
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: 14 – 20 days, performed weekly

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 blood collection. 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:	<p>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.</p> <p>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.</p> <p>Technical limitations: In some cases, DNA variants 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.</p> <p>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</p>

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.

Evaluation tools:

Multiple in-silico evaluation tools are used to assist the interpretation of this test result. These tools are updated regularly; therefore, changes to these algorithms may result in different predictions for a given alteration. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated.

Unless reported or predicted to cause disease, alterations in protein coding genes that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

Reclassification of Variants-Policy:

All detected alterations are evaluated according to American College of Medical Genetics and Genomics recommendations. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential significance. At this time, it is not standard practice for the laboratory to systematically review likely pathogenic alterations or variants of uncertain significance that have been previously detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

Methodology:

Custom Sequence Capture and Targeted Next-Generation (NGS) followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing (when appropriate) and Gene Dose Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA)

References:

[Mayo Clinical Laboratories](#) April 2019
Phone: 1-800-533-1710 Fax: 507-284-4542

Updates:

4/9/2019: Updated methodology.