

ACMG releases statement on noninvasive prenatal screening

April 8 2013

The American College of Medical Genetics and Genomics (ACMG) has just released an important new Policy Statement on "Noninvasive Prenatal Screening for Fetal Aneuploidy." The Statement can be found in the Publications section of the ACMG website at <http://www.acmg.net> and will soon be published in the peer-reviewed medical journal, *Genetics in Medicine*.

As background, in recent decades there have been many changes and improvements in prenatal genetic screening and diagnosis. The risk, however, of testing with specimens obtained by [invasive procedures](#) such as amniocentesis and chorionic villus sampling (CVS) has led to the search for new methods using mother's blood specimens obtained noninvasively. The most recent advances in genomics and genomic technologies have resulted in such noninvasive prenatal screenings (NIPS). The acronym NIPS is used to emphasize the screening nature (false positives and false negatives do occur) of tests currently on the market.

The new ACMG Statement on Noninvasive [Prenatal Screening](#) addresses:

- The current limitations of NIPS
- The advantages of NIPS compared with current screening approaches
- Pretest and posttest [genetic counseling](#)

- The reporting of results by laboratories performing NIPS
- Oversight of analytical and bioinformatic components by testing of the laboratories performing NIPS

The Statement says that while studies are promising and demonstrate high sensitivity with low false-positive rates, there are limitations to NIPS, "NIPS for fetal aneuploidy has arrived; however, as with most new technologies, there is room for refinement." The report strongly states that positive results should be followed-up with an invasive [diagnostic test](#) before any decision is made regarding [pregnancy termination](#).

Lead author of the ACMG Statement Anthony R. Gregg, MD, FACOG, FACMG and high- risk pregnancy physician said, "Obstetric care providers must become familiar with the advantages and disadvantages of the use of this approach. Clinicians should provide patients with both pretest and posttest counseling with the goal of avoiding patient harm or confusion - I can't stress this enough."

Gregg added, "Most of the companies that are developing these tests have referred to it as NIPDiagnosis or NIPTest. In our view, it is NOT a diagnostic test such as chorionic villus sampling [CVS] or amniocentesis; hence, we coined the term Noninvasive Prenatal SCREENING (NIPS)."

NIPS was initially validated for Down syndrome screening and has been applied to other trisomies including 13 and 18 with sex chromosomes being added now.

ACMG Medical Director Barry Thompson, MD, FACMG, another author of the Statement added, "NIPS is now one of many approaches available to women who desire Down syndrome screening. Unlike other methods, it is minimally invasive in that it only requires a blood sample from the pregnant mother rather than the more invasive [amniocentesis](#) or

CVS that have associated risks of miscarriage."

"NIPS is a very accurate screening test, " said Michael S. Watson, PhD, Executive Director of the American College of [Medical Genetics](#) and Genomics. However, it is well known that the cells originate from 'extraembryonic tissues' around the fetus so aneuploidy status may not always be identical to the genetics of the fetus. The advantages include that the detection rate is higher, the negative predictive value is greater, and the false positive rate is lower, than any other current screening approaches for Down syndrome. It must be followed up, however, by a diagnostic test since NIPS is a [screening](#) test."

"Finally, NIPS does not replace a first trimester ultrasound (12-14 weeks); rather, it complements it," Watson added.

Provided by American College of Medical Genetics

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