Ambry Genetics®

Understanding Your *CFTR* Carrier Genetic Test Result

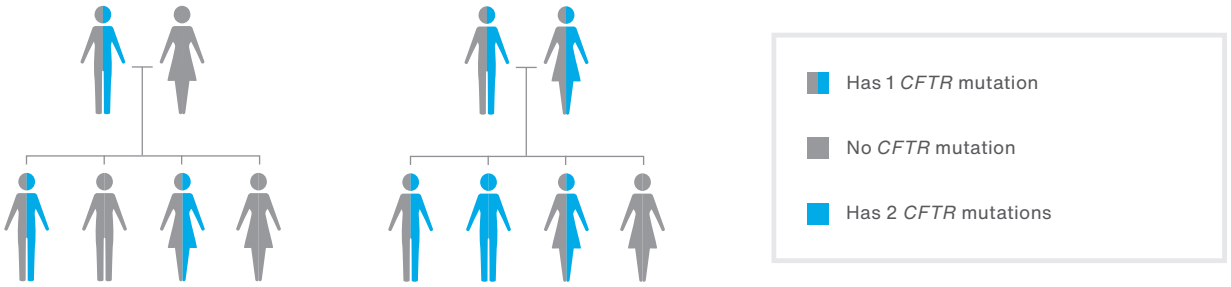
INFORMATION FOR PATIENTS WITH ONE PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things To Know

1	*CFTR* mutation	Your testing shows that you have ONE pathogenic mutation or variant that is likely pathogenic in the *CFTR* gene.
2	Carrier	People with one *CFTR* mutation are carriers of cystic fibrosis and typically do not have cystic fibrosis. People with two *CFTR* mutations have cystic fibrosis (CF*) or a *CFTR*-related disorder. Your result shows you do not have cystic fibrosis, but your family members may be at risk for it. One mutation in the *CFTR* gene does not cause CF, but may slightly increase your risk to develop pancreatitis.
3	Cancer risks and other medical concerns	Cancer risks associated with being a *CFTR* carrier are not significantly increased compared to the general population. Individuals with only one *CFTR* mutation may have a slightly increased risk to develop pancreatitis, bronchiectasis (a chronic lung condition), or have male infertility. However, most *CFTR* carriers will not have any of these medical concerns.
4	What you can do	There may be personalized risk management options available. It is important to discuss these options with your healthcare provider, and decide on a plan that best manages your risks.
5	Family	Family members may be at risk- they can be tested for the *CFTR* mutation that was identified in you, as well as other mutations in the *CFTR* gene. It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.

CFTR Mutations in the Family

There is a 50/50 random chance to pass on the *CFTR* mutation to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations. If your partner also carries a mutation in *CFTR*, there is an increased risk (25%) to have a child with CF* or a *CFTR*-related disorder.



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

Reach Out

RESOURCES

- National Society of Genetic Counselors nsgc.org
- Genetic Information Nondiscrimination Act (GINA) ginahelp.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. 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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