ABOUT THE COMPANY

Sequenom Laboratories, a wholly owned subsidiary of Sequenom, Inc., is a CAP-accredited and Clinical Laboratory Improvement Amendment (CLIA)-certified molecular diagnostics laboratory dedicated to improving patient outcomes by offering revolutionary laboratory-developed tests for a variety of prenatal conditions. Sequenom Laboratories pioneered NIPT with the launch of its MaterniT21 PLUS test for fetal abnormalities, and offers a broad menu of prenatal tests.

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ABOUT THE TEST

The MaterniT GENOME test is a laboratory-developed test that was developed, validated and performed exclusively by Sequenom Laboratories. The test has not been cleared or approved by the US Food and Drug Administration (FDA). Although laboratory-developed tests to date have not been subject to US FDA regulation, certification of the laboratory is required under the Clinical Laboratory Improvement Amendments (CLIA) to ensure the quality and validity of the test. Sequenom Laboratories is certified under CLIA as qualified to perform high complexity clinical laboratory testing and is accredited by the College of American Pathologists.

No test is perfect. While the results of the MaterniT GENOME test are highly accurate, 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. Cell-free DNA (cfDNA) testing does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis. A patient with a positive MaterniT GENOME test result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. A negative MaterniT GENOME test result does not ensure an unaffected pregnancy. 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. The MaterniT GENOME test is not intended to identify pregnancies at risk for neural tube defects or ventral wall defects. cfDNA 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 minor, or no, clinical significance. Evaluating the significance of a positive or a non-reportable test result may involve both invasive prenatal testing and additional studies on the mother. Such investigations may lead to detection of maternal chromosomal or subchromosomal abnormalities, which on occasion may be associated with benign or malignant maternal neoplasms. cfDNA testing 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 body mass index (BMI), maternal weight, and/or maternal systemic lupus erythematosus (SLE). The results of this testing, including the benefits and limitations, should be discussed with a qualified health care provider. Pregnancy management decisions, including termination of the pregnancy, should not be based on the results of this test alone.

REFERENCES

1. Di Gregorio E, et al. Large cryptic genomic rearrangements with apparently normal karyotypes detected by array-CGH. *Mol Cytogenet.* 2014;7(82).
2. Zhao C, et al. Detection of fetal subchromosomal abnormalities by sequencing circulating cell-free DNA from maternal plasma. *Clin Chem.* 2015 Apr;61(4):608-616.

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THE SCIENCE OF

UNPARALLELED INSIGHT

MaterniT™ GENOME

Noninvasive prenatal test for genome-wide fetal chromosomal abnormalities

sequenom Laboratories

QUALITY OF SCIENCE™

YOUR GOAL AND OURS: TO DELIVER THE MOST ADVANCED CARE POSSIBLE TO EVERY PATIENT.

To help you meet that goal: the MaterniT™ GENOME laboratory-developed test, a new noninvasive prenatal test (NIPT) that analyzes every chromosome in the genome. Now, for the first time ever, there is an NIPT to provide you with this unparalleled insight, enabling a new level of patient care.

The MaterniT GENOME test is the only genome-wide NIPT that provides karyotype-level insight.

MaterniT™ GENOME

GAIN MORE INSIGHT THAN EVER BEFORE

The MaterniT GENOME test starts with the ease of an ordinary blood draw, taken as early as 10 weeks gestation. Though not a fetal karyotype, it is currently the only NIPT that offers a level of information previously only available from a karyotype analysis. It provides information about gains or losses of chromosome material ≥ 7 Mb across the genome. The test also analyzes seven clinically significant microdeletion regions. By combining increased sequencing depth with industry-leading expertise, MaterniT GENOME offers a breadth of coverage unlike any other noninvasive prenatal test available to date.

COMPLICATED SCIENCE. UNCOMPLICATED REPORTING.

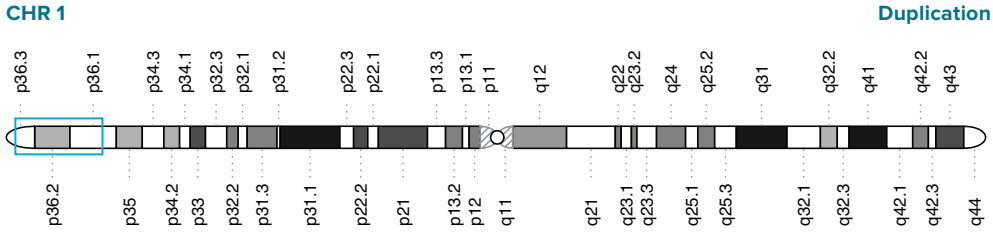
The MaterniT GENOME test delivers sophisticated DNA analysis in straightforward terms. The simple reporting style is designed to facilitate communication between you and your patient.

