

SUNY Upstate Medical University
Authorization for the Genetic Test for Cystic Fibrosis

1. What is 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 prevents the normal function 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.

2. What is the purpose of the test and what are its limitations? Individuals affected with CF have two abnormal copies of a gene known as CFTR. Individuals who have one abnormal copy and one normal 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. The Molecular Diagnostics Laboratory can perform a test for CF gene detection to identify CF carriers and affected individuals. Currently, there are over 1000 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, 97% in Ashkenazi Jews, 57% in Hispanics, 69% in African Americans, and a lower and variable percent of mutations in American Asians. For Upstate New York Caucasian, approximately 1% of CF affected individuals and about 10% of CF carriers will not be identified. A positive result by itself should not be used as the sole criteria for diagnosis. 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.

3. What is required to perform the test? A 10 ml blood sample is necessary. This 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.

4. Is there a cost for the test? This is a routine clinical laboratory test and the results from it may aid in diagnosis, so you or your health insurer will be billed for the procedure.

5. What will happen to the DNA once the test is complete? The only testing that will be performed on this sample is the test for CF. The residual DNA may be stored indefinitely (this does not constitute DNA banking) to be used as a laboratory control in which case all identifying information will be removed.

6. How will I obtain results from the test? The test will be provided to your physician who will discuss it with you. 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.

If you have any questions about the test that will be performed, you may 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 <i>(for office use only)</i>
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For the Patient:

Please print the name, phone number, and address (if known) of all health care professionals, physicians (in addition to the referring physician), or other individuals/organizations (such as a health insurer) to whom you authorize the release of the CF 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 CF testing.

Date: _____	Signature of Patient
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Name of Parent/Guardian	Signature of Parent/Guardian if patient is a minor
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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 genetic test indicated above be performed.

Printed name of Physician/ Health Care Professional	Signature of Physician/Health Care Professional
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