Single-Nucleotide Polymorphism–Based Noninvasive Prenatal Screening in a High-Risk and Low-Risk Cohort

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1. What is the primary objective of this study? Identify the study design used by the investigators and discuss the strengths and weaknesses of this approach for this study question.

2. Is noninvasive prenatal screening for aneuploidy currently offered to all patients at your institution? If not, which patients at your institution might be candidates for noninvasive screening? How effective is your screening process? Discuss any existing barriers to screening eligible patients at your institution.

3. The authors use a single-nucleotide polymorphism–based technique for noninvasive prenatal screening. List and briefly describe all available genetic techniques for noninvasive prenatal screening for aneuploidy (refer to any resources, including American College of Obstetricians and Gynecologists Committee Opinion No. 545: Noninvasive Prenatal Testing for Fetal Aneuploidy).

4. Describe the key findings from this study. Discuss which findings are novel.

5. “No-call” samples were excluded when computing the predictive characteristics of noninvasive prenatal screening in this study. Explain the meaning of “no-calls” and discuss whether or not you agree with their exclusion from the screening performance computations.

6. Conduct a literature search to identify any randomized studies comparing noninvasive prenatal screening to other screening approaches for aneuploidy. Describe your search strategy and state the number of trials you identified. In your opinion, is there a need for a randomized trial of prenatal genetic screening methods? Why or why not?

7. The authors conclude that the prenatal screen performed with high sensitivity and specificity in both high-risk and low-risk cohorts. How did the authors define both low-risk and high-risk cohorts? Based on the findings of this study should routine noninvasive prenatal screening be offered to low-risk women? Discuss the advantages and disadvantages of screening low-risk women.