

PATIENT INFORMED CONSENT FOR DNA TESTING

Patient Name: \_\_\_\_\_

Date of Birth: \_\_\_\_\_ Gender  Male  Female

Name of Referring Physician: \_\_\_\_\_

Test(s) to be performed:  Cystic Fibrosis Mutation Panel  Other \_\_\_\_\_

Reason for Testing:  Diagnosis  Carrier Status / Screening  Prenatal  Other \_\_\_\_\_

Ethnicity  African American  Asian  Caucasian  Jewish  Hispanic  Multiethnic  Native American  Other \_\_\_\_\_

Family History  Yes Specify Mutation (s) \_\_\_\_\_  No

The signature on this document constitutes authorization to collect and test samples for the above-designated genetic testing. The signature of the mother, or other legally authorized individual, provides authorization to collect and test samples from a child. The signatures also affirm that the benefits, risks, and limitations of this testing have been explained by a health care professional, including but not limited to the following:

- 1. that the results may diagnose the condition, indicate status as a carrier of the condition, and/or disclose a risk that a family member may develop or be a carrier of this condition.
2. that each party has been informed that there is a small risk associated with the collection of specimens. These risks are as follows:
- oral swab collection - inflammation of the lining of the mouth
- blood collection - hematoma or infection
3. that the significance of the results based on family history have been explained.
4. that the U.S. Food and Drug Administration (FDA) has not approved this test, but that approval is not necessary for the performance of this test. The results of this test should not be used as the only source of information for clinical diagnosis or patient management decisions.
5. that the test will not detect all mutations possible within this gene nor mutations in other genes. In addition, clinical misdiagnosis may occur due to sample misidentification or inaccurate family history.
6. that participants understand they may ask questions about the collection, testing, or reporting process.
7. that consent is given to use samples for medical research, test validation, or education. Refusal to give consent does not affect the completion of the test or reporting of the results.
8. that results will be released only to the ordering physician. Results will be released to a third party only if permission has been submitted in writing by the patient.

I consent to the use of my sample for research purposes as long as my privacy and confidentiality are maintained: Yes [ ] No [ ]
Signature \_\_\_\_\_ Date \_\_\_\_\_

PLEASE ENSURE THE PATIENT READS AND SIGNS THE INFORMED CONSENT and SEND TO MPLN



MOLECULAR PATHOLOGY LABORATORY NETWORK, INC.

Phone 800.932.2943 | 865.380.9746 | fax 865.380.9191
250 East Broadway Avenue | Maryville, TN 37804
www.mplnet.com

## What is cystic fibrosis?

Cystic fibrosis (CF) is one of the most common inherited genetic diseases. It affects approximately 30,000 people in the United States and one in 2,500 Caucasian newborns has CF. The incidence of CF in other ethnic groups varies from 1:8000 in Hispanics, 1:15,000 in African Americans, to 1:32,000 in Asian Americans.

CF is clinically characterized by pulmonary and gastrointestinal problems of varying severity. These symptoms can include difficulty with breathing and increased susceptibility to respiratory infections. Gastrointestinal problems are generally related to difficulty with the proper digestion of food and can result in malnutrition and poor weight gain. About 10% of infants with CF will have a bowel obstruction requiring surgical repair at birth. Treatment for CF depends on the degree and severity of symptoms but generally involves daily respiratory therapy for breathing problems, antibiotics for respiratory infections, and nutritional supplementation and special diets for the digestive symptoms. While treatment has greatly improved the quality of life for many CF patients, there is no cure for the disease and life expectancy is reduced with progressive respiratory failure as the leading cause of death.

CF is inherited in an autosomal recessive manner. What this means is that to be affected with CF, you must inherit two defective (abnormal) copies of the CF gene, one from each parent. CF carriers will carry one normal CF gene and one defective (abnormal) CF gene. Carriers do not have CF, but are at increased risk of having children with CF.

