Lab Dept: Anatomic Pathology

Test Name: CFTR GENE, KNOWN MUTATION

General Information

Lab Order Codes:	CFTRK
Synonyms:	CF, Carrier Detection; Cystic Fibrosis (CFTR), Known Mutation
CPT Codes:	81221 – CFTR (cystic fibrosis transmembrane conductance regulator) gene analysis; full gene sequence
Test Includes:	PCR sequencing specific to a previously identified CFTR mutation. An interpretive report will be provided.

Logistics

Test Indications:	Diagnostic confirmation of cystic fibrosis when familial mutations have been previously identified. Carrier screening of at-risk individuals when a mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene has been identified in an affected family member.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	Mayo Medical Laboratories (MML Test: FMTT)
Phone Numbers:	MIN: 612-813-6280 STP: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	2-3 weeks
Special Instructions:	Please fill out the Mayo Molecular Genetics – Congenital Inherited Diseases Patient Information Sheet (Supply T521) form available from the laboratory. Documentation of the specific familial mutation(s) must be provided with the specimen in order to perform this test. 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

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Interpretive

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

Limitations:

The identification of a disease-causing mutation in an affected family member is necessary before predictive testing for other family members can be offered. If a familial mutation has not been previously identified, Order: Cystic Fibrosis Mutation Analysis, 106-Mutation Panel or CFTR Gene, Full Gene Analysis.

Analysis is performed for the familial mutation(s) provided only. This assay does not rule out the presence of other mutations within this gene or within other genes that may be associated with Cystic Fibrosis (CF).

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.

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.

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 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]) is utilized to test for the presence of a specific mutation(s) previously identified in an affected family member.

References:

Mayo Medical Laboratories August 2015
Phone: 1-800-533-1710 Fax: 507-284-4542