

Patient ID SA00000928	Patient Name SAMPLEREPDMGLM, VLD20150713A0189	Birth Date 1981-01-01	Gender M	Age 34
Order Number SA00000928	Client Order Number SA00000928	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Jul 2015 17:14		

Result Summary

MCR

Negative

Result

MCR

No reportable variants were identified.

Interpretation

1 MCR

This result decreases the likelihood of, but does not rule out, carrier or affected status for a CASR-related disorder. Some individuals who are carriers of or affected with a CASR-related disorder may have a pathogenic CASR variant that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant(s) in another gene(s).

Several conditions are known to have phenotypic overlap with those exhibited in individuals with CASR-related disorders. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. urinary calcium to creatinine ratio). If testing was performed due to a family history of a CASR-related disorder, genetic testing of an affected family member is necessary to determine whether this test is of predictive value for this family. Identification of the pathogenic variant in this family would allow for more accurate risk assessment of at risk individuals.

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 benign variants identified for this patient are available upon request.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of sequence variants in all six coding exons and intron/

exon boundaries of the CASR gene (GenBank accession number NM_000388; 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



Patient ID SA00000928	Patient Name SAMPLEREPDMGLM, VLD20150713A0189	Birth Date 1981-01-01	Gender M	Age 34
Order Number SA00000928	Client Order Number SA00000928	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Jul 2015 17:14		

Specimen

WB Whole Blood

MCR

Released By

Katrina Kotzer

MCR

Received: 13 Jul 2015 20:25

Reported: 23 Jul 2015 15:42

Test Environment
ETBM Template

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	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