

Genetics



Cystic Fibrosis

Cystic fibrosis (CF) is an autosomal recessive disease characterized by pulmonary and gastrointestinal symptoms of varying severity. Inherited mutations in the cystic fibrosis transmembrane regulator (CFTR) gene affect transmembrane chloride ion transport, resulting in decreased chloride secretion and increased viscosity of extra cellular fluids. These thick secretions lead to duct obstruction with resultant organ dysfunction or failure.

Cystic Fibrosis affects 1:2,500 Caucasians, 1:8,400 Hispanics, 1:14,400 African Americans and 1:32,400 Asian Americans. When both parents are carriers of the CF mutation, each child has a one in two chance of also carrying the mutation and a one in four chance of being affected with CF.

More than 1,500 mutations in the CF gene have been reported to date. The American College of Medical Genetics recommended core mutation panel can be used for the most frequent mutations to identify CF carriers.

The CF mutation panel offered by Molecular Pathology Laboratory Network, Inc. will detect 32 mutations. This mutation set includes the 23 core mutations now recommended by the ACMG and additional mutations that are rare, accounting for only a small percentage of CF cases.

ACOG Recommendations

In late 2001, the American College of Obstetricians and Gynecologists (ACOG) began recommending that OB/GYN practitioners make DNA screening for CF available to all couples seeking preconception or prenatal care — not just those with a personal or family history of CF. This recommendation is endorsed by ACMG.

Recommendations for Mutation Testing

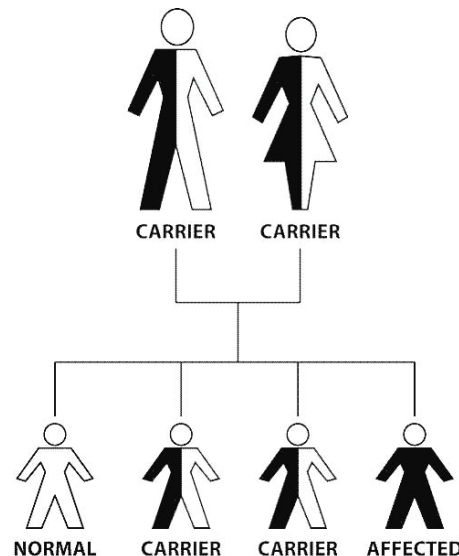
- Diagnostic: To confirm clinical diagnosis, identify familial mutations.
- Prenatal (pre-implantation) diagnosis (both partners are carriers).
- Carrier Testing / Population Screening: As a part of routine preconception or prenatal care, or for the test of reproductive partners if an individual with CF, a CF carrier, or a male with congenital bilateral absence of vas deferens.

| CF Carrier Detection Rate | |
|---------------------------|-----|
| Ashkenazi Jewish | 97% |
| European Caucasian | 80% |
| Hispanic | 57% |
| African American | 69% |

| Mutation $\Delta F508$ | |
|-----------------------------|-----|
| Northern European Caucasian | 70% |
| Ashkenazi Jewish | 30% |

Risk of an Affected Child

For a child to be born with cystic fibrosis, she/he needs to inherit two altered copies of the CF gene; one altered copy from the mother and one altered copy from the father. However, even when both members of the couple are carriers of CF there is a 3 out of 4 chance that the child will be unaffected. If only one member of the couple is a carrier of CF, the chance of having an affected child is low.





Specimen Requirements

- 5.0 ml (min. 3.0 ml) whole blood EDTA preferred, ACD whole blood accepted.
- Buccal swab, placed in transport medium (provided).
- 2.0 ml (min. 1.0 ml) ThinPrep® PAP solution or SurePath™ Test Pack

CPT Codes

83890, 83894, 83902, 83896 x32, 83912

References

1. Preconception and Prenatal Carrier Screening for Cystic Fibrosis. Clinical and Laboratory Guidelines. The American College of Obstetricians and Gynecologists. Oct. 2001.
2. Cystic Fibrosis Foundation. www.cff.org
3. National Society of Genetic Councilors www.nsgc.org
4. Genetic Alliance, Inc. www.geneticalliance.org

About MPLN

Since 1989, Molecular Pathology Laboratory Network, Inc. (MPLN) has offered an expanding selection of tests in molecular oncology, infectious diseases and human genetics to hospitals, medical laboratories and private physician groups nationwide.

Headquartered in Maryville, Tennessee, we are a fully licensed laboratory, certified by the Clinical Laboratory Improvement Amendment, and accredited by the College of American Pathologists and the states of Tennessee, Florida, New York and Maryland.

At MPLN, our philosophy is simple - we build strong professional relationships, deliver personalized service, and offer advanced diagnostic technology to support high-quality patient care.

For more information, contact your local representative or call MPLN client services at 800.932.2943.

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