Each chromosome target receives a distinct result of Positive or Negative

CONTENT	RESULT
AUTOSOMAL ANEUPLOIDIES	
Trisomy 21 (Down syndrome)	Negative
Trisomy 18 (Edwards syndrome)	Negative
Trisomy 13 (Patau syndrome)	Negative
Other autosomal aneuploidies	Negative
SEX CHROMOSOME ANEUPLOIDIES	
Fetal sex	Consistent with female
Monosomy X (Turner syndrome)	Negative
XYY (Jacobs syndrome)	Negative
XXY (Klinefelter syndrome)	Negative
XXX (Triple X syndrome)	Negative
GENOME-WIDE COPY NUMBER VARIANTS ≥ 7 Mb	
Gains/Losses ≥ 7 Mb	Positive

CONTENT	RESULT
SELECT MICRODELETIONS	
22q11 deletion (associated with DiGeorge syndrome)	Negative
15q11 deletion (associated with Prader-Willi / Angelman syndrome)	Negative
11q23 deletion (associated with Jacobsen syndrome)	Negative
8q24 deletion (associated with Langer-Giedion syndrome)	Negative
5p15 deletion (associated with Cri-du-chat syndrome)	Negative
4p16 deletion (associated with Wolf-Hirschhorn syndrome)	Negative
1p36 deletion syndrome	Negative

The report features a chromosome ideogram, which illustrates abnormal results to facilitate comprehension



Description: An approximate 15.3 Mb gain of chromosome 1 material was observed, suggestive of a duplication in the region p36.3-p36.1.

SEE BEYOND PRENATAL KARYOTYPE

Resolution of prenatal karyotype is often poorer than peripheral blood karyotype. Cryptic deletions or duplications larger than 7 Mb can go undetected by routine prenatal karyotype[1], the clinical consequences of which can be lethal or lead to complex, severe fetal anomalies. The MaterniT GENOME test offers a novel capability of noninvasive testing, identifying > 95% of genome-wide deletions or duplications ≥ 7 Mb. This enables the most comprehensive fetal chromosomal test currently available noninvasively.

We understand that advanced genetic reporting can introduce challenges to patient counseling, especially when you need more context for a particular result. You can rely on peer-to-peer consultations with our expert genetic counselors to provide the deeper level of insight you need.

For questions about the MaterniT GENOME test, or to speak to a genetic counselor about results, contact us at genome@sequenom.com.

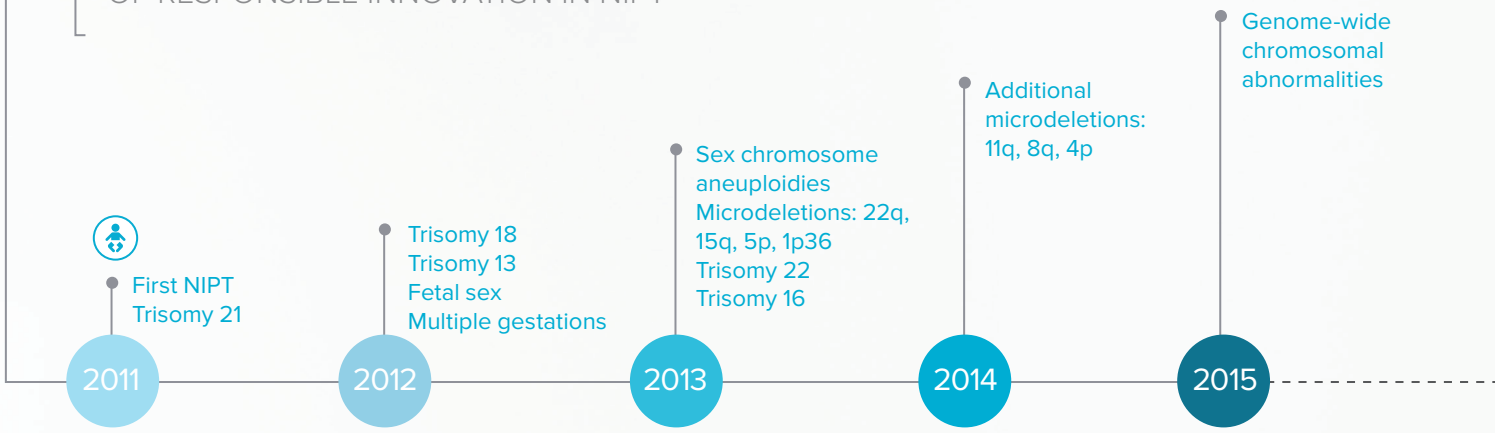
	Fetal karyotype test	MaterniT GENOME test
Analyzes every chromosome	Yes	Yes
Requires an invasive procedure	Yes	No
Detects large, unbalanced translocations	Yes	Yes
Detects marker chromosomes	Yes	Yes
Detects balanced translocations or inversions	Yes	No
Detects chromosome gains or losses as small 7 Mb	No	Yes
Detects select microdeletions	No	Yes
Detects triploidy	Yes	No
Considered diagnostic	Yes	No

