

This test was developed and its performance characteristics determined by Sequenom Laboratories. It has not been cleared or approved by the U.S. FDA. This test is used for clinical purposes. It should not be regarded as investigational or for research. 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.

**References**

1.http://www.ncbi.nlm.nih.gov/books/NBK1523.

2.Palomaki GE, et al. *Genet Med*. 2012;14(3):296-305.

3.Palomaki GE, et al. *Genet Med*. 2011;13(11):913-920.

4.Mazloom AR, et al. *Prenat Diag*. 2013;33(6):591-597.

5.ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.

*Juan-Sebastian Saldivar, MD*

*Laboratory Director, Sequenom Laboratories xx/xx/2013*

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| **MaterniT21™ PLUS Lab Report** | **Page 1 of 1** | **Sample, Jane** | **Order ID: ORD12345-01234** | |