

ACMG releases updated position statement on noninvasive prenatal screening for detection of fetal aneuploidy

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Noninvasive prenatal screening using cell-free placental DNA circulating in maternal blood (NIPS) has been rapidly integrated into prenatal care since the American College of Medical Genetics and Genomics (ACMG) released its 2013 Position Statement on its use. "New data and provider and patient demand required an updated ACMG position on the use of NIPS in prenatal care," said lead author of the ACMG statement and high-risk pregnancy physician, Anthony R. Gregg, MD, MBA, FACOG, FACMG. "We provide a framework for understanding how genetic technology moves from an idea into clinical practice. ACMG continues to stress that NIPS is a screening rather than a diagnostic test and positive results should be followed by a diagnostic test such as chorionic villus sampling or amniocentesis." (Diagnostic tests eliminate the problem of false positive and false negative results associated with a screening test).

The updated statement, "Noninvasive prenatal [screening](#) for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics," will be published in the ACMG's peer-reviewed academic journal, *Genetics in Medicine* online Ahead of Print (AOP) Thursday, July 28, 2016.

The ACMG statement focuses on topics that obstetric care providers and patients should discuss when considering prenatal aneuploidy screening options. It includes 44 specific recommendations for providers and the

laboratories that offer NIPS. One goal is to ensure patients receive accurate and up-to-date information. Another goal is to ensure obstetric care providers have clear, precise and accurate laboratory reports as well as educational tools and resources in order to counsel patients.

Key questions and recommendations include:

1. Should NIPS be offered to all pregnant patients?
 - ACMG recommends:
 - informing all pregnant women that NIPS is the most sensitive screening option for traditionally screened aneuploidies involving chromosomes 13, 18 and 21 (i.e., Patau, Edwards and Down syndrome).
2. How are "no calls" avoided, interpreted and managed? ("No call" is the technical term reported when the laboratory is unable to obtain a NIPS result; it is most commonly due to insufficient fetal cell-free DNA in the sample).
 - ACMG recommends
 - offering diagnostic testing for a "no call" NIPS result due to low fetal fraction if maternal blood for NIPS was drawn at an appropriate gestational age. A repeat blood draw is NOT appropriate.
 - offering aneuploidy screening other than NIPS in cases of significant obesity.
3. Should NIPS be offered to screen for sex chromosome aneuploidies?
 - ACMG recommends
 - informing all pregnant women as part of pre-test counseling for NIPS, of the availability of the expanded use of screening for sex chromosome aneuploidies.
 - providers should make efforts to deter patients

from selecting sex chromosome aneuploidy screening for the sole purpose of biologic sex identification, in the absence of a clinical indication for this information.

4. Should NIPS be offered for detection of copy number variation (CNV)?
 - ACMG recommends
 - informing all pregnant women of the availability of the expanded use of NIPS to screen for clinically relevant CNVs when specific conditions can also be met.

The position statement offers guidance to obstetric care providers and makes the following specific recommendations:

- referring patients to a trained genetics professional for any positive NIPS result.
- offering diagnostic testing for a positive NIPS result.
- providing accurate, balanced, up-to-date information, at an appropriate literacy level when a fetus is diagnosed with a condition after prenatal screening and confirmatory diagnostic testing. These materials should reflect the medical and psychosocial implications of the diagnosis (see Patient and Provider Resources).
- informing patients of the possibility of identifying maternal genomic imbalances which could potentially impact maternal health, and that this possibility depends on the specific methodology used.

Some of the recommendations for laboratories that perform NIPS include that:

- laboratories provide readily visible and clearly stated detection

rate (DR), specificity (SPEC), positive predictive value (PPV) and negative predictive value (NPV) for all conditions being screened, in pre-test marketing materials and when reporting laboratory results, in order to assist patients and providers in decision-making and results interpretation.

- laboratories should not offer screening for any condition for which it is unable to report the DR, SPEC, PPV and NPV.
- all laboratories include a clearly visible fetal fraction on NIPS reports.
- all laboratories specify the reason for a "no call" when reporting NIPS results.
- laboratories work with public health officials, policymakers, and private payers to make NIPS, including the pre- and post-test education and counseling, accessible to all [pregnant women](#).

ACMG Executive Director, Michael S. Watson, PhD, FACMG, another author of the new ACMG statement and member of The Noninvasive Prenatal Screening Work Group of the American College of Medical Genetics and Genomics added, "Ultimately, we believe that a patient, with guidance from her provider, should be able to make an informed decision on the current use of [prenatal screening](#) options including NIPS and understand the ramifications of a positive, negative or "no call" result."

The ACMG statement also stresses the importance of patient counseling before and after testing, and it provides a list of patient resources and useful references for medical providers.

Provided by American College of Medical Genetics

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