July 2017 | Members

Spotlight on Health

Expanded Carrier Screening

If you or your partner is pregnant, or planning a pregnancy, you may have thought about inherited disorders. Inherited disorders are conditions that children can get from their parents. About 4 in every 10,000 children are born with an inherited disorder.[1] Many of these conditions are "recessive." Parents can be "carriers" of a recessive disorder without actually having it. "Carrier screening" can help a couple understand their risk for having a child with an inherited disorder.

This newsletter discusses carrier screening. And how new guidelines can help guide your healthcare provider when working with you on family planning.

What Is a Carrier?

To understand what a carrier is, we first need to understand a little about genes and variants (sometimes called mutations). Genes are pieces of DNA that determine things like the color of hair and eyes. You have two copies of each gene: one from your mother and one from your father. Changes in genes are called variants. Like genes, variants can be inherited from parents. Getting a variant from both parents can cause some medical conditions. These types of conditions are called recessive conditions.

If a person has a variant in one copy of a gene for a recessive condition, the person is considered a "carrier". Carriers of genetic disorders usually do not have any signs or symptoms of the condition, and most don't know they are carriers. For example, it is estimated that over 10 million Americans are carriers for cystic fibrosis.[2] And most are probably not aware of their carrier status.

If only you or your partner is a carrier of a recessive condition, then your child is not at high risk of getting the condition. However, if you and your partner are both carriers for the same recessive condition, then your child has a 25% chance of getting it.

Carrier Screening

If you are pregnant, or planning a pregnancy, you may want to find out if you are a carrier for certain inherited disorders. Doctors, medical organizations, and patient advocacy organizations recommend carrier screening. Screening before a pregnancy is ideal. But screening can take place during pregnancy as well.[3]

Carrier screening used to focus just on family history and ethnicity. Some conditions are more common in certain ethnicities.[4] For example, Caucasians of European and Ashkenazi Jewish heritage have a greater chance to be a carrier of cystic fibrosis (CF). Asians and African Americans are less likely to be CF carriers.

How Expanded Carrier Screening Works with Couples

1. The mother is screened first to see if she is the carrier of a condition.
2. The father is screened next, but only for the conditions the mother tests positive for.
3. If mother and father both test positive for the same condition, the fetus can be tested.

But they have a greater chance to be carriers of other genetic conditions. However, ethnicity-based screening can overlook important insights.[5]

Inherited conditions can affect people from all racial or ethnic groups. In addition, the United States has an ethnically diverse population. Individuals may have a mixed ethnicity, or they may not know their background. Or, they may even be incorrect about their background. To address this issue, *expanded* carrier screening is now available. With this method, many different conditions are screened for at once, regardless of racial or ethnic background.

Counseling

It is important to know your carrier status if you are pregnant or planning a family. But there is a lot of information to consider. Many genetic tests are available, but not all of them are useful to everyone. Fortunately, specially trained doctors and counselors are available to help. They can advise based on established guidelines.[6]

These healthcare professionals can also help you understand what a screen can and cannot tell you. By working with an expert, individuals and couples can make well-informed personal decisions.

How the Laboratory Can Help

Quest Diagnostics offers the QHerit™ Expanded Carrier Screen. This pan-ethnic screening panel for select genetic conditions is based on guidelines and recommendations.[7] Quest also offers ethnicity-based screening options, as well as a variety of screening tests for individual conditions.

Additional Information

For more information, visit QuestDiagnostics.com/home/patients/tests-a-z/prenatal.html or these helpful websites:

- Genetics Home Reference: ghr.nlm.nih.gov/primer#
- March of Dimes: marchofdimes.org/
- Genetic and Rare Disease Information Center: rareconditions.info.nih.gov/
- National Society of Genetic Counselors: aboutgeneticcounselors.com/

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