Majority of women prefer non-invasive testing for trisomy 21

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(HealthDay)—The majority of both high-risk and intermediate-risk women prefer a non-invasive cell-free prenatal DNA screening (NIPT) as a secondary screening test for trisomy 21, according to a study published online Nov. 10 in *BJOG: An International Journal of Obstetrics and Gynaecology*.

Yvonne Kwun Yue Cheng, M.B.Ch.B., from The Chinese University of Hong Kong, and colleagues examined preference for follow-up testing in women who were screened as high or intermediate risk in first- or second-trimester Down syndrome screening. Testing options included either an invasive test plus chromosomal microarray to obtain more detailed fetal genetic information or a NIPT to detect trisomies 13, 18, and 21 to avoid procedure-related miscarriage; the participants could also decline further testing.

The researchers found that of the 347 high-risk women, nearly all (99.1 percent) opted for follow-up testing, of whom 62.2 percent (216 women) chose NIPT. Of the 614 participants with intermediate risk, 82.6 percent (507 women) chose NIPT. Twenty-one percent of 34 women with nuchal translucency ?3.5 mm opted for NIPT.

"In a setting where reporting times are similar and there is no cost difference between options, approximately 60 percent of high-risk women would opt for NIPT offering simple but limited aneuploidy assessment over a diagnostic procedure with comprehensive and more detailed assessment," the authors write.

One author disclosed a royalty-producing patent related to non-invasive prenatal diagnostics.

More information: Abstract
Full Text (subscription or payment may be required)