

Certain abnormal prenatal testing results and subsequent diagnosis of maternal cancer

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In preliminary research, a small number of occult (hidden) malignancies were subsequently diagnosed among pregnant women whose noninvasive prenatal testing results showed chromosomal abnormalities but the fetal karyotype was subsequently shown to be normal, according to a study appearing in *JAMA*. The study is being released to coincide with its presentation at the 19th International Conference on Prenatal Diagnosis and Therapy in Washington, D.C.

Understanding the relationship between aneuploidy detection (an abnormal number of chromosomes) on [noninvasive prenatal testing](#) (NIPT) and occult maternal malignancies may explain abnormal NIPT results that are discordant with the actual fetal karyotype (the chromosomal characteristics of a cell) and improve maternal clinical care. Many professional societies have recommended that NIPT be offered to pregnant women at high risk for having a fetus with autosomal (pertaining to a chromosome that is not a sex chromosome) aneuploidy, with follow-up diagnostic testing (amniocentesis or chorionic villus sampling) recommended to confirm a positive test result, according to background information in the article.

Diana W. Bianchi, M.D., of Tufts Medical Center, Boston, and colleagues examined DNA sequencing data in a series of pregnant women with abnormal NIPT results involving aneuploidies of certain chromosomes, who were diagnosed with cancer after prenatal testing occurred. The case patients were identified from 125,426 samples submitted between February 2012 and September 2014 from

asymptomatic pregnant women who underwent plasma cell-free DNA sequencing for clinical prenatal aneuploidy screening.

Among the clinical samples, 3,757 (3 percent) were positive for 1 or more aneuploidies involving chromosomes 13, 18, 21, X, or Y. These were reported to the ordering physician with recommendations for further evaluation. From this set of 3,757 samples, 10 cases of maternal cancer were identified. Detailed clinical and sequencing data were obtained in 8. Maternal cancers most frequently occurred with the rare NIPT finding of more than 1 aneuploidy detected (7 known cancers among 39 cases of multiple aneuploidies by NIPT, 18 percent). In 1 case, blood was sampled after completion of treatment for colorectal cancer and the abnormal pattern was no longer evident.

"Here we have shown that occult maternal malignancies may provide a biological explanation for some discordant NIPT results. This is presumably due to the cell-free DNA that is released into maternal circulation from apoptotic [death of cells] malignant cells," the authors write.

The researchers add that these data underscore the necessity of performing a diagnostic procedure to determine the true fetal karyotype whenever NIPT results reveal [chromosomal abnormalities](#). "When there is discordance between the fetal karyotype and NIPT result, occult maternal malignancy, although very uncommon, may be an explanation for the findings. Based on the results of the study, we estimate there is between a 20 percent and 44 percent risk of maternal cancer if multiple aneuploidies are detected. However, until further studies are done to assess the clinical implications of discordant NIPT and fetal karyotype results, it is not clear what, if any, follow-up clinical evaluation is appropriate."

"At this time, there is insufficient evidence about the benefits, risks, and

costs of reporting the incidental findings, as Bianchi et al mention," write Roberto Romero, M.D., D.Med.Sci., of the Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health, Bethesda, Md., and Maurice J. Mahoney, M.D., J.D., of the Yale University School of Medicine, New Haven, Conn., in an accompanying editorial.

"As the authors correctly recommend, the data emphasize the need for performing a diagnostic procedure to determine the fetal karyotype in all situations in which there is an abnormal NIPT result. Given that it is likely that NIPT will increase in the coming years, an active dialogue among stakeholders (obstetricians, patients, laboratories, ethicists, policy makers, etc.) needs to take place to provide informed advice to potentially affected [pregnant women](#) and to guide the care of such patients."

More information: *JAMA*, [DOI: 10.1001/jama.2015.7120](https://doi.org/10.1001/jama.2015.7120)
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