
Lab Dept: **Anatomic Pathology**

Test Name: **CFTR GENE, FULL GENE ANALYSIS**

General Information

Lab Order Codes: CFTRG

Synonyms: CF, Full Gene Analysis; Cystic Fibrosis (CFTR), Full Gene Sequencing

CPT Codes: 81223 – CFTR (cystic fibrosis transmembrane conductance regulator) gene analysis; full gene sequence
81222 – CFTR gene analysis; duplication/deletion variants

Test Includes: Full gene sequencing of the CFTR gene and CFTR large deletion/duplication MPLA, if appropriate.

Logistics

Test Indications: Follow-up testing to identify mutations in individuals with a clinical diagnosis of cystic fibrosis (CF) and a negative targeted mutation analysis for the common mutations. Identification of mutations in individuals with atypical presentations of CF (eg, congenital bilateral absence of the vas deferens or pancreatitis). Identification of mutations in individuals where detection rates by targeted mutation analysis are low or unknown for their ethnic background.

This is not the preferred test for carrier screening or initial diagnosis. For these situations, order Cystic Fibrosis Mutation Analysis, 106-Mutation Panel.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Mayo Medical Laboratories (MML Test: CFTRZ)

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 available from the laboratory. 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:	3 mL (Minimum: 1 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

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A

Limitations:

A small percentage of individuals who have a diagnosis of cystic fibrosis (CF) may have a mutation that is not identified by this method (eg, promoter mutations, deep intronic alterations). The absence of a mutation(s), therefore, does not eliminate the possibility of positive carrier status or the diagnosis of CF. For carrier testing, it is important to first document the presence of a cystic fibrosis transmembrane conductance regulator (CFTR) gene mutation in an affected family member.

In some cases, DNA alterations of undetermined significance may be identified. 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.

A previous bone marrow transplant for an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

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.

In addition to disease-related probes, the multiplex ligation dependent probe amplification (MLPA) technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.

Methodology:

Polymerase Chain Reaction (PCR)/DNA Sequencing/Dosage Analysis (Multiplex Ligation/Dependent Probe Amplification [MLPA])

References:

[Mayo Medical Laboratories](#) August 2015
Phone: 1-800-533-1710 Fax: 507-284-4542