MaterniT® GENOME

A fully validated genome-wide NIPT

Integrated GENETICS
LabCorp Specialty Testing Group

Why it's different

By combining increased sequencing depth with industry-leading expertise, the MaterniT® GENOME test offers a breadth of coverage unlike any other noninvasive prenatal test available to date.

After more than 20,000 tests resulted by Sequenom Laboratories, up to 30% of all positive findings could only be detected by MaterniT GENOME methodology.[1]

Because most other NIPTs don't analyze for that 30%, they don't report on it. But that doesn't mean there's nothing to report.

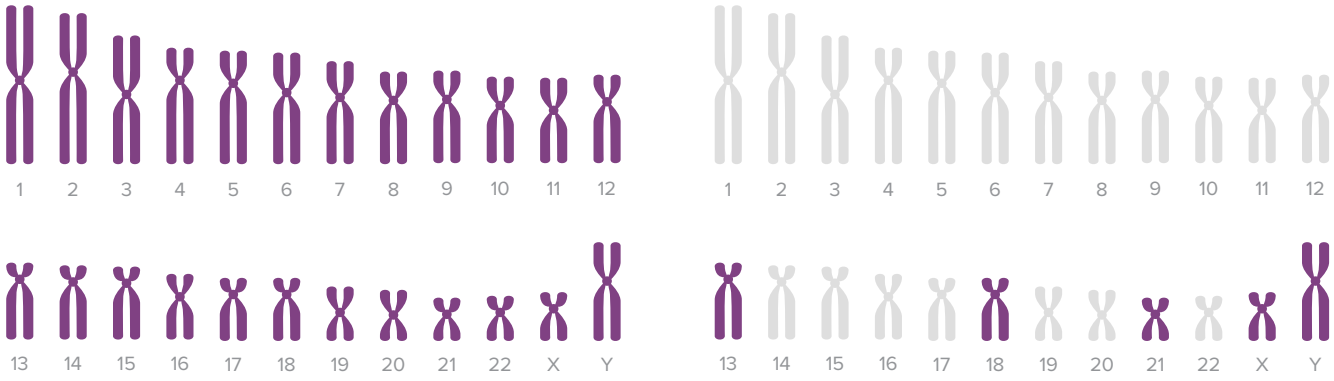
REPORTS ON DELETIONS/DUPLICATIONS ≥ 7 MB… AND CLINICALLY RELEVANT MICRODELETIONS < 7 MB[2]

Like most NIPTs, MaterniT GENOME starts with the ease of an ordinary blood draw, taken as early as nine weeks gestation.

It screens for common trisomies (such as 21, 18, and 13), sex chromosome aneuploidies, and analyzes seven clinically significant microdeletion regions.

It also analyzes every chromosome and can provide information about clinically relevant microdeletions and gains or losses of chromosome material ≥ 7 Mb across the entire genome—something other validated NIPTs do not currently do.

Whole chromosome analysis



Its capacity to analyze chromosomal material genome-wide makes MaterniT GENOME an ideal fit for high-risk cases where a patient may wish to avoid a diagnostic procedure, or where screening for common aneuploidies may not be enough. Recent MaterniT GENOME case studies present findings not detectable by conventional NIPT (ask a Sequenom Laboratories representative for details, or visit sequenom.com/company/clinical-updates).

Though not a fetal karyotype, MaterniT GENOME offers a level of information that previously was only available from a karyotype analysis.

In fact, cryptic deletions or duplications larger than 7 Mb can sometimes go undetected by routine prenatal karyotype.[3] The clinical consequences of this can lead to complex, severe fetal anomalies. Fortunately, models of available abnormal cases show that MaterniT GENOME can identify > 95% of genome-wide deletions or duplications ≥ 7 Mb.[4] This enables a comprehensive fetal chromosomal screen noninvasively.

A HIGHER STANDARD FOR DIGEORGE RESULTING

The 22q microdeletion is associated with DiGeorge syndrome, which, according to the US National Library of Medicine, impacts one in 4,000 pregnancies.[5]

With a reportable fetal fraction threshold of ≥ 4%, sensitivity of 74%, and specificity of 99.9% for 22q11.2 microdeletions,[6] MaterniT GENOME sets a higher standard in reporting for this critical chromosomal abnormality.

