Ambry Genetics

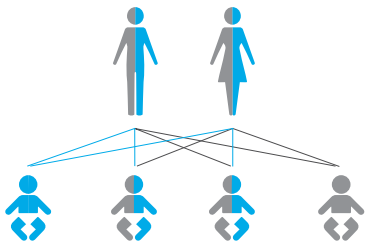
Understanding Your Cystic Fibrosis (CF) Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH **ONE PATHOGENIC MUTATION** OR **VARIANT THAT IS LIKELY PATHOGENIC**

Result	**CARRIER**	Your testing shows that you have one pathogenic (disease-causing)mutation, or a variant that is likely pathogenic, in the gene that causes CF (the CFTR gene). Both of these should be treated as the same type of result. This means you are what is called a "carrier" for CF. It is not likely that you will have symptoms of the common type of CF, but some carriers may have mild symptoms.
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. Carriers have one mutation in a copy of their CFTR gene. CF carriers do not usually have the symptoms of the common form of CF, but can have children with the condition.
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 mutation(s) found in your family.
Next Steps	**DISCUSS**	Please share this with family members so they can talk with their doctors and learn more. They can now be tested for the same mutation(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