Study finds massively parallel sequencing can detect fetal aneuploidies, including Down syndrome

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In a study to be presented today at the Society for Maternal-Fetal Medicine's annual meeting, The Pregnancy Meeting, in Dallas, Texas, researchers will report findings that indicate that massively parallel sequencing can be used to diagnose fetal aneuploidies, including Down syndrome, Edwards syndrome, Patau syndrome and Turner syndrome.

Because of the importance of the clinical data, the abstract, entitled Genome Wide Fetal Aneuploidy Detection by Sequencing of Maternal Plasma DNA: Diagnostic Accuracy in a Prospective, Blinded, Multicenter Study, was given late-breaker status. The study demonstrated that it was possible to use massively parallel sequencing of maternal plasma DNA in combination with a proprietary algorithm to detect the three most prevalent fetal aneuploidies (an abnormal number of chromosomes in the fetus). With this clinical evidence, this prenatal test may be incorporated into routine prenatal care.

"This is the first prospective, multicenter, blinded clinical study to demonstrate detection of all fetal aneuploidies across the genome. It demonstrates the efficacy of massively parallel sequencing of maternal plasma DNA with optimized normalization," said Diana W. Bianchi, M.D., of the Mother Infant Research Institute at Tufts Medical Center, Boston, Mass., the study's presenting author. "Our sample included 2,882 women undergoing prenatal diagnostic procedures at 60 different U.S. locations. Importantly, this study is representative of clinical practice, by

taking into account and reporting results from all whole chromosome aneuploidies."

The study correctly identified 89 of 89 cases of trisomy 21 (Down syndrome) with 100 percent sensitivity and specificity, and had a 100 percent positive predictive value for the three most common autosomal aneuploidies, trisomies 21, 18, and 13. The study also detected monosomy X (Turner syndrome) and other chromosome aneuploidies such as trisomies 16 and 20. This approach, if incorporated into prenatal care, will require far fewer invasive procedures to diagnose fetal aneuploidy.

In addition to Bianchi, the study was conducted by Lawrence Platt, David Geffen School of Medicine at UCLA, Obstetrics and Gynecology, Los Angeles, Calif.; James Goldberg, San Francisco Perinatal Associates, Prenatal Diagnosis Center, San Francisco, Calif.; Alfred Abuhamad, Eastern Virginia Medical School, Department of Obstetrics and Gynecology, Norfolk, Va.; Richard Rava, Verinata Health, Inc., Research & Development, Redwood City, Calif.; and Amy Sehnert, Verinata Health, Inc., Clinical Research, Redwood City, Calif.

More information: A copy of the abstract is available at www.smfmnewsroom.org/annual-me … 1-meeting-abstracts/

Provided by Society for Maternal-Fetal Medicine