MAYO CLINIC

1-800-533-1710

Carrier Screen, Focused Panel

CSFP

PATIENT NAME				ORDER NUMBER
TESTRNV, IMPLEMENTATION				**M205000021**
PATIENT ID X100397472	DATE OF BIRTH 02/19/1968	AGE 54 Y	SEX Male	REQUESTED BY PROVIDER UNKNOWN
COLLECTED 9/5/2022, 9:00 AM	RECEIVED 9/6/2022, 2:49 PM	REPORTED 9/7/2022, 9:41 AM		

The collected, received, and reported dates and times on the report are in the time zone of the performing location.

7028846
MCL RochesterCampus
Rochester MN 55901

CLIENT ORDER NUMBER
X100397472

CLIENT MRN
321

RESULT SUMMARY

CFTR variant detected

RESULT

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

ADDITIONAL RESULTS

Fragile X syndrome (FXS): Normal (allele sizes: 16)

Spinal Muscular Atrophy (SMA): SMN1 Copies: greater than or equal to 2.
The g.27134T>G polymorphism is absent.

PATIENT NAME				ORDER NUMBER
TESTRNV, IMPLEMENTATION				**M205000021**
PATIENT ID X100397472	DATE OF BIRTH 02/19/1968	AGE 54 Y	SEX Male	REQUESTED BY PROVIDER UNKNOWN
COLLECTED 9/5/2022, 9:00 AM	RECEIVED 9/6/2022, 2:49 PM	REPORTED 9/7/2022, 9:41 AM		

The collected, received, and reported dates and times on the report are in the time zone of the performing location.

7028846
MCL RochesterCampus
Rochester MN 55901

CLIENT ORDER NUMBER
X100397472

CLIENT MRN
321

CLINICAL SUMMARY

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.

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.

FRAGILE X SYNDROME (FXS)

This screening panel detects >99% of carriers of FXS. Rarely, carriers of FXS may have a non-expansion type variant (e.g. single nucleotide variant, deletion) of the FMR1 gene not detected by this assay. There are no known differences in FXS carrier rates among those with differing ancestry.

HEMOGLOBINOPATHIES

This screening panel includes the most common hemoglobinopathy alterations, including many single nucleotide and some deletion-type alterations; however, it does not comprehensively rule out all known pathogenic alterations in hemoglobin genes. Further, carrier status cannot be completely excluded because this test cannot determine the chromosomal arrangement of deletions or duplications, if present. Therefore, this result should be interpreted in the context of clinical presentation and results of other laboratory tests [e.g. hemoglobin electrophoresis and complete blood count (CBC)].

SPINAL MUSCULAR ATROPHY (SMA)

SMN1 gene deletions account for 95% of SMA-causing pathogenic variants; however, carrier status cannot be completely excluded because this test cannot differentiate at least one copy of SMN1 exon 7 on each chromosome from all copies of SMN1 on one chromosome and none on the other chromosome. Individuals that carry zero copies of the SMN1 gene on one of their chromosomes may be at risk to have an affected child if their partner is also an SMA carrier. Other variants within the SMN1 gene, such as single nucleotide variants, are also not detected by this test.

This assessment assumes that this individual is neither clinically affected with nor has a family history of FXS, hemoglobinopathies, or SMA.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

ALTERNATE NOMENCLATURE

PATIENT NAME				ORDER NUMBER
TESTRNV, IMPLEMENTATION				**M205000021**
PATIENT ID X100397472	DATE OF BIRTH 02/19/1968	AGE 54 Y	SEX Male	REQUESTED BY PROVIDER UNKNOWN
COLLECTED 9/5/2022, 9:00 AM	RECEIVED 9/6/2022, 2:49 PM	REPORTED 9/7/2022, 9:41 AM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER X100397472 CLIENT MRN 321

The c.1521_1523del nucleotide change for this alteration is also called c.1653_1655del.

METHOD

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); copy number changes and select variants within the HBA1, HBA2, and HBB genes (GenBank accession numbers NM_000558, NM_000517, and NM_000518; SMN1 exon 7 and exon 8 copy numbers (GenBank accession number NM_000344).

A three-primer CGG repeat primed PCR assay, followed by fragment sizing by capillary electrophoresis, was used to determine the size of the CGG repeat in the 5' untranslated region (UTR) of the FMR1 gene (GenBank accession number [build GRCh37 (hg19)]: NM_002024). The reported number of CGG repeats is estimated to be correct to within +/-5%.

Reportable range:
Normal: 5-44
Intermediate (Gray Zone): 45-54
Premutation: 55-200
Full mutation: >200

A variety of methods including (but not limited to) multiplex ligation-dependent probe amplification (MLPA), digital droplet PCR, and Sanger sequencing are used to confirm variants detected when appropriate.

See www.mayocliniclabs.com (Test ID CSFP) for additional information about this test, including CFTR, HBA, and HBB mutations assessed by this assay.

DISCLAIMER

CAUTIONS

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.

PATIENT NAME				ORDER NUMBER
TESTRNV, IMPLEMENTATION				**M205000021**
PATIENT ID X100397472	DATE OF BIRTH 02/19/1968	AGE 54 Y	SEX Male	REQUESTED BY PROVIDER UNKNOWN
COLLECTED 9/5/2022, 9:00 AM	RECEIVED 9/6/2022, 2:49 PM	REPORTED 9/7/2022, 9:41 AM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER X100397472 CLIENT MRN 321

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.

TEST CLASSIFICATION
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.

SPECIMEN

WB Whole Blood

RELEASED BY

LINDA HASADSRI