Ambry Genetics®

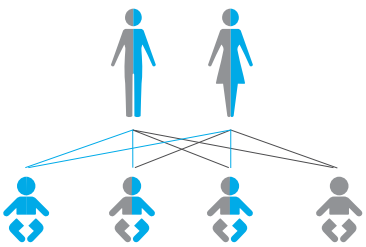
Understanding Your VUS Cystic Fibrosis (CF) Genetic Test Result

INFORMATION FOR PATIENTS WITH **VARIANTS OF UNKNOWN SIGNIFICANCE**

Result	**VUS**	Your testing shows that you have one or more variants of unknown significance (VUS) in the gene that causes cystic fibrosis (the *CFTR* gene). This result may be consistent with cystic fibrosis (CF) or a *CFTR*-related disorder, or being a carrier for either. A VUS is a gene change, but we do not know if it causes CF or not.
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. Mutations (or changes, like a spelling mistake) in a gene can cause it to not work properly and lead to medical problems (like CF). However, some changes in a gene (like a VUS) are not well understood, so we cannot predict if they will cause CF or not.
Diagnosis	**UNCERTAIN**	It is possible that you have CF or are a CF carrier. If you have symptoms of CF but genetic testing is not able to confirm this, you may also have another 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**	Testing for your family members may help explain this VUS. Talk with your doctor or genetic counselor about which family members may be helpful to test.
	FOR YOU	More genetic testing may be right for you. Please talk about this with your doctor or genetic counselor.
Next Steps	**DISCUSS**	Please discuss these results with your doctor or genetic counselor. They can tell you about any additional testing and/or medical care that might be right for you.
Reach Out	**RESOURCES**	• National Society of Genetic Counselors nsgc.org • Cystic Fibrosis Foundation cff.org • CFTR2 cftr2.org • Genetic Information Nondiscrimination Act (GINA) 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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