Society for Maternal-Fetal Medicine (SMFM) Special Report:

SMFM Statement: clarification of recommendations regarding cell-free DNA aneuploidy screening

Society for Maternal-Fetal Medicine (SMFM) Publications Committee

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The purpose of this statement is to clarify that the Society for Maternal-Fetal Medicine (SMFM) does not recommend that cell-free DNA aneuploidy screening be offered to all pregnant women, nor does it suggest a requirement for insurance coverage for cell-free DNA screening in women at low risk of aneuploidy. However, SMFM believes, due to the ethics of patient autonomy, that the option should be available to women who request additional testing beyond what is currently recommended by professional societies.

Key words: cell-free DNA aneuploidy screening, prenatal aneuploidy screening, diagnostic testing, genetic testing

The Society for Maternal-Fetal Medicine (SMFM) recent guidance and recommendations regarding cell-free DNA (cfDNA) aneuploidy screening are presented in the SMFM Consult Series no. 36 *Prenatal aneuploidy screening using cell-free DNA*[1] and the joint SMFM and American Congress of Obstetricians and Gynecologists Committee Opinion no. 640, *Cell-free DNA screening for fetal aneuploidy.*[2] In these documents, the key recommendations include that: (1) a discussion of the risks, benefits, and alternatives of various methods of prenatal screening and diagnostic testing, including the option of no testing, should occur with all patients; and (2) given the performance of conventional screening methods, the limitations of cfDNA screening performance, and the limited data on cost-effectiveness in the low-risk obstetric population, conventional screening methods remain the most appropriate choice for first-line screening for most women in the general obstetric population. Acknowledging the ethics of actively withholding available tests from one group, the recommendations further suggest that although any patient may choose cfDNA analysis regardless of risk status, the patient choosing this testing should understand the limitations and benefits in the context of alternative screening and diagnostic options and be provided discussion of the limitations of testing in the low-risk population.

The purpose of this statement is to clarify that SMFM does not recommend that cfDNA aneuploidy screening be offered to all pregnant women, nor does it suggest a requirement for insurance coverage for cfDNA screening in women at low risk of aneuploidy. However, SMFM believes, due to the ethics of patient autonomy, that the option should be available to women who request additional testing beyond what is currently recommended by professional societies. This is comparable to the recommendation that it is ethically permissible for physicians to perform chorionic villus sampling or amniocentesis for genetic testing upon maternal request to low-risk women.[3]

Limited data at the present time on the effectiveness and clinical utility for improving patient outcomes preclude a recommendation that cfDNA be actively offered to all pregnant women. This recommendation is supported by a recent study in which the authors found that cfDNA screening was only optimal as a first-line test at a maternal age of $\geq$40 years.[4]

SMFM recognizes the value of cfDNA screening for women at higher risk for aneuploidy but considers that cfDNA screening is not the appropriate choice for first-line screening for the low-risk obstetric population at the present time. For this population, conventional screening methods remain the preferred approach. Given the misconceptions regarding interpretation of cfDNA screening results and the serious consequences that have been documented,[5] there are significant concerns about the consequences of broad utilization of this test in low-risk women, the vast majority of whom do not undergo genetic counseling or detailed pretest counseling with a provider. The recommendation that pregnant women understand the limitations and benefits of this screening test should be an absolute

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requirement prior to performing the test, with a need for documentation of how that requirement was met. This might include genetic counseling by an independent counselor, documentation of review of unbiased educational materials, and other methods beyond simple ordering of the test. In such circumstances, SMFM would suggest that such counseling should be required prior to payment for this test.

SMFM is strongly committed to advancing care for women and children by raising the standards of prevention, diagnosis, and treatment of maternal and fetal disease. This mission requires a careful evaluation of the evidence surrounding the clinical utility of available interventions and providing advocacy and health policy leadership. SMFM will continue to closely follow advances in the area of genetic testing, as well as in all aspects of maternal-fetal care to assure optimal care for women and to provide guidance for maternal-fetal medicine subspecialists. ■

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