## Do I need to be tested for cystic fibrosis?

There are approximately 8 million individuals in the United States who are carriers of CF. The chance of being a CF carrier is dependent upon several factors including ethnic background, and family history (whether someone in your family has CF or is known to be a CF carrier). Whether you or your partner choose to have CF carrier testing remains a personal decision. However genetic testing is the only method available for determining carrier status and identifying couples who are at increased risk for giving birth to a child with CF.

## How is the test performed?

Your physician will obtain a sample (buccal swab, blood or liquid cytology) for testing. The cells obtained in this sample contain DNA, which is then analyzed for the presence of mutations in the CF gene. You will also be asked to provide information on your ethnic background and family history so that the laboratory results can be individually interpreted for you.

## What do the results mean?

This test detects the 25 core CF mutations recommended for carrier screening by the American College of Medical Genetics (ACMG) and the American College of Obstetricians and Gynecologists (ACOG) as well as eight additional mutations. The CF carrier-screening panel will either reveal the presence or absence of a CF mutation(s). If ordered for diagnostic purposes, the presence of two CF mutations will confirm a diagnosis of CF. The presence of one CF mutation indicates that the individual is a CF carrier. The absence of a detectable CF mutation indicates that the individual is less likely to be a CF carrier, but does not exclude the possibility that the person is a carrier of a rare mutation not tested for in the panel. In this instance, appropriate genetic counseling should follow CF mutation analysis so that the individual is fully informed of the significance of a negative test result.

## What if I am a carrier?

If you are found to be a carrier of a CF mutation there is a 1:2 (50%) chance that you will pass this mutation on to your offspring. However this does not mean that your child will have CF. This is dependent upon whether your partner also carries a CF mutation. As such, your physician will recommend that your partner be tested to determine the risk that a child of yours might have CF.

## What if we are both carriers?

If both partners are carriers, each time the couple conceives a child they will have a 1:4 (25%) chance of having a child with CF, a 1:2 (50%) chance of having a child who is an unaffected CF carrier, and a 1:4 (25%) chance that that child will not be a carrier.

## What if one of us has Cystic Fibrosis?

If one partner has CF and the other is a CF carrier there is a 1:2 (50%) chance of having a child with CF and a 1:2 (50%) chance that the child will be a CF carrier.

## Do I need to have genetic counseling?

Yes. Certain test results will generate the need for appropriate genetic counseling and it is important that individuals and couples receive accurate information about risks, prognostic factors, and the range of options available to make fully informed decisions.

## Where can I find out more about cystic fibrosis?

To learn more about cystic fibrosis, please talk with your health care provider. You can also find more information below: Cystic Fibrosis Foundation, 6931 Arlington Road, Bethesda, MD 20814. 1-800-FIGHT CF (1-800-344-4823), [www.cff.org](http://www.cff.org), E-mail: [info@cff.org](mailto:info@cff.org).

## Where can I find out more about genetic counseling?

National Society of Genetic Counselors, Executive Office  
233 Canterbury Dr. Wallingford, PA 19086-6617, 1  
610-872-7608. [www.nsgc.org](http://www.nsgc.org)

Genetic Alliance, Inc. 4301 Connecticut Ave. NW, Suite 404,  
Washington, DC 20008-2304. 1-800-336-4363.  
[www.geneticalliance.org](http://www.geneticalliance.org)

## Molecular Pathology Laboratory Network, Inc.

Molecular Pathology Laboratory Network Inc. (MPLN) is a preeminent provider of molecular diagnostic and flow cytometric testing in the areas of oncology, hematological and lymphoid malignancies, infectious diseases, genetic mutational analysis and human identity analysis.

MPLN is a fully accredited, CAP licensed and CLIA certified high complexity laboratory. We employ qualified medical technologists and actively pursue and participate in continuous quality improvement and assessment in all areas of the laboratory.



MOLECULAR PATHOLOGY  
LABORATORY NETWORK, INC.

Phone 800.932.2943 | 865.380.9746 | fax 865.380.9191  
250 East Broadway Avenue | Maryville, TN 37804  
[www.mplnet.com](http://www.mplnet.com)

v.June.2006