

Patient ID <b>SA00237586</b>	Patient Name <b>TESTINGRNV, BRGGP</b>	Birth Date <b>1975-01-01</b>	Gender <b>M</b>	Age <b>0 D</b>
Order Number <b>SA00237586</b>	Client Order Number <b>SA00237586</b>	Ordering Physician <b>CLIENT,CLIENT</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>01 Jan 1975 06:00</b>		

## Brugada Syndrome Multi-Gene Panel,B

### Gene(s) Evaluated MCR

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

### Result Summary MCR

**NEGATIVE**

### Result Details MCR

The results for all genes in this panel are negative.

### Interpretation MCR

This result decreases the likelihood but does not rule out involvement of the genes evaluated in this panel. Some affected individuals may have a pathogenic variant in one of these genes 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).

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.

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 MCR

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. 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

### 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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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:** 03 Nov 2015 14:13

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