Expanded Genetic Carrier Screening

Palmetto Health USC
OBSTETRICS AND GYNECOLOGY
GENETIC COUNSELING

What is expanded genetic carrier screening?

Traditional genetic carrier screening has largely focused on screening individuals for conditions that are more common in their ethnic group or ancestry in order to identify couples at risk for passing on genetic conditions to their offspring. For example, 1 in 10 African Americans are carriers of sickle cell disease; therefore, sickle cell screening is routinely offered to individuals of African American ancestry. Caucasians have a 1 in 25 chance to be carriers of cystic fibrosis, while individuals with other ethnic backgrounds have lower chances. There are a number of genetic conditions in which individuals of Ashkenazi Jewish ancestry have increased carrier chances. Expanded carrier screening simultaneously screens for common ancestry-based diseases in addition to a number of other genetic conditions. The list of conditions screened, known as a "panel," varies between laboratories and can include more than 100 genetic conditions. Expanded carrier screening panels may include rare conditions, conditions for which effective treatment may or may not be available, and/or conditions with variable presentations including age of onset.

What types of conditions are screened for?

Expanded panels screen for genetic conditions with a wide range of severity and age of onset. Many of the conditions are associated with significant adverse outcomes such as cognitive impairment, decreased life expectance, and need for medical or surgical treatment. Some conditions included in expanded carrier screening have no effective medical treatment. Because most of the conditions are rare, it is not uncommon for carrier frequencies to be unknown in some populations.

Not all of the conditions are inherited in the same way. Most of the conditions are autosomal recessive, which means both a mother and father must be carriers of a specific condition for there to be a 25% chance (1 in 4) of having a child affected with that condition. Some of the conditions are X-linked, and typically only females are carriers of these conditions. If a woman is a carrier of an X-linked condition, she has a 50% chance (1 in 2) of passing it on. If her child is a girl, she has a 50% chance of being a carrier. If the child is a boy, he has a 50% chance of being affected with the condition.

What happens if I am a carrier?

A carrier is someone who has a change in one copy of a gene pair. A change in a gene may be called "trait," "variant" or "mutation." A positive result from this screening test means that a change was identified in a gene(s) screened, and the individual is considered to be a carrier for that condition(s). Expanded carrier screening includes a large number of disorders; therefore, it is common to identify carriers of one or more conditions. Carriers are typically healthy individuals because the other copy of their gene works properly. Carriers do, however, have a chance of passing on their trait. Genetic counseling is recommended for the patient and their partner to review the condition and the chances for children to be affected with that condition.

When a patient is identified as a carrier of an autosomal recessive condition, it is recommended that we screen his or her partner. When each parent is a carrier of the same autosomal recessive condition, a 25% chance exists in each pregnancy for an affected child. If one or both parents screen negative for a trait, their offspring have a decreased chance to be affected with the conditions. It is important to realize that some carriers may be undetected by screening.

When a woman is identified as a carrier of an X-linked condition, she has a 50% chance to have a carrier daughter, and a 50% chance to have an affected son.

Couples that are identified to have an increased chance for an affected child with either an autosomal recessive condition or an X-linked condition may consider testing in pregnancy by CVS or amniocentesis to determine if the condition is present in the baby. Couples may also choose to test a baby at birth or may consider alternative parenting options such as adoption, egg or sperm donation, or preimplantation genetic diagnosis. Specific testing options may vary based on the condition involved.

2 Medical Park Rd.
Suite 103
Columbia, SC 29203
803-545-5775 phone
803-434-4596 fax

PHUSCMG.org

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What does a negative result mean?

A negative result means that there were no changes identified in the gene(s) screened for and the chance of being a carrier is significantly lowered. A negative screen does not eliminate risk to offspring because screening cannot detect all disease-causing changes in a gene.

Should I have expanded genetic carrier screening?

The decision to pursue screening is a personal one. Some women/couples want to know if their chance to have a child with a genetic condition is increased prior to or during pregnancy. Other women/couples do not wish to screen for a large number of conditions but would rather screen for conditions they are at risk for based on ancestry. Talk with your doctor and/or arrange to speak with a genetic counselor for more information.