When is cell-free DNA best used as a primary screen?

At age 38 years, cell-free DNA screening as the first-line test becomes the optimal strategy; at age 40 years, cell-free DNA as a primary screen becomes optimal and is cost-effective, according to this decision-analytic model study in which investigators compared the clinical outcomes, quality-adjusted life-years, and costs associated with 6 strategies of prenatal testing.

FAST TRACK

Cell-free DNA screening technology is rapidly changing, but ACOG's current guideline is the best approach for screening practices

Kaimal AJ, Norton ME, Kupperman M. Prenatal testing in the genomic age: clinical outcomes, quality of life, and costs. Obstet Gynecol. 2015;126(4):737–746.

EXPERT COMMENTARY

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The author reports no financial relationships relevant to this article.

Cell-free DNA screening, or so-called noninvasive prenatal testing (NIPT), has had greatly increased utilization recently as advances in technology have elevated it almost to the level of a diagnostic test for detection of certain aneuploidies. Although it is still considered a screening test, recent interest has arisen regarding population screening and whether or not this test should be universally used as first-line or whether it should still be restricted to specific high-risk populations.

WHAT THIS EVIDENCE MEANS FOR PRACTICE

For now, the best approach would be to adhere to current recommendations as outlined in the 2015 American College of Obstetricians and Gynecologists (ACOG) Committee Opinion.[1] Summarized, these are:

- Do not utilize NIPT in low-risk populations (although this opinion also suggests that patients may opt to have this test performed regardless of risk status with the understanding that detection rates are lower in low-risk populations and insurance coverage may be different).
- Offer NIPT to high-risk women as a first-line screen, as is suggested in the current study with respect to maternal age criteria (ACOG uses an age cut-off of 35 years, not 38).
- Utilize NIPT as a follow-up test after conventional testing suggests increased risk status in those patients wishing to avoid invasive diagnostic testing.
- The ACOG position remains that all women, regardless of age, who desire the most comprehensive information available regarding fetal chromosomal abnormalities should be offered diagnostic testing (chorionic villus sampling and amniocentesis).

Also, as mentioned in the committee opinion, this technology is evolving rapidly and all practitioners should closely follow this evolution with respect to changing efficacy and changing cost.

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Examining the
EVIDENCE

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In the current study, Kaimal and colleagues attempted to determine the best strategy for utilization of NIPT using a decision-analytic model. For their study many assumptions had to be made in order to allow for calculation of detection rates and for determination of cost and quality-adjusted life years. (The model followed a theoretical cohort of women desiring prenatal testing [screening or diagnostic or both] from the time of their initial test through the end of their pregnancy, the birth of their neonate, and the remainder of their own life expectancy.)

The conclusion of the authors is that traditional multiple marker screening remains the optimal choice for most women (those aged 20 to 38 years) but that NIPT becomes the optimal strategy at age 38. The goal of this study was not just to determine cost-effectiveness but also to attempt to devise a strategy that would optimize detection of aneuploidy and minimize the need for the performance of diagnostic procedures.

Data not available in this study included such things as population differences with respect to the acceptance of pregnancy termination as an option, and the potential utility of first-trimester ultrasound screening for structural defects that might not be accounted for with NIPT (such as thickened nuchal translucency, altered cardiac axis, cranial defects, and abdominal wall defects).

Reference

1. American College of Obstetricians and Gynecologists. Committee Opinion No. 640. Cell-free DNA screening for fetal aneuploidy. Obstet Gynecol. 2015;126(3):e31–e37.