MaterniT21® PLUS Lab Report

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Final Report

Ordering Provider:	**XXXX, XXXX**	Patient:	**XXXX, XXXX**
Provider Location:	**XXXXX**	DOB:	**XX/XX/XXXX**
Provider Phone:	**555-555-5555**	Patient ID:	**12345-01234**
Date Ordered:	**XX/XX/XXXX**	Specimen:	**0123456789**
Date Collected:	**XX/XX/XXXX**	Referral Clinician:	**XXXX, XXXX**
Date Received:	**XX/XX/XXXX**	Lab Director:	**XXXX, XXXX**
Order ID:	**12345-01234**	Date Reported:	**XX/XX/XX 00:00 PM PT**

SEE ADDITIONAL FINDINGS

Test Result for Chromosomes 21, 18 and 13

Negative	This specimen showed an expected representation of chromosome 21, 18 and 13 material. Clinical correlation is suggested.

Test Result for Y Chromosome

Y chromosome material detected	Consistent with a male fetus.

ADDITIONAL FINDINGS

This specimen showed a decreased representation of chromosome 4p	These findings are suggestive of a 4p deletion, affecting the 4p16.3 region associated with Wolf-Hirschhorn syndrome.

Wolf-Hirschhorn syndrome (4p minus) is caused by a deletion on the short arm of chromosome 4. The disorder is characterized by distinctive craniofacial anomalies, growth restriction, developmental delay, hearing loss and seizures. Incidence is estimated to be ~1/50,000 births. Most cases are not inherited and represent a *de novo* deletion.[1]

Lab Director Comments

Fetal Fraction: 12%

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 chromosome 21, 18, 13 representation and the presence of the Y chromosome.[2] Other chromosomal material, including fetal chromosome 22, 16, sex chromosome (X and Y) representation, and select regions (22q, 15q, 11q, 8q, 5p, 4p, 1p), was also evaluated and will only be reported as an Additional Finding when an abnormality is detected.

About the Test

The Materni1 PLUS test analyzes circulating cell-free DNA extracted from a maternal blood sample. The test is indicated for use in pregnant women with increased risk for chromosomal aneuploidy. Validation data on twin pregnancies is limited and the ability of this test to detect aneuploidy in a triplet pregnancy has not yet 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 abnormality.[2,3,4]

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 DNA does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis.

A patient with a positive test result or presence of an Additional Finding 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. The absence of an Additional Finding does not indicate a negative result. 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. Sex chromosomal aneuploidies are not reportable for known multiple gestations. The health care provider is responsible for the use of this information in the management of their patient.

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/NBK1183.
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.

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Laboratory Director, Sequenom Laboratories

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