Genetic Insights: quick reference guide for healthcare providers

This guide is intended to facilitate a discussion between a provider and their patient.

Genetic Insights test results: **Cystic fibrosis - carrier**

Key results

A pathogenic or likely pathogenic variant associated with being a carrier of cystic fibrosis was found in 1 copy of the *CFTR* gene. Carriers of cystic fibrosis do not have the condition but are at higher risk of having children with the condition if the other biological parent is also a carrier.

Next steps

Clinical recommendations	Resources
Genetic Insights is a screening test and not intended for diagnosis or to replace routine carrier screening for family planning or during pregnancy. *A follow-up genetic test should be performed in a clinical setting, especially if biological children are planned. Comprehensive analysis of the CFTR gene in a clinical setting should be considered for reproductive partners or gamete donors.*	**Ready to order?** Check with your institution and/or patient's insurance about the preferred testing laboratory. To confirm this result, Blueprint Genetics® offers **targeted variant testing** for the variant identified. You can order a confirmation test here: **Blueprint Genetics/TVT** **Have questions?** Call **1.866.GENE.INFO** (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare providers to discuss test selection and results.
Refer your patient to a genetic counselor.	Your patient can schedule a 1-on-1 remote genetic counseling session through their online Genetic Insights dashboard at no additional cost.
Genetic counselors can provide counseling on the implications of this test result and next steps for your patient.	To find a genetic counselor practicing in your patient's area for an in-person session, please visit FindAGeneticCounselor.NSGC.org

Patient conversation starters:

Patient conversation starters summarize the preceding information in plain language to support meaningful conversations between you and your patient.

FindAGeneticCounselor.NSGC.org

Your Genetic Insights test is a screening test. If you need to confirm your result, the next step is to have a second genetic test. It's also important that you talk with a genetic counselor. Genetic counselors are experts in genetics and can help you understand this result and potential next steps.

You can access a genetic counselor through your online Genetic Insights dashboard at no additional cost to you, and we can discuss a referral to a local genetic counselor.

Additional resources

The following advocacy groups have additional information and resources about cystic fibrosis:

CF Foundation: CFF.org

American Lung Association: Lung.org

Visit QuestDiagnostics.com/Genetic-Health-Screening for more information about this test.

What is cystic fibrosis?

Cystic fibrosis (CF) is a genetic condition characterized by the build-up of thick mucus in the lungs, pancreas, and other organs leading to multisystem disease.[1] Symptoms, such as chronic coughing, wheezing, and lung infections, typically start in infancy although the type and severity vary. There is no cure, but treatments are improving and people with CF may live well into adulthood.[1]

CF results from 2 DNA variants associated with CF, 1 in each copy of the *CFTR* gene.[1] People with a DNA variant associated with CF in only 1 copy of CFTR are considered carriers of CF and do not have the condition. Rarely, carriers may have mild symptoms such as pancreatitis.[1]

Patient conversation starters:

Cystic fibrosis (CF) is an inherited condition that primarily affects the function of the lungs. There is no cure, but treatments are helping people with CF live longer and healthier lives than was the case in the past.

People with 1 DNA variant in the *CFTR* gene are considered carriers of CF. Carriers do not have CF, but their biological children could have CF if their other biological parent (or sperm/egg donor) is also a carrier.

What this result means for family members

CF is an inherited condition. If both biological parents are carriers, each child has a 1 in 4 chance of having CF.

If only 1 parent is a carrier, each child has a 1 in 2 chance of being a carrier and a 1 in 2 chance that they will not be carriers. Parents and siblings of CF carriers may also be carriers.

Individuals with this result should discuss their carrier status with their reproductive partners, as well as family members who may also be carriers. Comprehensive *CFTR* gene analysis for family members may be warranted, especially for family planning purposes.[1,2]

In people with a confirmed DNA variant associated with being a carrier of CF, genetic testing for family members may help inform their risks.[1,2] Therefore, it is strongly recommended that individuals share these results with their biological relatives and reproductive partners.

Patient conversation starters:

Cystic fibrosis runs in families.

That means it can be passed down from parents to their children. One of your biological parents is likely a CF carrier. Your full siblings have a 1 in 2 (50%) chance of also being a CF carrier.

Your biological child(ren)'s chance of being a CF carrier or having CF depends on if their other parent is a carrier.

Sharing these results with your family members is important so they can decide if they want to have genetic testing.

References

1. Savant A, Lyman B, et al. Cystic Fibrosis . March 26, 2002. Updated November 10, 2022. In: Adam MP, Ardinger HH, Pagon RA, et al, editors. GeneReviews®. University of Washington, Seattle; 1993-2023. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1250
2. The American College of Obstetricians and Gynecologists Committee Opinion. Carrier Screening for Genetic Conditions. Number 691, March 2017 (Reaffirmed 2023). https://www.acog.org/Clinical-Guidance-and-Publications/Committee-Opinions/Committee-on-Genetics/Carrier-Screening-for-Genetic-Conditions