REMAIN AT THE FOREFRONT OF YOUR FIELD

The MaterniT GENOME test couples leading technology with unparalleled insight to offer the most scientifically advanced information available from a noninvasive test. It is a cutting-edge tool that complements your expertise, allowing you to enhance your practice and advance your patient management.

MateriT™ GENOME

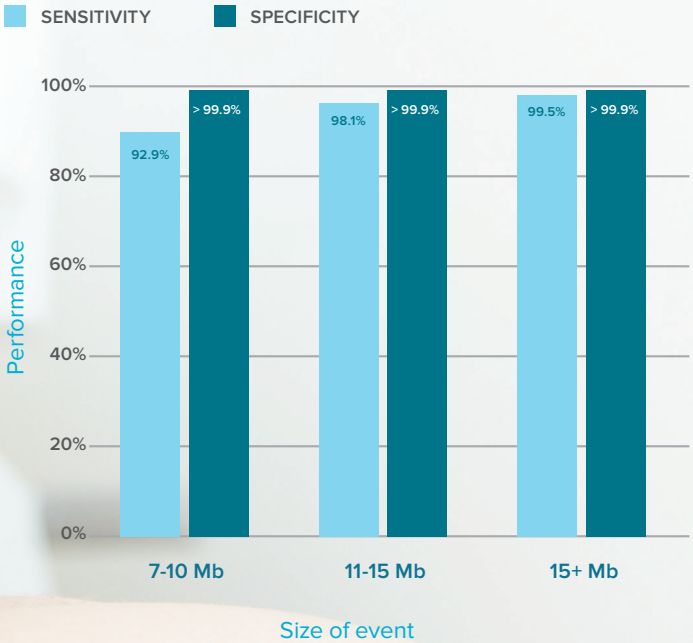
SEQUENOM LABORATORIES' HISTORY OF RESPONSIBLE INNOVATION IN NIPT



PERFORMANCE YOU CAN RELY ON

Sequenom Laboratories has a history of responsible innovation, with each new advancement in NIPT characterized by reliable results and supported by extensive validation studies. Validation testing of the MateriT GENOME test builds on this history, augmenting earlier work in genome-wide analysis, to ensure highly accurate results.

GENOME-WIDE PERFORMANCE



PERFORMANCE EVALUATION

GENOME-WIDE SUBCHROMOSOMAL EVENTS

Initial genome-wide study
- Prior work[2] was first foray into genome-wide analysis
- Used genomic DNA, maternal plasma mixtures
- Titration series to mimic fetal fraction distribution

Extensive modeling with ISCA data
- Used ISCA database as source for >2000 events
- *in silico* modeling of these events in >1000 maternal plasma sequences
- Enabled over 2 million analyses
- Aided understanding of algorithm performance

Analytical validation
- 4x more gDNA samples analyzed than prior work
- Samples with microarray or karyotype confirmed events
- Titration series to mimic fetal fraction distribution
- Measured performance confirmed *in silico* modeling

TRISOMIES, 21, 18, 13, SCA, AND FETAL SEX

Equivalency study
- Study of 448 clinical samples
- 2x sequencing depth than MateriT21® PLUS
- Compared MateriT21 PLUS results to MateriT GENOME results for trisomies 21, 18, 13, sex chromosome aneuploidies, fetal sex classification
- 100% concordance between the two tests