

Understanding Your *CFTR* Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH ONE PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things To Know

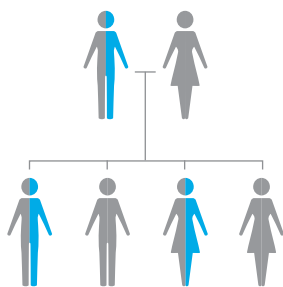
1	Result	Your testing shows that you have one pathogenic or likely pathogenic variant in the <i>CFTR</i> gene.
2	Carrier	People with one pathogenic or likely pathogenic <i>CFTR</i> variant are carriers of cystic fibrosis and typically do not have cystic fibrosis. People with two pathogenic or likely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

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

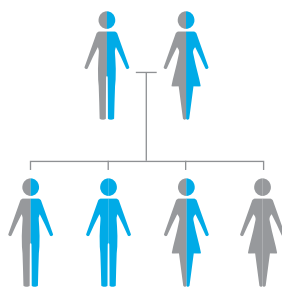
CFTR in the Family

There is a 50/50 random chance to pass on a pathogenic or likely pathogenic *CFTR* variant to each of your children. If your partner also happens to carry one pathogenic or likely pathogenic *CFTR* variant, there is a 25% chance that you will both pass on the *CFTR* variant to your child.

One carrier parent, one non-carrier parent



Two carrier parents



- Has two pathogenic or likely pathogenic *CFTR* variants (cystic fibrosis or related disorder)
- Has one pathogenic or likely pathogenic *CFTR* variant (carrier)
- No pathogenic or likely pathogenic *CFTR* variants

RESOURCES

- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously 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.