Patient ID	Patient Name		Birth Date	Gender	Age
SA00237587	TESTINGRNV, BRGGP ABN		1975-01-01	M	40
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00237587	SA00237587	CLIENT,CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		02 Nov 2015 06:00			

Brugada Syndrome Multi-Gene Panel,B

Gene(s) Evaluated

MCR

CACNA1C, CACNA2D1, CACNB2, GPDIL, KCNE3, KCNJ8, SCN1B, SCN3B, and SCN5A.

Result Summary

MCR

PATHOGENIC VARIANT(S) IDENTIFIED (SEE BELOW)

Result Details

MCR

The following variants were identified:

Gene (Transcript): SCN5A (NM_198056.2)
Genomic position: g.38627386_38627387
cDNA change: c.2582_2583delTT
Amino acid change: p.Phe861TrpfsX90
The patient is heterozygous for this variant
Classification: Pathogenic

The results for the remaining genes in this panel are negative.

Interpretation

MCR

The c.2582_2583delTT (p.Phe861TrpfsX90) variant in the SCN5A (NM_198056.2) gene is a known pathogenic variant.

The finding of a pathogenic variant is supportive of a diagnosis of Brugada syndrome for this individual but should be correlated with clinical findings. 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. Consultation with a genetics professional is recommended for interpretation of this result and to determine whether familial testing may be of benefit to this family. For questions regarding how to order variant specific testing, please call Mayo Medical Laboratories at 1–800–533–1710 or visit our website at www.mayomedicallaboratories.com. Please refer to family number 123 if ordering testing on family members of this individual.

Some of the genes tested by this panel may have more than one associated phenotype and/or inheritance pattern. Additionally, some genetic variants may have reduced penetrance and/or variable expressivity in some individuals. For information regarding the phenotypic spectrum which may be involved, see OMIM (www.ncbi.nlm.nih.gov/omim) and/or GeneReviews (www.genereviews.org) for this specific gene/disorder.

Next Generation Sequencing may not detect all types of genetic variants. If results do not match clinical findings, consider alternative methods for analyzing these genes.

ADDITIONAL INFORMATION

Laboratory developed test.

Method

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Next generation sequencing and/or Sanger sequencing was performed to test for the presence of a variant in all coding regions and intron/exon boundaries of the genes tested.

VARIANT EVALUATION

Evaluation and categorization of variants is performed using the most recent published ACMG recommendations as a guideline. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and predictions made by these tools may change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgment. 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.

Disclaimer

MCR

CAUTIONS:
CLINICAL CORRELATIONS

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data.

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
SA00237587	TESTINGRNV, BRGGP ABN		1975-01-01	M	40
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Misinterpretation of results may occur if the information provided is inaccurate or incomplete. If testing was performed because of a family history of cardiomyopathy or a related disorder, it is often useful to first test an affected family member. Identification of a pathogenic variant in an affected individual would allow for more informative testing of at risk individuals.

TECHNICAL LIMITATIONS

Next Generation Sequencing may not detect all types of genetic variants. Additionally, rare polymorphisms may be present that could lead to false negative or positive results. If results do not match clinical findings, consider alternative methods for analyzing these genes, such as Sanger sequencing or large deletion/duplication analysis. If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA.

RECLASSIFICATION OF VARIANTS POLICY

At this time, it is not standard practice for the laboratory to systematically review likely pathogenic variants or variants of uncertain significance that are detected and reported. 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. Consultation with a genetics professional should be considered for interpretation of this result.

A list of benign and likely benign polymorphisms identified for this patient is available from the lab upon request. Please contact the laboratory if additional information is required regarding the transcript and/or human genome assembly used for the analysis of this patient's results.

Reviewed by MCR

Linnea M. Baudhuin, Ph.D.

Received: 03 Nov 2015 09:31 **Reported:** 17 Nov 2015 13:58

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905