VALIDATED PERFORMANCE, STRAIGHTFORWARD REPORTING

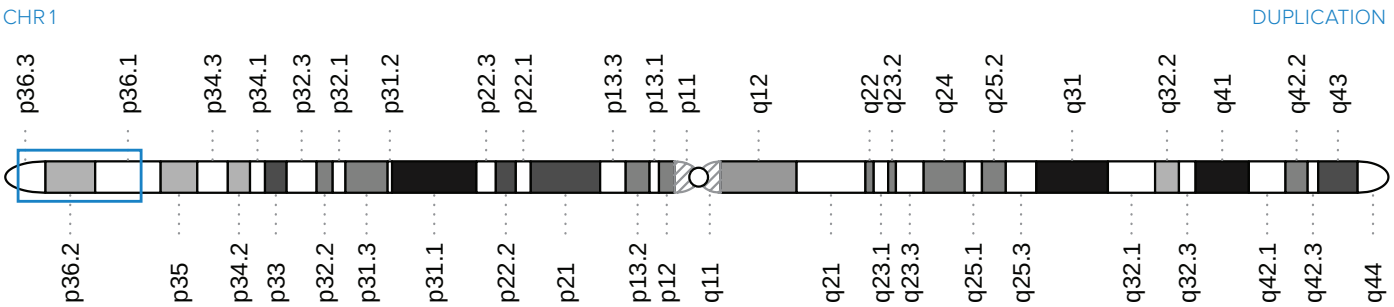
Sequenom and Integrated Genetics have a history of innovation, with each new advancement in NIPT characterized by reliable results and supported by extensive validation studies.

Validation testing of MaterniT GENOME built on this history, augmenting earlier work in genome-wide analysis, to ensure highly accurate results. (Visit sequenom.com/company/clinical-updates)

And the MaterniT GENOME test delivers sophisticated DNA analysis in straightforward terms. The reporting style (see example chromosome ideogram below) is designed to facilitate communication between you and your patient.

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 w/ 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
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

Each chromosome target receives a distinct result of *Positive* or *Negative*



The report features a chromosome ideogram, which illustrates abnormal results to facilitate comprehension. In this example, we see an approximate 15.3 Mb gain of chromosome 1 material, suggestive of a duplication in the region of p36.3-p36.1.

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View short videos on genetic testing:
sequenom.com/videos

Test name	MateriT GENOME
Test number	451941

Integrated GENETICS
LabCorp Specialty Testing Group

Professional services complement our pioneering science...

RAPID RESULTS

Overnight shipping of samples is available with a typical turnaround time of about five days after a test arrives at our labs.

COST ESTIMATOR

Send any patient with billing questions to our online cost estimator for answers and an estimate based on her specific insurance.

CONVENIENT BLOOD DRAWS

Blood draws just got easier for you and your patients. We are now part of LabCorp and have a nationwide network of patient service centers, allowing for convenient access to sample collection.
Visit www.LabCorp.com to find your nearest location.

GENETIC COUNSELING

Patients with a positive test result may be offered counseling, and Sequenom and Integrated Genetics offer the largest national commercial network of genetic counselors to help inform and support patients.

EVERY MOM PLEDGE

We believe every mom should have access to the best possible care. That's why we work directly with every patient to make sure our testing services are both accessible and affordable, no matter what.

REFERENCES

1. Internal data.
2. Lefkowitz RB, Tynan JA, Liu T, et al. Clinical validation of a noninvasive prenatal test for genomewide detection of fetal copy number variants. *Am J Obstet Gynecol* 2016;215:227.e1-16.
3. Di Gregorio E, et al. Large cryptic genomic rearrangements with apparently normal karyotypes detected by array-*CGH. Mol Cytogenet*. 2014;7(82).
4. Norton ME, Baer RJ, Wapner RJ, et al. Cell-free DNA vs sequential screening for the detection of fetal chromosomal abnormalities. *Am J Obstet Gynecol* 2016;214:727.e1-6.
5. "22q11.2 Deletion Syndrome." Genetics Home Reference. *US National Library of Medicine*, 6 Dec. 2016. Web. 8 Dec. 2016.
6. Internal data. Sensitivity estimated across the observed size distribution of DiGeorge syndrome [per ISCA database nstd37] and across the range of fetal fractions observed in routine clinical NIPT.

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