Precision medicine: What to know about cell-free DNA screening

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With more women seeking tests for common chromosome conditions in pregnancy, many are now opting for newly developed non-invasive cell-free DNA (cfDNA) screening. But like all screening tests, it has limitations and isn’t appropriate for all patients. Find out how cfDNA works and which of your patients may benefit from the screening.

A new continuing medical education (CME) module, developed by AMA in partnership with Scripps Translational Science Institute and The Jackson Laboratory, is helping physicians understand what the test detects, which patients benefit most from it, what to consider when ordering the test and a lot more.
First, how cfDNA screening works

Prenatal cfDNA screening detects small fragments of fetal DNA released by placental cells into the mother’s blood stream. cfDNA screening looks for a relative increase or decrease in specific regions of the fetal DNA that would suggest the presence of a chromosome condition.

The screening is now being offered for trisomies 21, 18 and 13. Some tests also include sex chromosome conditions and a few conditions caused by chromosomal micro-deletions or micro-duplications, but research is still underway to verify their clinical validity. Unlike other maternal serum screening tests, cfDNA cannot detect structural birth defects, so additional testing may be necessary.

Screening for cfDNA is not a diagnostic test, but rather a screening test. Results can only determine if a woman is at increased or decreased risk for chromosome conditions. If the results suggest an increased risk, more extensive diagnostic testing is needed.

Who benefits most from it?

While originally offered only to women at high risk for chromosome conditions, many professional societies now support offering cfDNA screening to women in the general obstetric population. Women at high risk for chromosome conditions include those of advanced maternal age and women with a positive screening test or who have a previous child with a chromosome condition.

The CME module covers numerous topics to help physicians determine if prenatal cfDNA screening is right for a patient, including:

- Benefits and limitations of the test
- Interpreting results
- Considerations when ordering the test
- Indications and contraindications
- Counseling women who are considering the test

The module also features point-of-care guidance about prenatal cfDNA screening, access to a predictive value calculator, comparisons of cfDNA screening to other prenatal screening and testing options and other tools for physicians, including resources physicians can share with patients.

Test your knowledge: A case study


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Megan is 30 years old and at 12 weeks gestation in her first pregnancy. She is worried about invasive testing but still wants to learn as much as possible about pregnancy risks. She read online about cfDNA screening and is interested in cfDNA as an alternative for chorionic villus sampling (CVS) or amniocentesis. What would you tell Megan about whether cfDNA screening is appropriate for her? Can cfDNA screening take the place of CVS or amniocentesis?

Case studies like these are included in the module and enable you to test your knowledge of cfDNA and practice applying it to patient scenarios.

You can also dig deeper into the benefits and limitations of cfDNA screening, find answers to logistical issues—like how to work with genetics experts and how to find labs that offer the test—and easily compare cfDNA screening to other prenatal testing options.

The prenatal cfDNA screening module is the second in the Precision Medicine for Your Practice education series. Genetic testing is a key element of precision medicine, a tailored approach to health care that accounts for the individual variability in the genes, environment and lifestyle of each person.

The first module, covering expanded carrier screening, was released in July. Future topics will include precision medicine and its applications in oncology, neurology and cardiology.

Find out more about precision medicine

- Precision Medicine For Your Practice: Expanded Carrier Screening module
- The Precision Medicine Initiative: Report of the AMA Council on Science and Public Health
- What is precision medicine?
- Personalized medicine resources for physicians


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