Obesity and no-call results: optimal timing of cell-free DNA testing and redraw

We thank the authors for their comments. Although cell-free DNA (cfDNA) testing has the highest detection rate among aneuploidy screening modalities, we agree that the reportedly high screening failure rates in women with obesity affects its use in this population. We also agree that the optimal aneuploidy screening option depends on the patient and her preferences.

Our study sought to determine the screening failure rate in women with obesity cases with an initial screen failure and to assess the increase in the fetal cfDNA fraction over time in the most obese women. We concluded that more than 80% of women weighing >400 lb received results between 9 and 12 weeks' gestation and that approximately 94% of women weighing >400 lb received results between 13 and 18 weeks' gestation. The no-call result rate owing to low fetal fraction is lower than previously reported, indicating that cfDNA screening is appropriate in women who desire aneuploidy screening with the highest detection rate and lower false-positive rates.

The authors' screening algorithm does not address the limitations of other forms of aneuploidy screening in women with obesity. In particular, there may be a higher rate of unobtainable nuchal translucency and greater scanning time required for women with obesity.[1,2] Studies have shown that targeted and standard sonography have higher failure rates and lower detection rates for soft aneuploidy markers in women with obesity.[3,4] In addition, the higher false-positive rate in screening with ultrasound or serum analyte screening may lead to increased invasive testing, which can be technically more challenging in the most obese women. Therefore, we believe that cfDNA screening offers a viable aneuploidy screening option for patients with obesity with the appropriate pretest counseling. ∎

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