Lab Dept: Anatomic Pathology

Test Name: CYSTIC FIBROSIS VARIANT PANEL

General Information

Lab Order Codes: CF106

Synonyms: CFTR Gene Variant Panel

CPT Codes: 81220 – CFTR (cystic fibrosis transmembrane conductance regulator) gene analysis; common variants (eg, ACMG/ACOG guidelines)

81222

Test Includes: This test includes targeted testing to evaluate over 500 genetic variants including 23 disease-causing variants recommended by the American College of Medical Genetics and Genomics.

For details regarding the specific variants identified by this test see Targeted Variants Interrogated by Cystic Fibrosis Variant Panel.

Logistics

Test Indications: Confirmation of a clinical diagnosis of cystic fibrosis

Reproductive risk refinement via carrier screening for individuals in the general population

Reproductive risk refinement via carrier screening for individuals with a family history when familial variants are not available

Identification of patients who may respond to cystic fibrosis transmembrane conductance regulator (CFTR) potentiator therapy

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Mayo Clinic Laboratories (MML Test: CFMP)

Phone Numbers: MIN: 612-813-6280

STP: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 1 – 3 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. If there is a family history of cystic fibrosis, the known variant in the family should be supplied for best interpretation of results.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube Alternate tubes: Yellow top ACD (Citric Acetate) tube
Draw Volume:	3.0 mL (Minimum: 1.0 mL) blood Note: Less than the optimal volume may not yield enough DNA for analysis.
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. Additional Information: 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed. 2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled
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:

This assay will not detect all known disease-associated variants that cause cystic fibrosis or CFTR-related disorders. Therefore, the absence of a detectable variant does not rule out the possibility that an individual is a carrier of or affected with this disease.

A negative result does not eliminate the risk of carrier status for any of the included conditions, due to the possibility that the patient carries a variant that is not interrogated with this assay or the rare chance of a false-negative result for a tested variant. For tested variants, the negative predictive value of this screen is greater than 98%. The patient's residual risk to be a carrier after a negative screen is dependent on ethnic background and family history.

A positive control was not available for all variants targeted on this panel. For more information regarding availability of a positive control for each variant see Targeted Variants Interrogated by Cystic Fibrosis Variant Panel. The negative predictive value of these targets is unknown.

Rare variants (i.e., 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.

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. This assay was designed to specifically target known disease-causing or likely disease-causing variants. In rare cases, DNA variants of undetermined significance may be identified. The laboratory encourages healthcare providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

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

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Methodology:

Targeted Genotyping Array

References:

Mayo Clinic Laboratories November 2024

Updates:

11/20/2024: Major revisions in most sections including the number of variants, specimen volumes and methodology.