MAYO CLINIC LABORATORIES

Patient ID	Patient Name	Birth Date	Sex	Age
321	TESTRNV, IMPLEMENTATION	1968-02-19	M	54

Order Number	Client Order Number	Ordering Physician	Report Notes
X100390252	X100390252	UNKNOWN,PROVIDER	

Account Information	Collected
C7028846 DLMP Rochester	30 May 2022 10:00

Cystic Fibrosis (CF) Mutation Panel

Result Summary

MCR

Positive (Carrier)

Result

MCR

The following heterozygous variant was identified:
CFTR, p.F508del (p.Phe508del), c.1521_1523del, Chr7:117199646_117199648, Pathogenic, Autosomal Recessive,
Cystic Fibrosis

Interpretation

1 MCR

This result indicates that this individual is a carrier of cystic fibrosis (CF). This interpretation assumes that this individual is not clinically affected with CF.

Of note, current data suggests that some patients with a clinical diagnosis of CF may respond to variant-specific therapies depending on the combination of variants.

Since a pathogenic variant has been identified, genetic testing of at-risk family members could be considered. If appropriate, genetic testing should be offered to this individual's reproductive partner to further clarify their risk of having a child with CF.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

CAUTIONS

This assay will not detect all known pathogenic variants causing cystic fibrosis (CF) 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 Expanded Carrier Screen Panels in Special Instructions. The negative predictive value of these targets is unknown.

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.

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. This assay was designed to specifically target known pathogenic or likely pathogenic variants. In rare cases, DNA variants of undetermined significance may be identified. 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.

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.

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects deidentified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

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Additional Information

MCR

ALTERNATE NOMENCLATURE

The c.1521_1523delCTT nucleotide change for this alteration is also called c.1653_1655delCTT.

Specimen

MCR

WB Whole Blood

Method

MCR

Targeted genotyping array (build GRCh37 (hg19)) was performed to test for the presence of select variants within the CFTR gene (GenBank accession number NM_000492).

Targeted DNA sequence analysis of the CFTR gene is used to clarify results of the initial analysis when appropriate.

See www.mayocliniclabs.com (Test ID CFMP) for additional information about this test, including variants assessed by this assay.

Released By

MCR

LINDA HASADSRI

Received: 31 May 2022 16:14 **Reported:** 16 Jun 2022 09:51

Laboratory Notes

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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