

Noninvasive test for trisomy 21 closer at hand

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In 1980 in the United States, approximately 4.5% of all pregnant women were of advanced maternal age. By 2007 that figure had increased to 14%. Women over 35 are at increased risk of giving birth to babies with trisomy 21. In a study published online today in the *American Journal of Obstetrics & Gynecology* (AJOG), researchers from the Sequenom Center for Molecular Medicine confirmed that DNA sequencing of maternal blood plasma could accurately detect trisomy 21.

"In this study we have taken the next step in evaluating the practical use of fetal DNA sequencing and have shown that it has the potential to be highly accurate in a clinical setting," commented senior author Dirk van den Boom, DPhil. "We have implemented technical improvements that will increase sample throughput and reduce costs. Following completion of an ongoing clinical validation study the above improvements will greatly facilitate introduction into clinical practice. "

Because the plasma of pregnant women contains circulating cell-free (ccf) fetal (ccff) DNA the DNA sequencing method is able to identify the extra chromosome 21 material present in a fetus with trisomy 21. Independent of gestational age assessments, this novel approach allows for direct fetal assessment using massively parallel shotgun sequencing to detect missing or extra chromosomes, rather than surrogate biochemical markers that are used today in clinical practice.

Using samples from 449 high-risk [pregnant women](#), the DNA sequencing method correctly identified 39 trisomy 21 samples and 409

normal samples, and misclassified 1 normal sample as trisomy 21. The overall classification showed 100% sensitivity and 99.7% specificity.

The data show that, in the future, a noninvasive prenatal trisomy 21 test from ccff DNA might be used in concert with other clinical assessments, such as ultrasound, and become an option to better identify those women who would, or would not, benefit from confirmatory invasive diagnostic tests.

According to prenatal testing expert, Lee P. Shulman MD, The Feinberg School of Medicine of Northwestern University, Chicago, "The results of this study point more to a continuation in the improvement in the various non-invasive aneuploidy detection technologies that have been studied for the past two decades; hence, the results of this study can be considered a small step...Many more steps and studies will be needed in order to arrive at the goal that most of us who provide and study prenatal screening and diagnosis have long sought after: a facile, economical, and more accurate approach to prenatal screening and, eventually, an effective noninvasive alternative to invasive prenatal testing. We may not be there yet, but it seems that we have come a bit closer to our ultimate objective."

"This important study, along with another article recently published, suggests that progress is being made towards the realization of prenatal diagnosis of fetal disorders based upon maternal blood analysis and the use of high throughput technologies. While this study shows feasibility for the detection of trisomy 21, further studies are required to establish utility in clinical practice. A new frontier is the diagnosis, not only of trisomy 21 and other genetic disorders of the human fetus, but also the assessment of the functional state of the unborn baby by analysis of circulating fetal and/or placental RNA in the maternal circulation," said Roberto Romero, Chief of the Perinatology Research Branch of NICHD, NIH and Professor of Molecular Obstetrics and Genetics at

Wayne State University. Dr. Romero is an Associate Editor of the *American Journal of Obstetrics & Gynecology*.

More information: The article is "Noninvasive detection of fetal trisomy 21 by sequencing of DNA in maternal blood: a study in a clinical setting " by Mathias Ehrich, MD et al. It will appear in the *American Journal of Obstetrics & Gynecology*, Volume 204, Issue 3 (March 2011)

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