Prenatal Carrier Screening

Introduction

Carrier screening is genetic testing that determines whether an asymptomatic person has a genetic mutation or abnormalities associated with a particular disorder that may be passed on to children. **The goal of genetic carrier screening is to provide individuals with meaningful information that they can use to guide pregnancy planning based on their personal values.** In recognition of how critical genetic testing is in preparing for and managing a successful pregnancy, The American College of Obstetricians and Gynecologists (ACOG) has expanded guidelines on carrier screening in two new Committee Opinions released February and March 2017.

There are two approaches to screening for carrier disorders: traditional carrier screening and expanded carrier screening. Both approaches share the same objective: - to inform couples of their risks so that they may consider their reproductive options.

In the past, ACOG recommended carrier screening based primarily on ethnicity. The focus was on specific ethnic populations with known increased risk for particular disorders. ACOG's two new Committee Opinions go beyond previous guidance to broaden who should be screened and for which genetic disorders.

Summary Of General Recommendations *ACOG Committee Opinion 690 & 691*

- **Ethnic-Specific, Pan-Ethnic, and Expanded Carrier Screening are acceptable screening strategies** pre-conception or during pregnancy.
- Each healthcare provider or practice should establish a **standard approach.**
- After counseling on test benefits, limitations, and alternatives, **a patient may choose to decline any or all carrier screening.**
- Carrier screening for **cystic fibrosis, spinal muscular atrophy, thalessemias and hemoglobinopathies, along with a complete blood count should be offered to all patients** who are pregnant or considering becoming pregnant regardless of ethnicity or screening strategy.
- **Prenatal Carrier Screening does not replace newborn screening,** nor does newborn screening diminish the benefit of prenatal carrier screening.
- **If an individual is found to be a carrier for a specific condition, the individual's reproductive partner should be offered carrier testing.** Concurrent screening of patient and her partner is suggested if there are time constraints for decisions about prenatal diagnostic evaluation.
- **If both partners are found to be carriers, genetic counseling is encouraged** so that reproductive options such as Preimplantation genetic diagnosis, prenatal diagnosis or donor gametes can be discussed.

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- **When an individual is found to be a carrier for a certain genetic condition, the individual's relatives are at risk of carrying the same mutation.** The patient should be encouraged to inform their relatives of the risk and availability of carrier
- **Family history should be obtained.** Individuals with a family history of a genetic disorder may benefit from identification of specific familial mutation(s). Knowledge of specific familial mutation(s) may allow for more specific and rapid prenatal diagnosis.
- **Carrier screening for a particular condition should be done once a person's lifetime** and results documented in the patient's chart. Additional mutations may be added to newer screening panels due to the rapid evolution of technology. The incremental benefit of repeat testing should be assessed before rescreening a patient.
- **The cost of carrier screening for individual conditions may be higher than cost of testing of using expanded disease panels.** Cost of each option should be discusses with each patient.

"Genetic conditions, including SMA, are not limited to one ethnic group and certain conditions are common enough that it's essential offer screening for them in every patient. A growing number of Americans are also of mixed or uncertain ethnic backgrounds, which means we may not identify some people who are at risk of passing genetic conditions to their children when we follow ethnic-based recommendations." *ACOG Committee Opinion author Joseph R. Biggio Jr., MD, Director of the Division of Maternal Fetal Medicine at the University of Alabama at Birmingham.*

Otogenetics Carrier Screening Panels

Basic 5 with CF Alpha-Thalessemia, Beta Hemoglobinopathies (Beta-Thalassemia and Sickle Cell), Cystic Fibrosis, Duchenne/Becker Muscular Dystrophy, Spinal Muscular Atrophy

ACOG/ACMG 13 with CF Alpha-Thalessemia, Beta Hemoglobinopathies (Beta-Thalassemia and Sickle Cell), Bloom Syndrome, Canavan Disease, Cystic Fibrosis, Duchenne/Becker Muscular Dystrophy, Familial Dysautonomia, Fanconi Anaemia, Gaucher Disease, Mucolipidosis IV, Niemann-Pick Disease, Types A and B, Spinal Muscular Atrophy, Tay-Sachs Disease

Ashkenazi Jewish Panel screens for 38 diseases

Pan-Ethnic Panel screens for 167 diseases

Panel Criteria

Conditions included in our Expanded Carrier Screening panel meet the following criteria:

- carrier frequency of one in 100 or greater
- well-defined phenotype
- have detrimental effect on quality of life
- cause cognitive or physical impairment,
- require surgical or medical intervention
- have an onset early in life
- diagnosed prenatally
- potential opportunities for antenatal intervention to improve perinatal outcomes
- changes to delivery management to optimize newborn and infant outcomes
- parental education about special care needs after birth