What Your Test Results Mean

Test results indicate that you are a carrier of cystic fibrosis. Carriers typically show no symptoms of cystic fibrosis; however, carriers are at an increased risk of having a child with cystic fibrosis. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Cystic Fibrosis Explained

Cystic fibrosis is an inherited condition that affects the lungs, pancreas, liver, and male reproductive system. Some males may not develop lung or gastrointestinal symptoms of the disease and will only present with male infertility caused by congenital absence of the vas deferens (CAVD). Both cystic fibrosis and CAVD are caused by abnormalities in the cystic fibrosis transmembrane conductance regulator (CFTR). Individuals with cystic fibrosis are unable to properly regulate epithelial chloride and sodium channels, leading to an increased risk for lung infections, digestion problems, diarrhea, poor growth and male infertility. Management of disease is lifelong to prevent pulmonary complications and enhance weight gain. The majority of individuals with cystic fibrosis will require oral pancreatic enzyme replacement. With advances in treatment and appropriate management, individuals with cystic fibrosis typically live into their late thirties. Individuals with CAVD may conceive children through assisted reproductive technologies.

How the Genetics Work

The clinical features of cystic fibrosis disease can be explained by mutations in the CFTR gene. In general, individuals have two copies of the CFTR gene. Carriers of cystic fibrosis disease have a single mutation in one copy of the CFTR gene while individuals with cystic fibrosis disease have mutations in both copies of their genes, one inherited from each parent.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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.