

## Physician Letter

### PATIENT INFORMATION

NAME: \_\_\_\_\_

DOB:        /        /        TEST DATE:        /        /

Dear Dr. \_\_\_\_\_,

### PATIENT TEST RESULTS

This letter is in reference to the above patient, who was tested for **Cystic Fibrosis (CF)** using a buccal swab test through LabCorp.

**Your patient was found to be heterozygous for the delta F508 mutation, also known as being a CF carrier.**

Your patient ordered testing from DNA Direct, a Web-based genetic testing service ([www.dnadirect.com](http://www.dnadirect.com)) that provides pre-test education and consent as well as post-test interpretation and resources. We have advised your patient to forward this letter to you. Please include this report in your patient's medical records.

The following is a brief description of the condition, risks to your patient, risks to the patient's family, and a listing of current literature and resources.

### OVERVIEW

As you are aware, CF is a disease that results from mutations in the CFTR gene (cystic fibrosis transmembrane conductance regulator). CF mutations cause abnormal chloride concentrations across the apical membrane of epithelial cells in the lungs, pancreas, intestine, sweat glands, and vas deferens, resulting in a complex multi-system disorder. Affected individuals have a range of symptoms including, but not limited to, progressive pulmonary disease, pancreatic insufficiency, repeated infections, difficulty gaining weight, and male infertility. Since the advent of genetic testing, adult patients with milder signs and symptoms — such as persistent/recurrent sinusitis, nasal polyps, or digestive issues — have been identified.

CF is estimated to affect at least 30,000 children and adults in the United States. The average lifespan of an individual with CF is 32 years, the main cause of death being pulmonary failure.

### GENETICS OF CF

CF is inherited as an autosomal recessive condition. This means that in order to have CF, a patient must inherit two mutations, one from each parent. A carrier is defined as a healthy individual with one mutation and one functional copy of the CFTR gene. When two carriers have children together, there is a 1 in 4, or a 25% chance that each child will have CF.

## LIMITATIONS OF GENETIC TESTING

The CFTR gene is very large with well over 1000 mutations identified to date. According to guidelines established by the American College of Medical Genetics and the American College of Obstetrics and Gynecology, CF testing should include the 25 most common mutations. At DNA Direct, individuals are tested for 32 CF mutations.

Detection rates and carrier frequencies vary based on ethnic background, as indicated below:

Ethnic Group	Carrier Frequency	Detection Rate
Ashkenazi Jewish, Caucasian	1 in 29	97%
Northern European/Caucasian	1 in 25	90%
Southern European/Caucasian	1 in 25	68-70%
African American	1 in 60-65	69%
Hispanic American	1 in 45	55-57%
Asian American	1 in 90	30%
Other	Unknown	Unknown

## DIAGNOSING CF

If your patient is experiencing symptoms of CF, he or she may have a second CF mutation that remains undetected through our CF testing panel. You may choose to do the following:

1. Offer your patient testing through a laboratory that provides an expanded mutation panel. Genzyme Laboratories offers testing for approximately 86 mutations.
2. Refer your patient to the appropriate specialist to obtain a sweat test the “gold standard” method of diagnosing CF. This is often conducted by a pulmonologist.
3. Discuss the option of gene sequencing, a method by which the entire CFTR gene is analyzed for mutations. A genetic counselor or medical geneticist can help facilitate this discussion.

## PATIENT CARE

More than 10 million people in this country are unaffected healthy carriers. For family planning purposes, your patient’s partner should consider CF testing. Carriers do not require any health modifications; however, a referral to a genetic counselor is highly recommended if both parents are identified as carriers. At-risk couples, who often carry different mutations; in the CFTR gene, face a 25% chance of having children with CF.

## TREATING CF

To date, there is no cure for CF. Treatments are tailored to the patients’ symptoms and can include daily antibiotics, oral pancreatic enzymes, and physiotherapy. The main goal of treatment is to prevent infections, reduce the amount and thickness of secretions in the lungs, improve airflow, and maintain adequate calories and nutrition.

A referral to a regional CF center is recommended for patients who are undergoing evaluation for diagnosis or patients who are already diagnosed with the condition. CF centers include a multidisciplinary team of physicians, nurses, respiratory therapists, nutritionists, genetic

counselors, and social workers. To find a CF center in your area, contact the CF Foundation or the National Society of Genetic Counselors. For assistance in this matter, you can speak to our experts at DNA Direct by calling 1-877-646-0222.

## FAMILY AND POPULATION RISKS

The chart below shows the chance that the relatives of a carrier have also inherited the same mutation.

Relation to a CF carrier	Risk of being a carrier
Identical twin	1 in 1 (100%)
Child, sibling, parent	1 in 2 (50%)
Grandparent, uncle, aunt, nephew, niece, half sibling	1 in 4 (25%)
First cousin	1 in 8 (12.5%)

Your patient may be interested in sharing this information with their at-risk relatives.

A Personalized Report was created for your patient that includes detailed information about inheritance patterns, family planning considerations, and strategies for sharing genetic information with relatives. With your patient's permission, you may obtain a copy of the complete report. You may also refer to the resources below for additional information geared toward medical professionals.

We appreciate your time and effort in considering this test result in the context of your patient.

Sincerely,

Elissa Levin, M.S., CGC  
Clinical Director, DNA Direct  
1-877-646-0222  
[expert@dnadirect.com](mailto:expert@dnadirect.com)

## LITERATURE REFERENCES

Preconception and Prenatal Carrier Screening for Cystic Fibrosis. October 2001 Guidelines published by the American College of Obstetricians and Gynecologists & American College of Medical Genetics.

Langfelder-Schwind E, Kloza E, Sugarman E, et al. 2005. Cystic Fibrosis Prenatal Screening in Genetic Counseling Practice: Recommendations of the NSGC. *Journal of Genetic Counseling*. 14: (1): 1-15.

## ONLINE RESOURCES

CFTR-Related Disorders. Gene Reviews. [www.genetests.org](http://www.genetests.org)

Cystic Fibrosis Foundation: [www.cff.org](http://www.cff.org)

National Society of Genetic Counselors: [www.nsgc.org](http://www.nsgc.org)

ACOG (American College of Obstetrics and Gynecology) Screening Recommendations: [www.acog.org/from\\_home/publications/press\\_releases/nr12-12-01-2.cfm](http://www.acog.org/from_home/publications/press_releases/nr12-12-01-2.cfm)