

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
E-mail: 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

Genetic Alliance, Inc.
4301 Connecticut Ave. NW
Suite 404 Washington, DC 20008-2304
1-800-336-4363
www.geneticalliance.org

About 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 cancer screening, hematological and lymphoid malignancies, genetic mutational analysis, infectious diseases 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.
250 East Broadway
Maryville, TN, 37804
1-800-932-2943, 865-380-9746
www.mplnet.com



MOLECULAR PATHOLOGY
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MOLECULAR PATHOLOGY
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Cystic Fibrosis Mutation Detection



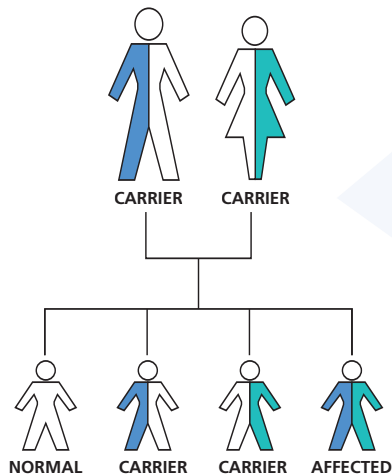
Patient Information

■ What is cystic fibrosis?

Cystic fibrosis is a common inherited disease affecting approximately 30,000 people in the United States. It causes digestive and breathing problems and may be diagnosed in infants with pneumonia or poor weight gain. Digestive problems are medically treatable; however, about 10% of infants born with cystic fibrosis have a bowel obstruction requiring surgical repair after birth. Breathing problems require respiratory therapy at home for about half an hour every day to clear mucus from the lungs. Lung infections are treated with antibiotics. Treatment has improved the quality of life for individuals with cystic fibrosis; however, there is no cure and the average life expectancy is approximately 30 years. Cystic fibrosis does not affect intelligence or appearance.

■ What causes cystic fibrosis?

Cystic fibrosis is a genetic disorder that occurs when a child inherits two copies of an altered gene. An altered gene is also referred to as a "mutation". One copy of an altered gene is inherited from the mother and the other copy from the father. In order to have cystic fibrosis, the child must inherit one copy of the altered gene from each parent. Although both the mother and the father have one copy of the altered gene, they also have an



unaltered copy which protects them from having the symptoms of cystic fibrosis.

Cystic fibrosis testing can tell you whether you have one or two copies of the altered gene. If you have one copy of the altered gene, you are a "carrier" of cystic fibrosis and do not have the medical symptoms. If you have two copies of the altered gene, you are "affected" with cystic fibrosis and will show some of the medical problems.

■ Do I need to be tested for cystic fibrosis?

The purpose of cystic fibrosis testing is to identify whether a couple is at increased risk for giving birth to a child with cystic fibrosis. Carriers of cystic fibrosis usually have no symptoms. Anyone can be a carrier for cystic fibrosis, even when there is no family history of cystic fibrosis and you already have healthy children. In the absence of a family history, the chance that an individual is a carrier for cystic fibrosis depends on their ethnic background. Finding out whether or not you are a carrier can benefit you in family planning. It may also benefit other family members; should you be a carrier for cystic fibrosis relatives may also be carriers and testing should be offered to them as well.

Ethnic Background	Carrier Risk	Chance of Child with CF
Caucasian	1 in 25	1 in 2,500
Hispanic	1 in 45	1 in 8,000
African American	1 in 60	1 in 15,000
Asian-American	1 in 90	1 in 32,000

■ How is the test performed?

There are several different ways of performing carrier testing for cystic fibrosis. It can be performed from a buccal swab (a swab sample from the inside of your mouth), blood work, or from your PAP smear. You and your doctor can determine which is the best and easiest way to perform your cystic fibrosis carrier screen.

■ What do the results mean?

The test detects 25 cystic fibrosis mutations recommended for carrier screening by the American College of Medical Genetics and the American College of Obstetricians and Gynecologists and eight additional mutations. Not all of the cystic fibrosis mutations are known. 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?

A positive result indicates that one of your two cystic fibrosis genes is altered. There is a one-in-two (50%) chance that you will pass this altered gene onto your child. It does not mean that the child will have cystic fibrosis. Carrier testing will be offered to the father of the child. If he is also tested and shown to have normal results then the chances are rare that the child will have cystic fibrosis.

■ What if we are both carriers?

If the father of the child is tested and he is also a carrier, the chance of your child having cystic fibrosis is then one-in-four (25%) with each pregnancy. Your doctor will discuss this with you in detail and may refer you to a genetic counselor. There are prenatal testing options available to determine if a baby will have cystic fibrosis or not.

■ What if one of us has cystic fibrosis?

If one of you has cystic fibrosis and the other is a carrier there is a one-in-two (50%) chance that the child will also have cystic fibrosis; however, there is also a one-in-two chance (50%) that the child will be a carrier and not have cystic fibrosis.