Study finds NIPT detects more than 80 percent of chromosomal abnormalities

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In a study to be presented on Feb. 6 at the Society for Maternal-Fetal Medicine's annual meeting, The Pregnancy Meeting, in New Orleans, researchers will report that noninvasive prenatal testing detected 83.2 percent of chromosomal abnormalities normally picked up by invasive diagnostic testing strategies, such as chorionic villus sampling (CVS) or amniocentesis. Noninvasive prenatal testing (NIPT) using cell free DNA provides accurate screening for the common trisomies, including trisomy 13 (Patau syndrome), 18 (Edwards syndrome), and 21 (Down syndrome).

In this study, titled Rare Chromosome Abnormalities Detected by Current Prenatal Screening Compared to Expected Performance using Non-Invasive Prenatal Testing (NIPT), 68,990 of 1,324,607 women tested positive for trisomy 18 or 21 when they underwent prenatal screening as part of the California Prenatal Screening Program between March 2009 and December 2012. Invasive diagnostic testing with CVS or amniocentesis was performed on 26,059 women who tested positive, and 2993 were found to have abnormal results. Of those chromosomal abnormalities, 2489 (83.2 percent) were abnormalities that would be detectable with NIPT, while 16.8 percent were less common aneuploidies that would not be detected.

One of the study's authors Mary Norton, M.D., said that more of the abnormal results were detectable in the women over 40, who are at higher risk for trisomy 13, 18 or 21. Conversely, fewer of the abnormalities in younger women would be detected by NIPT, as the risk

for common trisomies is lower in this group, while the rare aneuploidies are not typically associated with maternal age.

"While noninvasive prenatal testing with cell free DNA presents some real advantages in accuracy of screening for Down syndrome, as with everything there is a trade-off. Traditional aneuploidy screening with serum and ultrasound markers has higher false positive rates, but in these false positive cases are some fetuses with significant abnormalities that would not be found with NIPT. It is very important that patients and providers understand this trade-off," said Norton, professor and vice chair for Clinical and Translational Genetics, Department of Obstetrics, Gynecology and Reproductive Sciences at the University of California, San Francisco.

"In prenatal genetic testing, patient preferences are really the most important driver," Norton continued. "With this test, the patient makes a tradeoff between NIPT, which is noninvasive and detects most, but not all chromosome abnormalities—and is somewhat better in older women—and amniocentesis or CVS, which detect more chromosome abnormalities [8 to 25 percent more, depending on age] but with a small risk of miscarriage due to the procedure.

For an older woman, detecting 83 percent with the noninvasive test may be good enough, while for a 25-year-old, failing to detect 25 percent [which may include rare aneuploidies not usually associated with age] may be of concern."

Provided by Society for Maternal-Fetal Medicine