Cystic Fibrosis and Other *CFTR*-Related Disorders

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of cystic fibrosis or a *CFTR*-related disorder. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of cystic fibrosis or a *CFTR*-related disorder for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for cystic fibrosis or a *CFTR*-related disorder, each of your children has a 1 in 4 (25%) chance to have the condition.

Cystic Fibrosis and Other *CFTR*-Related Disorders Explained

What is Cystic Fibrosis?

Cystic fibrosis is an inherited condition that primarily affects the pulmonary and digestive systems. Most individuals with cystic fibrosis are diagnosed in childhood. Individuals with cystic fibrosis are unable to properly regulate epithelial chloride and sodium channels, leading to a buildup of mucus. This buildup of mucus increases the risk for lung infections and digestive problems. Additional signs and symptoms can include diarrhea, malnutrition, and poor growth. Men with cystic fibrosis are usually infertile because many do not have the vas deferens, an important organ responsible for transporting sperm. These men may be able to have children using assisted reproductive methods.

What is a *CFTR*-Related Disorder?

Some changes in the *CFTR* gene do not cause cystic fibrosis but instead cause a *CFTR*-related disorder. Examples of *CFTR*-related disorders include an increased risk for pancreatitis, an increased risk for pulmonary or lung problems, and an increased risk for a condition that causes male infertility known as congenital absence of the vas deferens.

Prognosis

Prognosis for cystic fibrosis is considered fair. While advances in research and treatment have greatly improved survival and quality of life, affected individuals still require lifelong medical care. The average expected lifespan for adults is approximately 35 years old. The prognosis for individuals with *CFTR*-related disorders can vary, as symptoms can be mild.

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Treatment

Treatment for cystic fibrosis involves long-term disease management that aims to prevent pulmonary complications and promote weight gain. Many individuals are pancreatic insufficient and therefore require oral pancreatic enzyme replacement. Some individuals may benefit from a lung transplant. Various medications have been introduced in recent years that treat the underlying issues in individuals with cystic fibrosis (ex. Kalydeco®, Trikafta™, Orkambi®). Individuals taking these medications have shown improvement and/or stabilization in lung function. Treatment for individuals with *CFTR*-related disorders can vary, as individuals may only have congenital absence of the vas deferens or pancreatitis.

Resources
Cystic Fibrosis Foundation
https://www.cff.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/cystic-fibrosis/
National Society of Genetic Counselors
https://www.nsgc.org/

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