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ACOG Recommends Expanded Genetic Carrier Screening

Ricki Lewis, PhD | February 27, 2017

Two Committee Opinions from the American College of Obstetricians and Gynecologists (ACOG) published in the March 2017 issue of *Obstetrics & Gynecology* expand guidelines on carrier screening for genetic disorders.

The recommendations are in response to lowered costs of testing for many conditions at once and increased genetic admixture that has diluted the ethnic population concentrations that have guided previous screening recommendations.

Committee Opinion 690, "Carrier Screening in the Age of Genomic Medicine," offers general guidelines, and Committee Opinion 691, "Carrier Screening for Genetic Conditions," addresses testing for specific diseases. Carrier screening tests asymptomatic individuals for single copies of specific mutations known to cause disease when present in two copies or in a male.

The opinions distinguish three scopes of genetic screening:

- ethnic-specific, such as for Tay-Sachs disease among people of Ashkenazi Jewish descent;
- panethnic (for everyone), such as a test for cystic fibrosis, spinal muscular atrophy, and fragile X syndrome offered to all patients; and
- expanded carrier screening, which analyzes up to hundreds of conditions.

The general recommendation urges individual healthcare providers to "establish a standard approach" they offer consistently to patients, including counseling and informed consent. Counseling should include discussion of "residual risk" resulting from de novo mutations and mutations not included in test panels. Minimally, all patients should be offered screening for cystic fibrosis, spinal muscular atrophy, and hemoglobinopathies because these are the more common recessive inherited conditions.

Preconception carrier testing enables couples to identify mutations they both carry. If they conceive, they can follow-up with prenatal diagnosis to identify inheritance of a disease genotype. Or they can use donor gametes or preimplantation genetic diagnosis to eliminate risk. Carrier testing is particularly valuable for consanguineous couples, whose offspring are at elevated risk of inheriting recessive mutations from shared ancestors. Prenatal carrier testing provides information for diagnostic testing of the fetus or newborn, termination, or arranging care.

Patients with a family history of a specific genetic disorder should be offered carrier testing for the family's specific mutation if it is not part of the disease panel the practitioner uses.

According to the committee opinions, a genetic disorder should be part of a carrier test panel if it:

- has a carrier frequency of 1/100 or greater;
- causes physical or cognitive impairment requiring medical or surgical intervention; and
- can be diagnosed prenatally and the information used to optimize pregnancy, labor, and childbirth.

Test panels should not include conditions with adult onset.

The opinions note that more than half of patients taking many-gene panels will learn that they carry something. Obstetrician-gynecologists can work with genetic testing companies to tailor panels to suit the particular patient

population.

"Ultimately, the goal of genetic screening is to provide individuals with meaningful information that they can use to guide pregnancy planning based on their personal values," the opinion "Carrier Screening in the Age of Genomic Medicine" concludes.

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