MaterniT® GENOME

CLINICAL UPDATE
COMMERCIAL EXPERIENCE n = 28,760

The MaterniT GENOME laboratory-developed test screens all 23 pairs of chromosomes in the entire genome, with high sensitivity, specificity, and proven commercial reliability. It is designed to detect whole chromosome abnormalities, sex chromosome aneuploidies (SCAs), subchromosomal copy number variants (CNVs) ≥ 7 Mb, and select microdeletions.

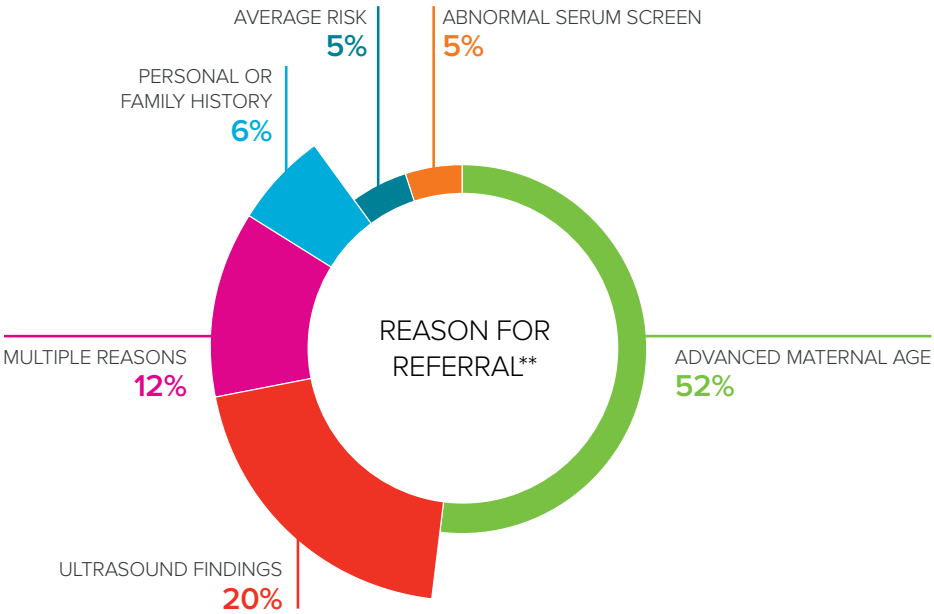
Performance*

Population sensitivity ≥97.7%

Specificity ≥99.9%

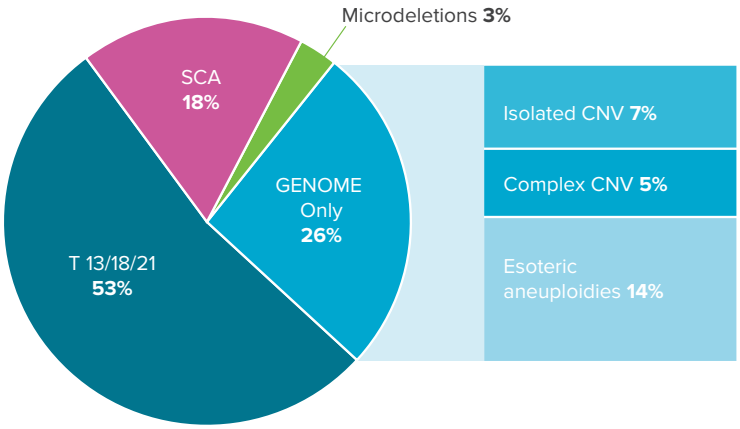
When should you use the MaterniT GENOME test?[1]

Based on peer experience, look for complex cases beyond advanced maternal age (AMA)



What will you see with the MaterniT GENOME test?[1]

Summary of the 1,392 positive results



* Sensitivity estimated from the samples in the published clinical validation study[2] and across the observed range of fetal fractions.

Actual sensitivity may also be influenced by other factors such as the size of the event, total sequence counts, amplification bias, or sequence bias.

