Ambry Genetics

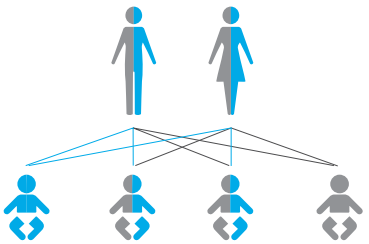
Understanding Your Cystic Fibrosis (CF) Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH A **5T VARIANT**

Result	**GENE VARIANT**	Your testing shows that you have a change (called a variant) in the gene that causes cystic fibrosis (the CFTR gene). This variant, called 5T, is considered a "disease-modifying variant." This means it may cause some signs or symptoms of CF, but the type of these depends on other changes (like a mutation or another variant) that may also be in the CFTR gene.
Gene	**DEFINITION**	Genes have instructions for traits that make us who we are, like eye color and how our bodies work. Everyone has two copies of each gene. We get one copy of each gene from each of our parents. A mutation (change in the gene, like a spelling mistake) in both copies of the CFTR gene can cause CF. Having a single mutation or 5T variant means you are a "carrier" for CF. Carriers do not usually have symptoms of the common form of CF, but can have children with the condition. If you have the 5T variant and a mutation in the CFTR gene, you could have the common type of CF or milder symptoms of CF.
Meaning	**5T VARIANT**	Your result needs to be understood based on your other test results and any medical problems that you might have. If you have symptoms of CF but your genetic test results have not confirmed this, it is possible that you have another medical condition (with similar symptoms) that this test cannot find. Your doctor or genetic counselor will talk to you about this further, including any medical care that might be helpful for you.
Screening Options	**FAMILY MEMBERS**	If your partner is also a CF carrier, you could have a 1 in 4 (25%) chance to have a child with CF in each pregnancy together (see below). CF genetic testing for your partner may help you both learn more about this. In addition, your adult family members may wish to be tested to see if they carry the 5T variant found in your family.
Next Steps	**DISCUSS**	Please discuss your results with your doctor or genetic counselor. They can tell you about more genetic testing and/or medical care that might be right for you. Also, please share this with family members so they can talk with their doctors and learn more. They can now be tested for the same variants(s), if they choose to.
Reach Out	**RESOURCES**	• National Society of Genetic Counselors www.nsgc.org • Cystic Fibrosis Foundation www.cff.org • CFTR2 www.cftr2.org • Genetic Information Nondiscrimination Act (GINA) www.ginahelp.org

How Cystic Fibrosis Inherited

People who carry a mutation in only one copy of their CF gene are called "carriers," and do not usually have symptoms. If their partner is also a CF carrier, there is a 1 in 4 (25%) chance for them to have a son or daughter with CF in each pregnancy together. There is a 2 in 4 (50%) chance for them to have a child who is a CF carrier (usually without symptoms), and a 1 in 4 (25%) chance for them to have a child who does not have CF, and is not a CF carrier.



CF carrier (one gene mutation)

Does not have CF, and is not a CF carrier (no gene mutations)

Has CF (two gene mutations)

Please talk with your doctor or genetic counselor about this. The field of genetics is continuously changing, so updates related to your result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or taken as medical advice.

7 Argonaut, Aliso Viejo, CA 92656 USA Toll Free +1 866.262.7943 Fax +1 949.900.5501 ambrygen.com