Cystic Fibrosis is an inherited condition that affects the lungs, pancreas, liver, digestive system, and male reproductive system. Individuals with cystic fibrosis are unable to properly regulate epithelial chloride and sodium channels, leading to an increased risk for lung infections, digestive problems, diarrhea, poor growth, and male infertility. Long-term disease management is aimed at preventing pulmonary complications and promoting weight gain. Many individuals will also be pancreatic insufficient and therefore require oral pancreatic enzyme replacement. While advances in research and treatment have greatly improved the lives of people with cystic fibrosis, affected individuals still require lifelong medical care.

The clinical features of cystic fibrosis and CFTR-related disorders can be explained by pathogenic variants in the CFTR gene. In general, individuals have two copies of the CFTR gene. Carriers have a single variant in one copy of the CFTR gene while individuals with cystic fibrosis or a CFTR-related disorder typically have variants in both copies of their genes, one inherited from each parent.

What Your Test Results Mean

Carriers of a pathogenic CFTR variant are very common and are not expected to show symptoms of cystic fibrosis or CFTR-related disorders. Because risk for offspring depends on both parents’ carrier status, carrier testing regardless of sex is recommended.

CFTR-Related Disorders Explained

Some CFTR variants do not cause classic cystic fibrosis but do cause CFTR-related disorders. Similar to carriers of cystic fibrosis, individuals who carry a CFTR-related disorder do not have any clinical signs or features of the condition. However, they may have an increased risk to have a child with a CFTR-related disorder should their partner or donor be identified as a carrier. Examples of CFTR-related disorders include an increased risk for pancreatitis, an increased risk for variable pulmonary or lung concerns and/or, specific to males, an increased risk to have congenital absence of the vas deferens (CBAVD), a condition that causes male infertility.

How the Genetics Work

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Questions?

Contact us at 1-855-776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.