

**AUTHORIZATION FOR THE  
GENETIC TEST FOR  
CYSTIC FIBROSIS**

Patient Name: \_\_\_\_\_ MR#: \_\_\_\_\_

Account #: \_\_\_\_\_ DOB: \_\_\_\_\_ Date: \_\_\_\_\_

**Cystic fibrosis** Cystic fibrosis (CF) is one of the most common recessive genetic diseases in Caucasians of North European descent, affecting approximately 1 in 2500 newborns. CF causes the body to produce abnormally thick mucous secretions which prevent the normal functioning of multiple organ systems. Symptoms vary among affected individuals, but the most common findings include chronic lung disease, severe breathing difficulties, pancreatic insufficiency, and abnormal sweat electrolytes.

Everyone has two copies of the CF gene (known as CFTR). Individuals affected with CF have two abnormal copies. Individuals who have one normal and one abnormal copy of the CF gene are known as "carriers" and are asymptomatic (show no symptoms of the disease). CF is inherited in an autosomal recessive pattern meaning that when both parents carry one abnormal CF gene, the couple has a 25% (1 in 4) chance of having an affected child. In Caucasians of Northern European descent, approximately 1 in 25-29 individuals carries an abnormal gene for CF. The CF carrier frequency in Hispanics, African Americans and Asian Americans is about one in 46, 65 and 90, respectively.

**Molecular test for Cystic fibrosis** You will be required to donate 10 mL of blood, which is equal to about two teaspoons. For prenatal diagnosis, 10-20 mL (2-4 teaspoons) of amniotic fluid collected by amniocentesis is necessary. To precisely interpret a prenatal diagnosis, the genotypes of both parents must be known or parental blood samples must be submitted for analysis with the prenatal sample. If parental genotypes are not determined, a less precise estimate of the risk of CF in the fetus will be provided. In addition, you may be asked to provide information regarding your medical history. A correct history is critical for proper interpretation of the data. This is a routine clinical laboratory test and the results from it may aid in diagnosis, so your or your health insurer will be billed for the procedure.

**Significance of the results** If one or more CF mutations are found by the testing procedure, the individual tested may be a carrier of, or affected by CF. The significance of the results will depend on which mutation(s) are found and what other symptoms are also present. **A positive result by itself should not be used as the sole criterion for diagnosis of Cystic fibrosis.** Rare (less than 1% of the time) errors may occur, for example due to sample mix-ups, or due to technical errors such as rare genetic variants that mimic or mask the mutation being tested. **To understand your results, you should consult your physician and may wish to consider further independent testing or pursue genetic counseling.**

**Limitations** As of 2010, there are over 1700 known gene alterations (mutations) associated with CF. Not all of these gene mutations are presently detected by typical CF screening tests. The test performed by the Molecular Diagnostic Laboratory will detect about 90% of CF mutations in Upstate New York Caucasians, 94% in Askenazi Jews, 74% in Hispanics, 66% in African Americans, and 55% in American Asians. For Upstate New York Caucasians, approximately 1% of CF affected individuals and about 10% of CF carriers will not be identified. A negative test does not rule out the possibility of being a CF carrier or CF affected individual.

**Results from the test** The test result will be provided to health care professionals directly involved with your care. Genetic counseling may also be appropriate as follow up. To the extent permitted by law, all of the records, findings, and results of this test are confidential and shall not be disclosed without your written consent specifically authorizing to whom such records, findings, and results are to be released. In accordance with NYS law, any remaining DNA will be discarded unless it is retained as a laboratory control, in which case all information identifying the DNA to you will be removed.

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Patient's Name: \_\_\_\_\_ Account #: \_\_\_\_\_ MR#: \_\_\_\_\_

I have read this entire document and understand its contents. In addition, I understand that I am free to withdraw any portion of my consent by crossing off and initialing unacceptable statements. Please note that crossing off the laboratory's option to bill for the testing will result in the testing being cancelled.

If you have any questions about the test to be performed, you may also wish to obtain genetic counseling prior to signing this form. You may also contact the Molecular Diagnostics Laboratory at (315) 464-6806.

Patient's Name (printed): \_\_\_\_\_ Patient's Medical Record Number: \_\_\_\_\_  
(for office use only)

**For the Patient:**

Please print the name, phone number, and address (if known) of all health care professionals, physicians (other than the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the Cystic Fibrosis test result. (Medical results cannot be sent to a patient, a patient's family member or guardian.) Please print legibly.

Name and Title	Address	Phone Number
_____	_____	_____
_____	_____	_____
_____	_____	_____

My signature below indicates that the above information has been explained to me and that I give consent for this Cystic Fibrosis testing.

Date: \_\_\_\_\_ Signature of Patient: \_\_\_\_\_

Name of Parent/Guardian: \_\_\_\_\_

Signature of Parent/Guardian if patient is a minor: \_\_\_\_\_

*As referring physician/health care professional, I understand the benefits and limitations of this clinical assay. I hereby attest to the fact that I have provided the patient or patient's guardian with the information contained above in compliance with the NYS Civil Rights Act, Section 79-l, have answered any questions fully, and have obtained a signed informed consent as appropriate.*

*I request that the above indicated genetic test be performed.*

Signature of Physician/Health Care Professional: \_\_\_\_\_

Printed Name/Stamp/Title: \_\_\_\_\_ Date/Time: \_\_\_\_\_