Ambry Genetics®

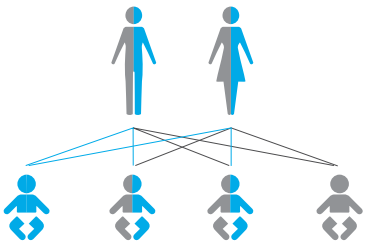
Understanding Your Negative Cystic Fibrosis (CF) Genetic Test Result

INFORMATION FOR PATIENTS

Result	**NEGATIVE**	Your testing did not find any disease-causing changes (mutations) in the gene that causes cystic fibrosis (the *CFTR* gene). Depending on the test you had, this means that you likely do not have cystic fibrosis (CF), and are likely not a carrier for CF.
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. People who carry one mutation in their *CFTR* gene are called "carriers." While most carriers do not have symptoms of the most common type CF, 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 (see below).
Diagnosis	**NO CHANGE**	It is possible that you may have another type of condition 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.
Next Steps	**DISCUSS**	More genetic testing may be right for you. Please talk about this with your doctor or genetic counselor.
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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