

CLINICAL AND  
HUMAN TESTING



READING NATURE

sequenom

## A PARTNERSHIP FOR SIMPLE AND ACCESSIBLE PRENATAL CARE

Test which fungicide is more effective for  
fungal control and eradication

## DISCOVER THE PREMIUM PRENATAL TEST THAT TAKES PRENATAL TESTING TO THE NEXT LEVEL.

TAAG Genetics, in partnership with Sequenom, offers a series of world-class prenatal tests. All these tests are based on cell-free DNA technology and massively parallel sequencing, they can provide you and your patients with unprecedented genetic information noninvasively.

These tests can report both common and rare fetal chromosomal abnormalities—from trisomies 21, 18 and 13 to fetal sex aneuploidies, trisomies 16, 22, and select copy number variants. Here, you'll find comprehensive, clinically relevant data that's transforming the way we assess prenatal health.

### THE MOST COMPREHENSIVE, VALIDATED TEST OF ITS KIND.

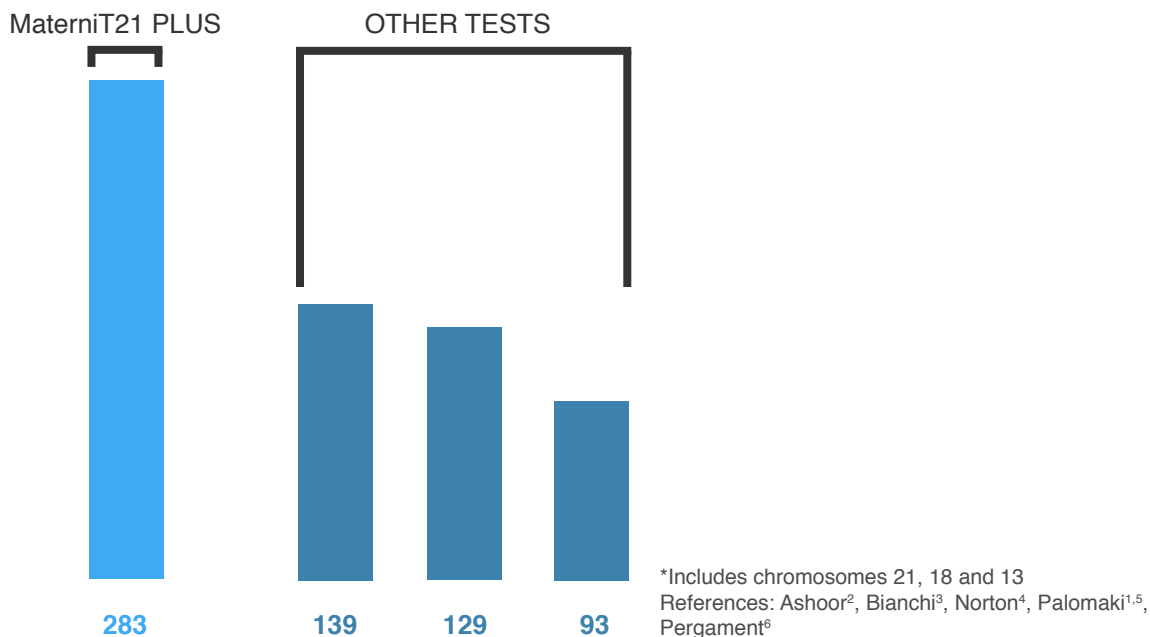
Sequenom's tests have been validated in the largest of-its-kind, independently designed, analyzed, and published clinical study with 4,664 pregnant women at increased risk for fetal chromosomal aneuploidies. Their robust data is derived from testing more than 2,800 pregnant women's blood samples and 375 trisomies.

### POSITIVE OR NEGATIVE RESULT

#### Get the answer you need the first time around.

Sequenom's tests report core results (trisomy 21, 18 and 13) as a positive or negative; never a percentage, and never a maybe. Our results provide you and your patients with fast, clear answers.

