MaterniT®

GENOME

Why it's different

MaterniT® Genome

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.

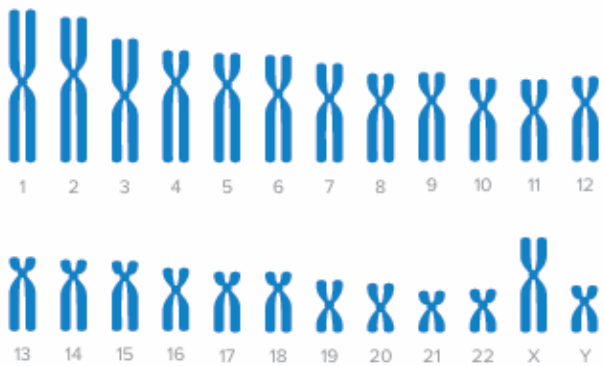
After more than 20,000 tests resulted by Integrated Genetics, up to 30% of all positive findings could only be detected by MaterniT GENOME methodology. Because most other NIPTs don't analyze for that 30%, they don't report on it. But that doesn't mean there's nothing to report.

REPORTS ON DELETIONS/DUPLICATIONS ⩾ 7 MB... ANDCLINICALLY RELEVANT MICRODELETIONS < 7 MB2

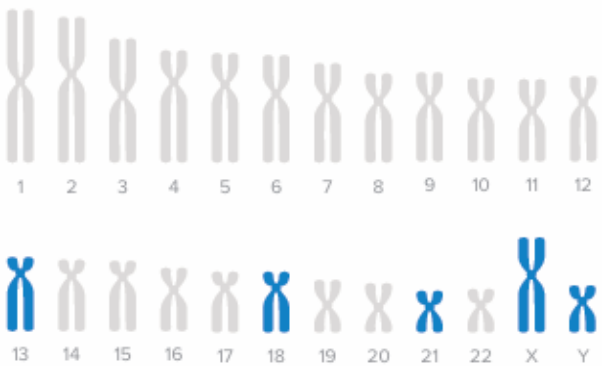
Like most NIPTs, MaterniT GENOME starts with the ease of an ordinary blood draw, taken as early as nine weeks gestation. It screens for common trisomies (such as21,18,and13), sex chromosome aneuploidies, and analyzes seven clinically significant microdeletion regions.

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—somethingother validated NIPTs do not currently do.

Whole chromosome analysis



MaterniT GENOME



Other NIPTs

Its capacity to analyze chromosomal material genome-wide makes MateriniT 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 MateriniT GENOME case studies present findings not detectable by conventional NIPT (ask a LabCorp/Integrated Genetics representative for details, or visitintegratedgenetics.com/providers/tests/prenatal/nipt/materniT genome).

Though not a fetal karyotype, MateriniT 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.

The clinical consequences of this can lead to complex, severe fetal anomalies. Fortunately, models of available abnormal cases show that MateriniT GENOME can identify > 95% of genome-wide deletions or duplications ≥ 7 Mb.4 This enables a comprehensive fetal chromosomal screen noninvasively.

AHIGHER 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.

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

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*

VALIDATED PERFORMANCE, STRAIGHTFORWARD REPORTING

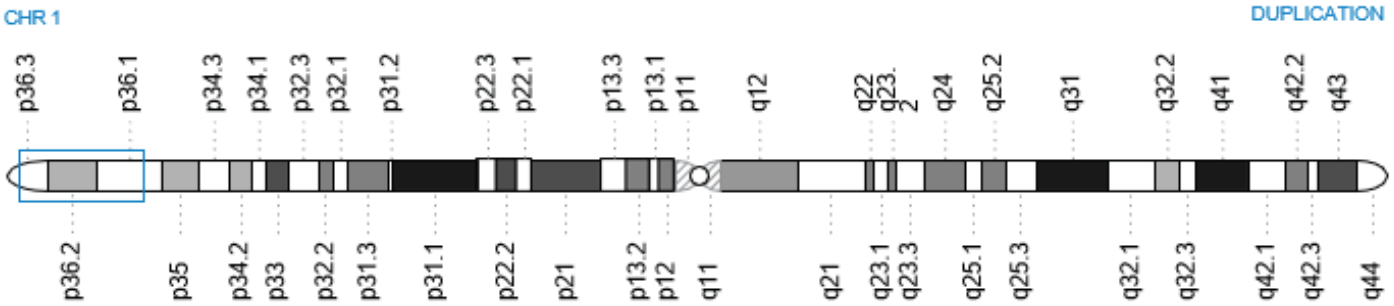
Integrated Genetics has a history of innovation, with each new advancement in NIPT characterized by reliable results and supported by extensive validation studies.

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.

What will MaterniT GENOME tell me?

Like most noninvasive prenatal screenings (NIPSs/NIPTs), MaterniT GENOME can tell you if you screen positive or negative for trisomies 21 (Down syndrome), 18 (Edwards syndrome), and 13 (Patau syndrome), and if you're having a boy or a girl. But it can also find other chromosomal changes that may go undiagnosed at birth. Having information about these chromosomal changes before birth can help ensure your baby receives the proper and necessary support.



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 1material, suggestive of a duplication in the region of p36.3-p36.1.