

# Genetic Carrier Screening



At Valley Medical Center, we partner with our patients to provide up-to-date information and options for medical care. After reading this information about genetic carrier screening, you may have more questions for your medical provider. Your provider may also refer you to a genetic counselor for further discussion.

## We all carry one or more genetic conditions

We typically inherit one copy of genes from our mother and one from our father. Carrier status describes the state where one of our genes does not work properly, while the other one does. When a gene isn't working, we may not know because there are often no outward signs or symptoms. In fact, carrier status may be passed on and on in a family without anyone being aware of it.

**Current medical guidelines recommend offering genetic carrier screening to all couples who are pregnant or those considering pregnancy.**

### What is genetic carrier screening?

This is a blood test which screens people to see if they may carry one or more conditions. Historically, carrier screening has been used for disorders more common in specific population groups (examples: cystic fibrosis in European Caucasians; sickle cell trait in African Americans; Tay-Sachs disease in the Ashkenazi Jewish population). More recently, “expanded carrier screening” is frequently used: It screens for a group of disorders all at once and isn't focused on certain population groups. For most of the screened conditions, concerns for the pregnancy only arise if both parents are found to carry the condition—this would mean the baby is at risk to inherit two copies of the gene which don't work, one from the mother and one from the father. *(continued)*

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If you pursue carrier screening and your test result is positive, your partner may be recommended to have carrier screening for the same condition to see if you both carry it.

## Why would I consider genetic carrier screening?

Some people choose carrier screening to help decide whether to conceive or continue a pregnancy. Others choose carrier screening for planning purposes so that they can learn more about the risk for genetic conditions before the baby's birth. Prenatal testing is usually available if the pregnancy is determined to be at risk for a condition. While some conditions would be detected at birth with the Washington State Newborn Screen, others are not. If a condition is diagnosed early, early treatment or therapy may improve the outlook, and could even save a child's life.

### Limitations

You can choose whether to limit genetic carrier screening to a certain condition, or whether to have "expanded carrier screening" for a greater variety of conditions all at once. If you screen positive to carry a condition, genetic counseling and screening your partner will probably be recommended.

If you screen negative to carry a condition, this means it is very unlikely that you carry it, but not impossible. Carrier screening is limited by:

- The number of disorders you test for
- The number of gene changes ("mutations") that the laboratory tests for with each condition
- How common or rare a condition is for individuals with your ancestry

Finally, it is important to know that carrier testing is not available for all genetic conditions, and that no test can absolutely guarantee the health of a baby.

### Insurance

All insurance plans are different. If you're interested in genetic carrier screening, you may wish to contact your insurer to ask about coverage. If your insurance asks for codes related to the screening, please check with your provider or genetic counselor.

### Questions

We hope that many of your questions have been answered. Please use the lines below to record your own questions, which can be answered at your next visit with your medical provider, or when you meet with a genetic counselor.

1. My opinion of genetic carrier screening:

- I want to do it.    I don't want to do it.    I have questions (below) to discuss before I decide.

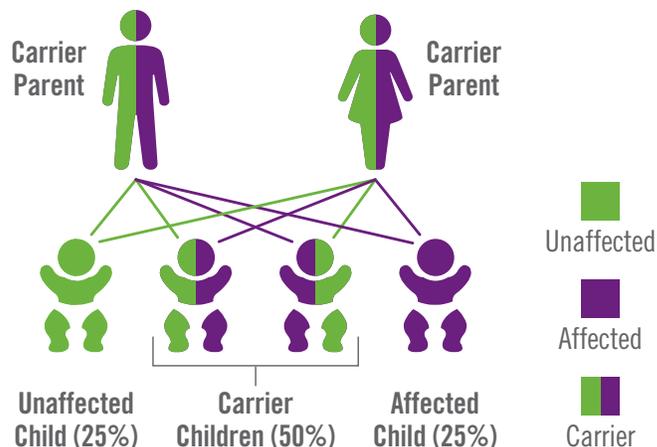
2. My questions about genetic carrier screening:

Q: \_\_\_\_\_

Q: \_\_\_\_\_

Q: \_\_\_\_\_

## Autosomal Recessive Inheritance Pattern



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