Genetics Uncoded:

NxGen MDx

Facts about

Cystic Fibrosis and *CFTR*-Related Disorders

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.

Cystic Fibrosis Explained

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 promote 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.

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

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.

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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

Revised 12/15/16