# Prenatal genetic carrier screening: How much do you know?

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As prenatal genetic screening options expand, physicians face questions about which screening is best for individual patients. Test your knowledge about new expanded carrier screening and what role it can play in clinical practice.

## Casting a wider net

Genomics is part of precision medicine, the science of crafting treatments to fit a patient's particular genes, environment and lifestyle. Unlike traditional one-size-fits-all treatments, precision medicine tools enable clinicians to better understand a patient's health and conditions, and better determine what treatments will be most effective for that particular person.

Carrier screening provides information about reproductive risks by identifying genetic variations in parents that usually do not affect their own health but could result in diseases in their children. Results allow patients to consider their reproductive options.

Traditional prenatal screening detects about a dozen conditions that are more prevalent in certain ethnicities. In contrast, new expanded carrier screening can test for more than 100 genetic conditions and isn't limited to only certain ethnicities.

As screening becomes available to more patients, physicians should consider additional factors and discuss them with patients before and after screening. For example, testing for more diseases, especially those that are less common, can lead to uncertainty about residual risks and clinical outcomes when data on those diseases is limited.

## Learn more about carrier screening

Extensive information on working with expanded screening can be found in a new continuing medical education (CME) module. It is the first of 12 modules the AMA is creating in partnership with Scripps Translational Science Institute and The Jackson Laboratory on the benefits and limitations of genetic testing and how and when it is appropriate to incorporate it into patient care.

In this first module, clinicians learn more about how to determine who is a good candidate for expanded carrier screening. The module includes patient scenarios illustrating issues that clinicians face as they weigh the merits of expanded carrier screening.

## Test your knowledge

Test your understanding of expanded carrier screening issues in clinical practice by considering three patient scenarios:

**Scenario 1:** Sasha and Eli are planning to start a family soon and have been advised by their rabbi to consider preconception screening for “Jewish diseases.” They want to rule out as many serious disorders as possible before pregnancy.

**Scenario 2:** Shonda is in her first pregnancy. She doesn’t know her family medical history or ethnicity but wants to know if her baby might have a treatable condition.

**Scenario 3:** Martha is of Southeast Asian descent and is 16 weeks pregnant. The father of the baby is not involved. She is highly anxious about her ability to raise a child with special needs as a single mother.

How would you answer questions for each of the three scenarios? They include:

> Is the patient more suited to expanded carrier screening or ethnicity-based screening?
> What is the impact if just one parent is a genetic carrier?
> What steps should be taken if screening reveals that one or both prospective parents are carriers or have disease risk themselves?
> What information is important to discuss with the patient before carrier screening?
> What is the best option if the father of the pregnancy is not available for testing?

The CME module on Expanded Carrier Screening offers answers to these questions and more. The module breaks down into three parts: A video overview of the topic, an opportunity to practice applying key points to real-world patient cases and referral to additional information for those who want to dig deeper into expanded carrier screening.

Additional CME genomics modules will follow. Module 2, expected to launch later this summer, addresses prenatal cell-free DNA screening, sometimes referred to as non-invasive prenatal screening. Other topics will include precision medicine and its applications in oncology, neurology and cardiology.

## Find out more about precision medicine

- The Precision Medicine Initiative: Report of the AMA Council on Science and Public Health
- What is precision medicine?
- Personalized medicine resources for physicians