

Patient ID SA00000865	Patient Name SAMPLEREPDMGLM, VLD20150713A0126	Birth Date 1981-01-01	Gender M	Age 34
Order Number SA00000865	Client Order Number SA00000865	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Jul 2015 16:01		

Result Summary

MCR

Positive

Result

MCR

The following heterozygous alteration was identified:

Amino Acid change: p.R122H (Arg122His)

DNA change: c.365G>A (g.142459789)

Classification: PATHOGENIC

Interpretation

1 MCR

The c.365G>A (p.R122H) alteration is a known pathogenic mutation.

This result is consistent with a diagnosis of hereditary pancreatitis (HP) for this individual.

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 Molecular Genetics 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 intron 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 a mutation in all coding regions and intron/exon boundaries of the PRSS1 gene (GenBank accession number NM_002769; 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 Clinic 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	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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Specimen

WB Whole Blood

MCR

Released By

EMILY LAUER

MCR

Received: 13 Jul 2015 20:23

Reported: 27 Jul 2015 13:33

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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