Ambry Genetics®

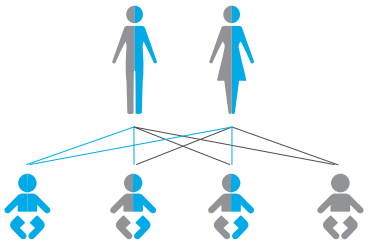
Understanding Your Positive Cystic Fibrosis (CF) Genetic Test Result

INFORMATION FOR PATIENTS WITH **PATHOGENIC MUTATIONS** OR **VARIANTS THAT ARE LIKELY PATHOGENIC**

Result	**POSITIVE**	Your testing shows that you have a combination of two pathogenic or likely pathogenic (disease causing) mutations in the gene that causes CF (the *CFTR* gene). If these two mutations are on different copies of the gene (each inherited from a different parent), this result is consistent with CF or a *CFTR*-related disorder. Testing of other family members, especially parents, can be helpful in finding out if these mutations are on the same or different copy of the 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.
Diagnosis	**CF**	People with this result have CF, a disease with a broad range of severity, which typically affects the lungs and digestive systems of the body.
Management Options	**FOR PATIENTS WITH CF**	Treatment options may include: medications, inhalers, chest physical therapy, antibiotics, and other options to decrease the risk of infection. Talk to your doctor about which options may be right for you.
Screening Options	**FAMILY MEMBERS**	People who carry one mutation in their *CFTR* genes are called "carriers." While most carriers do not have symptoms of CF, sometimes carriers may have mild symptoms of CF. If their partner is also a carrier, the couple has a 1 in 4 (25%) chance to have a child with CF in each pregnancy together. 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.

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