**Lab Dept:** Anatomic Pathology  
**Test Name:** CYSTIC FIBROSIS MUTATION ANALYSIS, 106-MUTATION PANEL

### General Information

**Lab Order Codes:** CF106  
**Synonyms:** CF 106 Mutation Panel; CFTR 106 Mutation Panel  
**CPT Codes:** 81220 – CFTR (cystic fibrosis transmembrane conductance regulator) gene analysis; common variants (eg, ACMG/ACOG guidelines)  
**Test Includes:** Testing includes the 23 mutations recommended by the American College of Medical Genetics (ACMG) for Cystic Fibrosis (CF) testing as well as 83 other mutations.

### Logistics

**Test Indications:** Confirmation of a clinical diagnosis of cystic fibrosis. Risk refinement via carrier screening for individuals in the general population. Prenatal diagnosis or familial mutation testing when the familial mutations are included in the 106-mutation panel. Risk refinement via carrier screening for individuals with a family history when familial mutations are not available.

**Lab Testing Sections:** Anatomic Pathology - Sendouts  
**Referred to:** Mayo Medical Laboratories (MML Test: CFP)  
**Phone Numbers:**  
- MIN: 612-813-6280  
- STP: 651-220-6550  
**Test Availability:** Daily, 24 hours  
**Turnaround Time:** 1 – 2 weeks  
**Special Instructions:** Please fill out the Mayo Molecular Genetics – Congenital Inherited Diseases Patient Information Sheet (Supply T521) form. If specimens are submitted without this information, processing will be delayed. Specimen must arrive at the reference laboratory within 96 hours of collection.

### Specimen

**Specimen Type:** Whole blood
**Container:** Lavender top (EDTA) tube

Alternate tubes: Yellow top ACD (Citric Acetate) tube

**Draw Volume:** 2.5 mL (Minimum: 0.5 mL) blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine venipuncture. Mix tube thoroughly by gentle inversion.

**Special Processing:** Lab Staff: Do Not centrifuge. Send whole blood specimen in original collection container at room temperature. Forward promptly. Specimen must arrive at reference lab within 96 hours of collection.

**Patient Preparation:** None

**Sample Rejection:** Improper specimen, improper information will delay sample processing; mislabeled or unlabeled specimens

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**Interpretive**

**Reference Range:** An interpretive report will be provided.

**Critical Values:** N/A

**Limitations:**

This assay will not detect all of the mutations that cause cystic fibrosis. Therefore, the absence of a detectable mutation(s) does not rule out the possibility that an individual is a carrier of or affected with the disease.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in interpretation of results may occur if information given is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

In rare cases, DNA alterations of undetermined significance may be identified. A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

**Methodology:** Multiplex polymerase chain reaction (PCR)–based assay utilizing the Sequenom Mass Array platform is used to test for mutations associated with cystic fibrosis (106-mutation panel).

**References:**[Mayo Medical Laboratories](https://www.mayoclinic.org) August 2015

Phone: 1-800-533-1710  Fax: 507-284-4542