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**

Key results

A pathogenic or likely pathogenic variant associated with cystic fibrosis was found in each of the 2 copies of the *CFTR* gene.

Next steps

Clinical recommendations	Resources
Genetic Insights is a screening test and not intended to diagnose whether an individual has cystic fibrosis. Consider a referral to a specialist for diagnostic evaluation of cystic fibrosis, if not previously completed.	The Cystic Fibrosis Foundation maintains a list of care centers. To find a cystic fibrosis care center, visit: https://www.cff.org/managing-cf/care-centers **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 Cystic Fibrosis Report 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

Genetic Insights is a screening test. That means it's not meant to tell you for sure if you have a certain health condition. If you haven't already, it's important to talk to a specialist about additional testing for cystic fibrosis.

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 the next steps.

You can access a genetic counselor through your online Genetic insights report at no additional cost, 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 buildup of thick mucus in the lungs, pancreas, and other organs leading to multisystem disease.[1] Diagnosis of CF includes clinical evaluation in addition to genetic testing and/or sweat chloride testing.[1]

There is no cure, but treatments are improving and people with CF may live well into adulthood.[1] Symptoms typically start in infancy and the type and severity vary. The severity of CF can vary, and certain DNA variants are linked to less severe forms of CF.[1]

Signs and symptoms of CF may include chronic coughing, wheezing or lung infections, gastrointestinal abnormalities such as malabsorption or pancreatic insufficiency, salty skin, and obstructive azoospermia in males.[1,2]

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.

Patient conversation starters:

Cystic fibrosis (CF) is an inherited condition that mostly 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.

CF is caused when 2 DNA variants are found, 1 in each copy of the *CFTR* gene. People with a DNA variant in only 1 copy of the gene are considered carriers of CF.

Carriers do not have CF, but could pass it on to their biological children if the other biological parent is also a carrier.

What this result means for family members

CF is an inherited condition. Parents of people with CF are most likely carriers. Full siblings of people with CF have a 1 in 4 chance to also have CF, a 1 in 2 chance to be carriers, and a 1 in 4 chance to neither be a carrier nor have the condition. Children of people with CF are carriers and would only be at risk of having CF if the other biological parent (or sperm/egg donor) is also a carrier.

People with confirmed DNA variants associated with CF should talk with their reproductive partners and at-risk family members. Comprehensive *CFTR* gene analysis for family members may be warranted, especially for family planning purposes.[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 the DNA variants linked to CF can be passed down from parents to their children.

For people who have CF, their biological parents are most likely carriers. Their siblings may have CF, may be a CF carrier, or may not have any DNA variants linked to CF.

Talking to your family members and reproductive partners about your CF status is important so they can decide if they want to have genetic testing for CF.

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. https://www.ncbi.nlm.nih.gov/books/NBK1250
2. Cystic Fibrosis Foundation. Accessed November 2022. www.cff.org