CASRZ

CASR Gene, Full Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00001072	SAMPLEREPDMGLM, VLD20150713A0030		1981-01-01	M	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00001072	SA00001072	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		13 Jul 2015 14:02			

Result Summary

MCR

Positive

Result

MCR

The following heterozygous alteration was identified:
Amino Acid change: p.T436LfsX20 (Thr436LeufsX20)
DNA change: c.1305_1306delTA (g.121981187_121981188del)
Classification: PATHOGENIC

Interpretation

1 MCR

The c.1305_1306delTA (p.T436LfsX20) variant in the CASR gene results in premature termination (stop) codon and is therefore predicted to be pathogenic.

The result is supportive of a diagnosis of familial hypocalciuric hypercalcemia (FHH) for this individual but should be correlated with clinical findings. Additionally, this result indicates carrier status for neonatal severe hyperparathyroidism (NSHPT). Appropriate screening and/or management procedures should be considered.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing. A genetic consultation may assist with interpretation of this result and help to determine whether familial testing may be of benefit to this family. Familial testing for the sequence variants identified in this individual is available at Mayo Medical Laboratories by ordering test code FMTT (Familial Mutation, Targeted Test).

For information regarding the phenotypic spectrum associated with the CASR gene, see OMIM (www.ncbi.nlm.nih.gov/omim).

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.

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 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 Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

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Specimen MCR

WB Whole Blood

Released By MCR

MICHELLE KLUGE

Received: 14 Jul 2015 14:24

Reported: 24 Jul 2015 10:03

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 Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905