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HIGHLIGHTS FROM ACOG-SMFM

PRACTICE BULLETIN #226 ON SCREENING FOR FETAL CHROMOSOMAL ABNORMALITIES

The American College of Obstetricians and Gynecologist (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) issued an ACOG Practice Bulletin on screening for fetal chromosomal abnormalities (PB #226). PB#226 replaces the previous PB#163 from May 2016 (reaffirmed 2018).

This information sheet highlights key points related to ACOG Practice Bulletin # 226, which includes expanded information regarding the use of cell-free DNA (cfDNA) in all patients, regardless of maternal age or baseline risk.

What is new in cfDNA screening?

ACOG now endorses cell-free DNA (cfDNA) screening as a first-line screening option for all pregnant women. ACOG also recognizes cfDNA screening as the most sensitive and specific screening test for fetal aneuploidies.

Key Highlights

- **cfDNA screening is an option for all pregnant women, regardless of maternal age or background risk**
  - While the previous guideline endorsed offering screening or diagnostic testing to all pregnant women, PB#226 emphasizes that cfDNA screening is an appropriate test to offer as a first-tier screen for all pregnant women, regardless of risk.
- **cfDNA screening is the most sensitive and specific screening option for the common aneuploidies**
  - PB#226 clearly endorses cfDNA screening as having the highest detection rate and lowest false positive rate of all screening options, regardless of maternal age or baseline risk.
  - As with all screening tests, there is the potential for false-positive and false-negative results and is not a substitute for diagnostic testing.
- **Pregnant women should make an informed choice regarding screening and diagnostic testing after discussion with their health care provider**
  - Prenatal screening and testing choices are influenced by many factors. Ultimately, the decision is personal and can be based on discussions between the patient and her health care provider.

Additional Recommendations

- cfDNA screening can be performed in twin pregnancies.
- NIPT results which include fetal fraction measurements are preferable.
- Technical failures of cfDNA screening (no-call results) are associated with a higher risk of aneuploidy. Women who receive a no-call NIPT result should have genetic counseling and be offered ultrasound and diagnostic testing. In some cases, repeat NIPT may be considered.
- Multiple aneuploidy screening tests performed simultaneously should not be used.
- Expanded NIPT for microdeletions, rare autosomal aneuploidies, or genome-wide changes is not recommended.