Carrier Identification

What does it mean to be a genetic carrier?

What is a "carrier?"

Sometimes, when a baby's newborn screen is out-of-range for a condition, the follow-up testing shows that he or she is a ***carrier*** of the condition. This means that he or she carries ***one*** genetic variant associated with the disorder. Genetic carriers are not going to have symptoms of the disorder.

Most of the conditions identified through newborn screening are inherited in an autosomal recessive pattern. For these medical conditions, a person needs to have ***two*** copies of the disease gene variant in order to have the diagnosis.

If a child only inherits one disease gene variant, he or she is considered a "carrier" of the condition and has not been diagnosed with the disorder.

KEY POINTS:

- **Being a "carrier" is not the same as being diagnosed with a condition.**
- **No treatment or dietary changes are needed.**
- **No additional follow up is needed for the newborn screening process.**

Is My Child Really Healthy?

In recessive conditions, a single non-working gene is not enough to cause the serious symptoms that are typically associated with a condition. In fact, most carriers are healthy and do not even know they have a non-working copy of the gene.

What Does This Mean For My Family?

People inherit two copies of each gene. We inherit one copy from our Mother and one copy from our Father.

When a child is identified as a carrier of a genetic condition, it is possible that one or both parents are carriers of the condition, as well. Therefore, the parents may want to have genetic counseling and testing to see if they also carry the gene. Some couples may want to know this information if they plan on having more children in the future, or to share with other family members who may also be planning on having children.

What is the chance of passing on the disease gene variant?

When one parent is a carrier of a disease gene variant and the other parent has 2 working genes:

- There is a 1 in 2 chance (50%) of having a child with 2 working genes.
- There is a 1 in 2 chance (50%) of having a child who is a carrier (1 working copy and 1 non-working copy of the gene).

When both parents are carriers of a recessive genetic condition:

- There is a 1 in 4 chance (25%) of having a child who will have the disorder because they inherited two copies of the disease gene variant.
- There is a 1 in 4 chance (25%) of having a child with 2 working genes.
- There is a 1 in 2 chance (50%) of having a child who is a carrier (1 working copy and 1 non-working copy of the gene).

Depending on the carrier status of the parents, siblings may have inherited a non-working copy of the gene, as well. They may wish to pursue carrier testing when they reach reproductive age to determine their own chance of having a child with a genetic condition.

In addition, your child that was recently identified as a carrier may benefit from speaking to a prenatal genetic counselor when they reach reproductive age and are planning to start their own family.

What If I Still Have Questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30 am -4:30 pm. We also recommend the website www.babysfirsttest.org as an accurate and informative resource.

Adapted from www.babysfirsttest.org.

This fact sheet was written for information purposes only. It should not replace medical advice, diagnosis or treatment.