Date Collected: **02/19/2024** Date Received: **02/19/2024** Date Reported: **02/24/2024** Fasting: **No**

Ordered Items: **MaterniT21 PLUS Core+SCA; Venipuncture**

Date Collected: **02/19/2024**

MaterniT21 PLUS Core+SCA

	Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
	Gestation [01]	Singleton			
	Fetal Fraction [01]	6%			
	Gestational Age > or = 9w: [01]	Yes			
▶	**Test Result** [01]	**Positive** **Abnormal** Trisomy 18			
	Lab Director Comments [01]	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.			
	Approved By [01]	Juan-Sebastian Saldivar, MD, Director, Sequenom Laboratories			
	Trisomy 21 (Down Syndrome) [01]	Negative			
▶	**Trisomy 18 (Edwards Syndrome)** [01]	**See below:** **Abnormal** Positive T18 PPV*: 83.6%			
	Trisomy 13 (Patau Syndrome) [01]	Negative			
	Fetal Sex [01]	Consistent with Male			
	Monosomy X (Turner Syndrome) [01]	Not Detected			
	XYY (Jacobs Syndrome) [01]	Not Detected			
	XXY (Klinefelter Syndrome) [01]	Not Detected			
	XXX (Triple X Syndrome) [01]	Not Detected			
	Negative Predictive Value [01]	Note 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.			
	Positive Predictive Value [01]	Note * 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,			

MaterniT21 PLUS Core+SCA (Cont.)

personal/family history, ultrasound findings, etc.), and refer to the table below.
A Priori Risk (1:10); PPV (96.5%) TRISOMY 18
A Priori Risk (1:20); PPV (92.9%) TRISOMY 18
A Priori Risk (1:30); PPV (89.6%) TRISOMY 18
A Priori Risk (1:40); PPV (86.5%) TRISOMY 18
A Priori Risk (1:50); PPV (83.6%) TRISOMY 18
A Priori Risk (1:100); PPV (71.6%) TRISOMY 18
A Priori Risk (1:200); PPV (55.7%) TRISOMY 18
A Priori Risk (1:300); PPV (45.5%) TRISOMY 18
A Priori Risk (1:400); PPV (38.5%) TRISOMY 18
A Priori Risk (1:500); PPV (33.4%) TRISOMY 18
A Priori Risk (1:1000); PPV (20.0%) TRISOMY 18
A Priori Risk (1:1500); PPV (14.3%) TRISOMY 18
A Priori Risk (1:2000); PPV (11.1%) TRISOMY 18
A Priori Risk (1:2500); PPV (9.1%) TRISOMY 18
A Priori Risk (1:3000); PPV (7.7%) TRISOMY 18
A Priori Risk (1:5000); PPV (4.8%) TRISOMY 18

About the Test [01]

The MaterniT(R) 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 [01]

See Notes
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 [01]

The performance characteristics of the MaterniT(R) 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]

Performance Characteristics [01]

Note

Fetal Sex	Accuracy: 99.4%	
Region (associated syndrome)	Est. Sens#	Est. Spec
Trisomy 21 (Down Syndrome)	99.1%	99.9%
Trisomy 18 (Edwards Syndrome)	>99.9%	99.6%

MaterniT21 PLUS Core+SCA (Cont.)

Trisomy 13 (Patau Syndrome)	91.7%	99.7%
Sex Chromosome Aneuploidies##	96.2%	99.7%

* As reported in ISCA database nstd37 [https://www.ncbi.nlm.nih.gov/dbvar/studies/nstd37/]
# Estimated Sensitivity. 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.
Singleton gestation only.

Limitations of the Test[01]

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

MaterniT21 PLUS Core+SCA (Cont.)

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(R), Xaparin(R), Clexane(R) and Fragmin(R)).

Note [01]

See Notes
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 [01]

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.

PDF [01]

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Disclaimer
The Previous Result is listed for the most recent test performed by Labcorp in the past 5 years where there is sufficient patient demographic data to match the result to the patient. Results from certain tests are excluded from the Previous Result display.

Icon Legend
▲ Out of Reference Range ■ Critical or Alert

Performing Labs
01: SEQCA - Sequenom Ctr for Molecular Med, 3595 John Hopkins Court, San Diego, CA 92121-1121 Dir: Phillip Cacheris, Dir
For Inquiries, the physician may contact Branch: 800-859-0391 Lab: 800-762-4344

Patient Details
Lindsay, Nadine T
5230 TUCKERMAN LN APT 1423, ROCKVILLE, MD, 20852

Phone: **443-370-6503**
Date of Birth: **09/06/1978**
Age: **45**
Sex: **Female**
Patient ID: **GUH FIN-790070023**
Alternate Patient ID: **006361507**

Physician Details
O MIMS JR
Georgetown Univ-Phc OB GYN
2233 Wisconsin Avenue Northwest, Washington, DC, 20007

Phone: **202-481-1060**
Account Number: **08747782**
Physician ID: **1326019589**
NPI: **1326019589**

Specimen Details
Specimen ID: **050-530-0396-0**
Control ID: **236030961**
Alternate Control Number: **236030961**
Date Collected: **02/19/2024 1603 Local**
Date Received: **02/19/2024 0000 ET**
Date Entered: **02/19/2024 1641 ET**
Date Reported: **02/24/2024 0006 ET**

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) + SCA
Singleton Gestation

Ordering Provider:	**MIMS JR, O**	Patient:	**LINDSAY, NADINE**
Provider Location:	**Georgetown Univ-Phc OB GYN**	DOB:	**09/06/1978**
Provider Phone:	**202-481-1060**	Specimen:	**2405101824**
Date Ordered:	**02/20/2024**	Fetal Fraction:	**6%**
Date Collected:	**02/19/2024**	Gestational Age ≥ 9w:	**Yes**
Date Received:	**02/21/2024**	External Accession:	**2405053003960**
Order ID:	**ORD24051-02821**	Referral Clinician:	
Patient ID:	**Not Available**	Date Reported:	**02/23/2024 02:54 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

Content	Result
FETAL SEX	**Consistent with Male**
AUTOSOMAL ANEUPLOIDIES	
Trisomy 21 (Down syndrome)	**Negative**
Trisomy 18 (Edwards syndrome)	**Positive T18 PPV*: 83.6%**
Trisomy 13 (Patau syndrome)	**Negative**
SEX CHROMOSOME ANEUPLOIDIES	
Monosomy X (Turner syndrome)	**Not Detected**
XYY (Jacob's syndrome)	**Not Detected**
XXY (Klinefelter syndrome)	**Not Detected**
XXX (Triple X syndrome)	**Not Detected**

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

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Test Method
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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%

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References

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6. ACOG/SMFM Practice Bulletin No. 226, Oct 2020.

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02/23/2024