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Clinical validation study on MaterniT GENOME test published in American Journal of Obstetrics and Gynecology

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Sequenom, Inc., (NASDAQ: SQNM), a life sciences company committed to enabling healthier lives through the development of innovative products and services, today announced the publication of a clinical validation study on the MaterniT® GENOME laboratory-developed test in the *American Journal of Obstetrics and Gynecology*.

The study, titled *Clinical Validation of a Non-Invasive Prenatal Test for Genome-Wide Detection of Fetal Copy Number Variants (Lefkowitz, et al.),* demonstrated high resolution, sensitive and specific detection of a wide range of sub-chromosomal and whole chromosomal abnormalities that were previously only detectable by karyotype analysis of invasively obtained samples. In some instances, genome-wide noninvasive prenatal testing (NIPT) also provided additional clarification about the origin of genetic material that had not been elucidated by karyotype analysis.

"In 2011, Sequenom Laboratories pioneered cell-free DNA prenatal testing with the first laboratory-developed NIPT for trisomy 21, and we continued to innovate by adding content, such as other aneuploidies and select microdeletions," said Dirk van den Boom, PhD, President and CEO of Sequenom. "In 2015, we further evolved NIPT with the first genome-wide test, MaterniT GENOME. The results published in this clinical study reflect the diligent effort, expertise and scientific process that went into validating the MaterniT GENOME test."

The study compared the MaterniT GENOME test results for 1,166 clinical samples with results from analysis of invasively-obtained amniocentesis or chorionic villus sampling (CVS) material. It included 151 samples with common aneuploidies (trisomies 21, 18, and 13 and sex chromosome aneuploidies), as well as 8 rare aneuploidies and 35 subchromosomal abnormalities distributed across the genome. The study demonstrated that the MaterniT GENOME test was able to detect changes in copy number for full chromosomes, as well as subchromosomal copy number changes as small as seven megabases—a resolution comparable to traditional cytogenetic karyotyping.

"The ability to non-invasively detect chromosomal and subchromosomal abnormalities across the genome is critical in clinical practice; abnormalities beyond the major trisomies (21, 18, and 13) and sex chromosome aneuploidies comprise more than 20% of all karyotype-level abnormalities in the general obstetric population," said Dr. van den Boom. "The MaterniT GENOME test was designed to bridge this information gap, and our clinical laboratory experience to date with over 6,000 MaterniT GENOME tests has in fact shown that approximately 25% of samples with abnormal results had rare abnormalities across the entire genome that would have been undetectable by other cfDNA testing methods."

In this study, the MaterniT GENOME test showed:

- Sensitivity of >99.9% for trisomy 21 (CI 94.6-100%), trisomy 18 (CI 84.4-100%), trisomy 13 (CI 74.7-100%), and sex chromosome aneuploidies (CI 84-100%)
- Sensitivity of 97.7% (CI 86.2-99.9%) for genome-wide copy number variants other than trisomy 21, trisomy 18, trisomy 13, and sex chromosome aneuploidies
- Specificity of 99.9% or greater (CI 99.4-100%) for all chromosomes or chromosome regions analyzed
- Fetal sex classification accuracy of 99.6% (CI 98.9-99.8%)

"The MaterniT GENOME test has been extensively validated, both analytically and clinically," said Dr. van den Boom. "In addition to providing physicians and patients the most comprehensive information from any NIPT available today, the MaterniT GENOME test demonstrates Sequenom Laboratories' commitment to leadership in the NIPT marketplace and women's health."

Source:
Sequenom, Inc.
