Ambry Genetics®
A Konica Minolta Company

Understanding Your *CFTR* Genetic Test Result

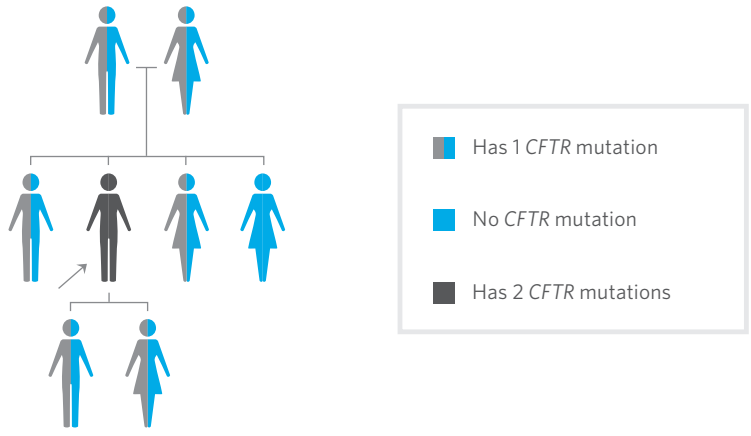
INFORMATION FOR PATIENTS WITH **TWO PATHOGENIC MUTATIONS** OR **VARIANTS, LIKELY PATHOGENIC**

5 Things To Know

1	*CFTR* mutation	Your testing shows that you have two pathogenic mutations or variants that are likely pathogenic in the *CFTR* gene.
2	Cystic fibrosis (CF*) or CFTR-related disorder	People with two *CFTR* mutations have CF or a *CFTR*-related disorder such as pancreatitis.
3	Cancer risks and other medical concerns	Cancer risks associated with *CFTR* mutations are not well defined. Individuals with pancreatitis may be at an increased risk for pancreatic cancer.
4	What you can do	There may be personalized risk management options available. It is important to discuss these options with your doctor, and decide on a plan that best manages your risks.
5	Family	This information may also be important for your family members – they can be tested for the *CFTR* mutations that were identified in you.

CFTR Mutations in the Family

You have two *CFTR* mutations, therefore, any children you have will inherit one of them. Your children should not be at risk to have CF* or a *CFTR*-related disorder unless your partner has at least one *CFTR* mutation as well. Each of your parents carries at least one *CFTR* mutation. This means your brothers and sisters have a 25% chance of also having CF or a *CFTR*-related disorder, a 50% chance to inherit one *CFTR* mutation, and a 25% chance to inherit **no** *CFTR* mutations. The image below shows that both men and women can carry and pass on these mutations.



*CF is characterized by chronic lung disease, pancreatic insufficiency, and high sweat chloride levels.

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Result	**MUTATIONS**	Your testing shows that you have two pathogenic mutations (disease-causing changes in the gene, like a spelling mistake) or variants that are likely pathogenic in the *CFTR* gene. This result should be considered positive for cystic fibrosis (CF) or a *CFTR*-related disorder.
Gene	***CFTR***	Everyone has two copies of the *CFTR* gene, which we randomly inherit from each of our parents. This means you inherited one of your mutations from each of your parents. Having two mutations in the *CFTR* gene is associated with having CF or a *CFTR*-related disorder such as pancreatitis.
Other Medical Concerns	**MAY BE PRESENT**	Individuals who carry two *CFTR* mutations are at risk to develop CF or a *CFTR*-related disorder such as acute recurrent or chronic pancreatitis or congenital absence of the vas deferens.
Cancer Risks	**VARIES**	Cancer risks associated with *CFTR* mutations are not well defined. Individuals with pancreatitis may be at an increased risk for pancreatic cancer.
Management Options	**FOR PANCREATITIS**	Lifestyle modifications to reduce the chance of pancreatitis may include: smoking cessation and low-fat diet. Talk to your doctor about what options may be right for you.
Management Options	**FOR PANCREATIC CANCER**	Options for screening and early detection may be available. There are no standard recommendations for pancreatic cancer screening currently available, so it is recommended that you talk to your doctor about what options may be right for you.
Risk Management	**VARIES**	Risk management decisions are very personal, and the best option depends on many factors. Screening typically begins earlier than the general population and is often more frequently performed. It is important to discuss these options with your doctor.
Family Members	**25-50% CHANCE**	Your parents each carry at least one *CFTR* mutation. Since you have two *CFTR* mutations (one from each parent), each of your children will inherit one of them and will be considered *CFTR* carriers. Your children should not be at risk to have CF or a *CFTR*-related disorder unless your partner has at least one CFTR mutation as well. If your partner has one *CFTR* mutation, your children each have a 50% chance to have CF or a *CFTR*-related disorder (have 2 *CFTR* mutations). Your brothers and sisters have a 25% chance of also having CF or a *CFTR*-related disorder and a 50% chance to be a carrier. Other close family members (like your aunts, uncles, cousins) may also be carriers. Talk to your healthcare provider about your family history to find out if other family members may benefit from genetic testing.
Next Steps	**DISCUSS**	It is recommended that you share this information with your family members so they can learn more and discuss this with their healthcare providers.
Reach Out	**RESOURCES**	• National Society of Genetic Counselors nsgc.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

Please discuss this information with you healthcare provider. The genetics field is constantly evolving, so updates related to your *CFTR* 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 interpreted as medical advice.

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