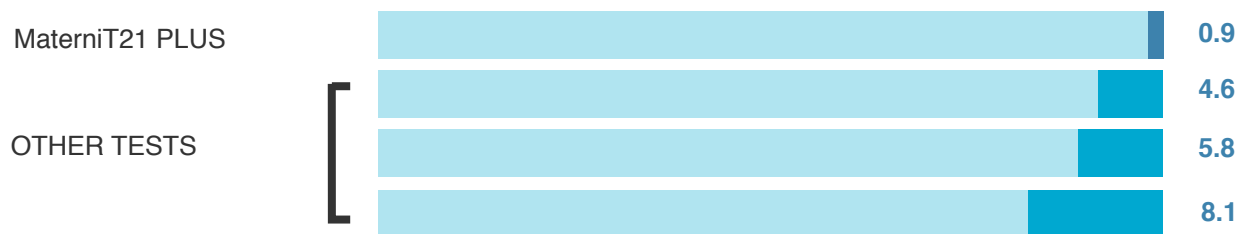
### TRISOMIES ANALYZED IN VALIDATION STUDIES



## INDEPENDENT VALIDATION<sup>1,5,7,8,9</sup>

POSITIVE RESULT	SENSITIVITY	SPECIFICITY
210 of 212 trisomy 21	99.1%	99.9%
59 of 59 trisomy 18	>99.9%	99.6%
11 of 12 trisomy 13	91.7%	99.7%
8 of 8 multiple gestations: 7 of trisomy 21 1 of trisomy 13	>99.9% detection rate	
Fetal gender	99.4% accuracy	
25 of 25 combined sex aneuploidies	96.2%	99.7%

## VALIDATION STUDY NON-REPORTABLE RATES (%)



References: Bianchi<sup>3</sup>, Norton<sup>4</sup>, Palomaki<sup>1,5</sup>, Pergament<sup>6</sup>

## INNOVATION TRANSLATING TO PREMIUM CONTENT

The premium sequenom's test (MaterniT21 PLUS) analyzes more chromosomal regions than any other noninvasive prenatal test, to date. Genomic regions/associated syndromes include:

### Trisomies:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Trisomy 16
- Trisomy 22

### Fetal Sex chromosomes:

- Fetal gender
- 45,X (Turner syndrome)
- 47,XXY (Klinefelter syndrome)
- 47,XXX (Triple X syndrome)
- 47,XYY (XYY syndrome)

### Enhances sequencing series:

- 22q (DiGeorge syndrome)
- 5p (Cri-du-chat syndrome)
- 1p36 deletion syndrome
- 15q (Angelman/Prader-Willi syndromes)
- 11q (Jacobsen syndrome)
- 8q (Langer-Gledion syndrome)
- 4p (Wolf-Hirschhorn syndrome)

## SOME OF OUR SOLUTIONS FOR PRENATAL CARE

### VisibiliT:

Simplest NIPT in the world. Massively parallel sequencing. Designed to mimic a serum screen.

- **Patient types:** Singletons, egg donors, surrogates, people of high consanguinity, bone marrow transplant recipients

- **Trisomy 21, 18:** Results presented as High Risk or Low Risk
- **Gender:** Available at no additional cost. Patient can opt out
- **Specimen:** 10 mL of maternal blood collected after 10 weeks of pregnancy
- **Turn around time:** On Average 20 days from receipt of sample
- **Non reportable rate:** 1.4%

### MaterniT21Plus:

Currently the largest panel in the world. Massively parallel sequencing.

- **Patient types:** Singletons, Twins, Triplets, Quads, egg donors, surrogates, people of high consanguinity, bone marrow transplant recipients

- **Trisomy 21, 18,13:** Reported as a Positive or Negative
- **Gender:** Available at no additional cost. Patient can opt out
- **Specimen:** 10mL of maternal blood collected after 10 weeks of pregnancy
- **Enhanced Sequencing Series reported as Additional Findings:**
- **Trisomies:** Trisomy 16, 22
- **Sex Chromosome Aneuploidies:** X, XXX, XXY, XYY, (not available in multifetal gestation)
- **Microdeletions:** DiGeorge, Prader-Willi/Angelman, Cri-du-Chat, 1p36, Jacobsen, Langer-Giedion, & Wolf-Hirschhorn Syndrome
- **Turn around time:** On Average 20 days from receipt of sample
- **Non reportable rate:** 1.4%

### MaterniTGenome:

New test, will be available very soon. Massively parallel sequencing

- **Patient types:** Singletons, egg donors, surrogates, people of high consanguinity, bone marrow transplant recipients

- **Karyotype like product:** Genome wide coverage (Chromosomes 1 through 22 as well as sex chromosomes). Anything greater than 7MB will be reported as a Positive or Negative (which is what you would see in an actual standard karyotype)

- **Gender:** Available at no additional cost. Patient can opt out
- **Specimen:** 10 mL of maternal blood collected after 10 weeks of pregnancy
- **All current MaterniT21Plus microdeletions with resolution down to 1.5MB will also be reported as a Positive or Negative.**
- **Turn around time:** On Average 20 days from receipt of sample
- **Non reportable rate:** 1.4%

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