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Invasive Procedures Down With Noninvasive Prenatal Testing

Overall trisomy 21 detection similar before, after noninvasive testing introduction

THURSDAY, Dec. 18, 2014 (HealthDay News) -- The number of invasive diagnostic procedures, including amniocentesis, is down significantly after the introduction of noninvasive prenatal testing (NIPT), according to a study published in the January issue of the *Journal of Clinical Ultrasound*.

Joseph R. Wax, M.D., from the Maine Medical Center in Portland, and colleagues compared maternal demographics, aneuploidy risk factors, rates of genetic counseling, invasive diagnostic procedures, and trisomy 21 detection in women at increased risk for aneuploidy, before (1,464 patients) and after (1,046 patients) the availability of NIPT. All women had an ultrasound examination between 10 0/7 and 21 6/7 weeks' gestation and were eligible for NIPT because of age ≥ 35 years, ultrasound findings suggestive of increased aneuploidy risk, positive aneuploidy screen, prior trisomic fetus, or parental balanced translocation with increased risk for trisomy 13 or 21.

The researchers found that all 33 fetuses with trisomy 21 in the two groups were identified by positive aneuploidy screening. Genetic counseling for aneuploidy risk increased after the introduction of NIPT (adjusted odds ratio [aOR], 1.77; $P < 0.0001$). After NIPT introduction, the overall invasive diagnosis (aOR, 0.42; $P < 0.0001$), including amniocentesis (aOR, 0.37; $P < 0.0001$), decreased, while the prenatal diagnosis of trisomy 21 remained similar (88 versus 100 percent; $P = 0.86$).

"NIPT in clinical practice uses more genetic counseling resources but requires significantly fewer invasive procedures to maintain the detection rates of trisomy 21," the authors write.

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April 21, 2015

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