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**		
X100390253	X100390253	UNKNOWN,PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		30 May 2022 11:00			

CF and SMA Carrier Screen 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.

The result indicates a reduced carrier risk for spinal muscular atrophy (SMA).

CYSTIC FIBROSIS (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.

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. An individual's residual risk for SMA carrier status varies by ancestry and SMN1 copies.

Ancestry	Prior Risk	2/0 Copies	3/0 Copies
European	1/35	1/632	1/3500
Ashkenazi	1/41	1/350	1/4000
Asian	1/53	1/628	1/5000
African	1/66	1/121	1/3000
Latinx	1/117	1/1061	1/11000
General Population	1/54	1/536	1/5450

*These calculations are based on the pathogenic variant detection rates and population carrier frequencies noted in the chart and assume no family history of SMA. We are unable to provide a revised risk assessment for ancestries other than those listed above as there is insufficient information available about the carrier frequency and pathogenic variant detection rates in other populations.

We are unable to provide a revised risk assessment for ancestries other than those listed above as there is insufficient information available about the carrier frequency and pathogenic variant detection rates in other populations. These calculations are based on the pathogenic variant detection rates and population carrier frequencies noted in the chart. They assume that this individual is neither clinically affected with nor has a family history of SMA.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

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

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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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.

Additional Information

MCR

ALTERNATE NOMENCLATURE

The c.1364C>A alteration is also known as c.1496C>A.

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

Specimen

MCR

WB Whole Blood

Method

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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) and for SMN1 exon 7 and exon 8 copy numbers (GenBank accession number NM_000344).

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 by when appropriate. SMN2 will only be reported in conjunction with relevant genotype findings.

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

Released By

MCR

LINDA HASADSRI

Received: 31 May 2022 16:16 **Reported:** 16 Jun 2022 09:57

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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