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# Noninvasive Prenatal Screening 2016 ACMG Position Statement

Narrator:
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Dr. Chavez:
I am Dr. Martin Chavez, Director of Maternal Fetal Medicine and Fetal Surgery Program in the Department of Obstetrics, Gynecology at Winthrop University Hospital.

Today I'm going to be talking about the American College of Medical Genetics and Genomics latest position statement on Non-Invasive Prenatal Screening, or NIPS. These guidelines came out in July 2016 and are the most current guidelines concerning this topic. While they cover many aspects of NIPS, we will focus only on two specific points, but the listener is strongly encouraged to read the position statement in its entirety.

Since the introduction of noninvasive prenatal screening there has been an increase in utilization of this technology. It has rapidly clinically expanded not only for high risk patients but for "low risk" patients as well. Noninvasive prenatal screening for aneuploidy in the past had relied on maternal serum analytes and/or ultrasonography. Detection rate varied from 50-95% depending on which analytes or if the ultrasound was incorporated. False positive rate was approximately 5%.

Two major points of the latest position statement have to do with offering all pregnant patients NIPS as well as how to interpret and manage "no calls". The statement also discusses clinical utility and whether a screening test is reliable and useful to the patient. This technology has shown to be not only reliable but extremely useful to the patient and clinician as well.

In its recommendations, it states that clinicians should be "Providing up-to-date, balanced, and accurate information early in gestation to optimize patient decision making, independent of the screening approach used."

When dealing with the question about whether NIPS should be offered to all patients, including low risk patients, the statement makes clear that it is appropriate to offer this technology to all. Pregnant women should be informed about NIPS and that it is the most sensitive screening option for the traditionally screened aneuploidies such as trisomy 21, 18 and 13. Also, it mentions the importance of referring patients who have an increased risk of aneuploidy by NIPS to a trained genetics professional. They emphasize the importance of diagnostic testing when a positive screening test result is reported after NIPS as well.

The position statement also dealt with interpretation and management of "No Calls" and how to avoid them. It recommends that diagnostic testing, CVS or amniocentesis, be offered for NIPS results due to low fetal fraction, assuming maternal blood for NIPS was done at the appropriate gestational age. This is in response to an increased risk of fetal aneuploidy with a "No Call" result. A repeat blood draw would not be appropriate under these circumstances. In cases of significant obesity, offering aneuploidy screening other than NIPS would be reasonable. The statement recommends that all laboratories include a clearly visible fetal fraction on NIPS results. They also further emphasize the importance that all laboratories specify the reason for a "No-Call" when reporting NIPS results.

In summary, all at-risk patients, including low risk patients, can be offered NIPS. All patients should have pre- and post-test counseling. With a positive result or a "No-Call" result, diagnostic testing should be offered as well as referral to a genetic professional for further counseling.

Narrator:
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