



Patient ID SA00378481	Patient Name TESTING, REPORTS	Birth Date 1983-01-01	Gender M	Age 34
Order Number SA00378481	Client Order Number SA00378481	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 31 Oct 2017 00:00		

Result Summary

MCR

Positive

Result

MCR

The following heterozygous alteration was identified:

Amino Acid change: p.E288V (Glu288Val)

DNA change: c.863A>T (g. 94847262)

Classification: PATHOGENIC. This alteration is commonly known as Pi*S

Interpretation

1 MCR

The c.863A>T (p.E288V) alteration is a known pathogenic mutation.

This result indicates that this individual is at minimum a carrier of alpha 1 antitrypsin (A1A) deficiency. However, we predict that there are individuals with a diagnosis of A1A deficiency who have pathogenic mutations that are not detectable by the method described (e.g. promoter mutations, large deletions or duplications, or deep intronic mutations). This result is best interpreted along with the total serum level of A1A; therefore, serum A1A testing is recommended if this has not already been performed.

Since a mutation has been identified, genetic testing of at risk family members could be considered. Mutation specific testing is available at Mayo Medical Laboratories by ordering FMTT / Familial Mutation, Targeted Testing. Please contact the Genomics Laboratory at 1-800-533-1710 with questions about this test.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the introns or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of mutations in all coding regions and intron/exon boundaries of the SERPINA1 gene (GenBank accession number NM_000295; build GRCh37 (hg19)). 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 de-identified 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 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.

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.

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

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.

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905



Patient ID SA00378481	Patient Name TESTING, REPORTS	Birth Date 1983-01-01	Gender M	Age 34
Order Number SA00378481	Client Order Number SA00378481	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 31 Oct 2017 00:00		

Additional Information

MCR

The p.E288V alteration is also known as p.E264V.

Released By

MCR

Victoria Waugh

Specimen

MCR

Buffy Coat

Received: 31 Oct 2017 08:42

Reported: 31 Oct 2017 10:55

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.

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905