** Per test requisition

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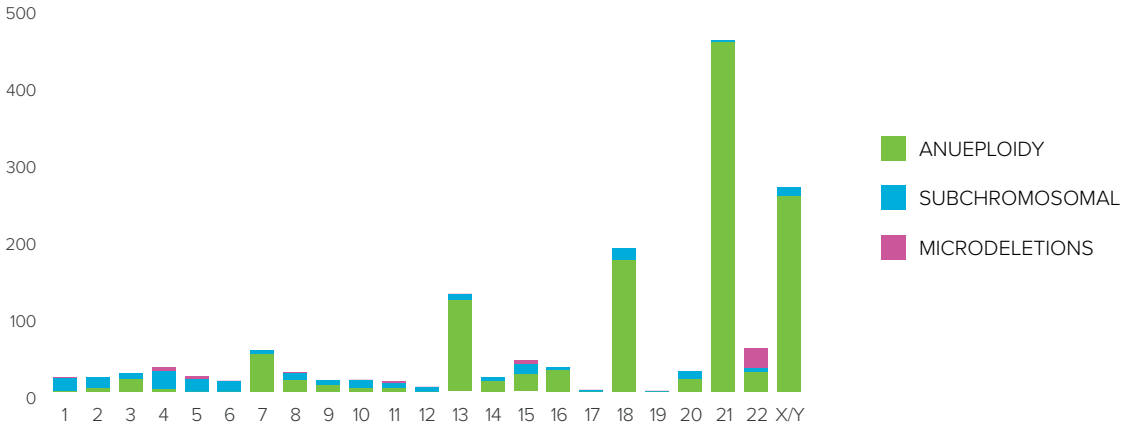
Key points[1,3]

- Genome-wide screening, 23 pairs of chromosomes
- Analyzes genome-wide aneuploids and events ≥7 Mb
- n = 28,760
- Leading reasons for referral beyond AMA
  - 20% Ultrasound findings
  - 12% Multiple indications
  - 6% Personal/family history
- Positive results were observed for every chromosome
  - 4.8% of all tests positive (n=1,392)
    - Of all positives, 14% reported as positive for esoteric aneuploidy (excluding chromosomes 21, 18, 13, X and Y)
    - The most common single esoteric aneuploidies involved chromosomes 7, 16, 22, 3, and 15
  - Nearly half of all cases positive for an esoteric aneuploidy had a clinically relevant outcome

MaterniT GENOME clinical experience finds up to 30% more chromosomal information than traditional NIPT as test adoption continues.[4]

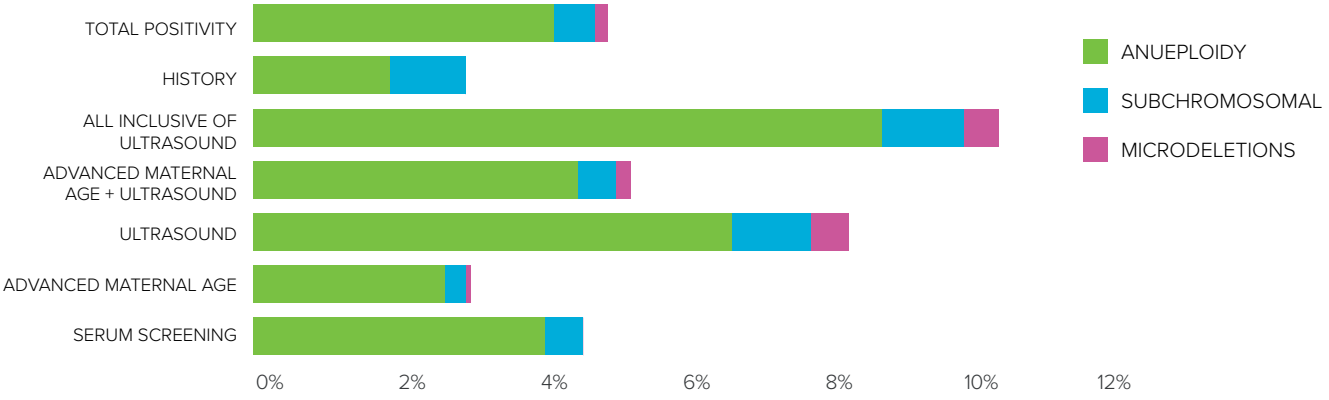
Abnormal findings identified across the entire genome[1]

Every chromosome is represented among the positive cases (n = 1,392)



Positivity rate varies by reason(s) for referral[5]

Abnormal ultrasound associated with higher positivity rate



Results from case studies are not predictive of results in other cases. Results in other cases may vary.

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1. Boomer T, Caldwell S, Almasri E, et al. Genome-wide cfDNA: Emerging data trends in 28k clinical samples. Poster presented at: 2017 National Society of Genetic Counselors; September, 2017; Columbus, OH.
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 Aug; 215(2):227.e1-227.e16. doi: 10.1016/j.ajog.2016.02.030.
3. Soster E, Boomer T, Dyr B, et al. Esoteric aneuploidies and genome-wide cell-free DNA. Poster presented at: 2017 National Society of Genetic Counselors; September, 2017; Columbus, OH.
4. Ehrich M, Tynan J, Mazloom A, et al. Genome-wide cfDNA screening: clinical laboratory experience with the first 10,000 cases. *Genet Med.* 2017;19(12):1332-1337.
5. Internal data