MAYO CLINIC LABORATORIES

1-800-533-1710

# KVAR1

Known Variant Analysis-1 Variant, Varies

| Patient ID | Patient Name | | Birth Date | Gender | Age |
|---|---|---|---|---|---|
| SA00081786 | SAMPLEREPORT, KVAR1 | | 1977-07-07 | F | 39 |
| **Order Number** | **Client Order Number** | **Ordering Physician** | **Report Notes** | | |
| SA00081786 | SA00081786 | CLIENT, CLIENT | | | |
| **Account Information** | | **Collected** | | | |
| C7028846 DLMP Rochester | | 16 Nov 2016 09:00 | | | |

## Known Variant Analysis-1 Variant

### Variant Tested

MCR

Testing for the following familial variant was requested:
KCNQ1(NM_000218.2), c.1781G>A, p.Arg594Gln

See Method Summary for details on analysis performed.

### Result Summary

MCR

NEGATIVE

### Result Details

MCR

The KCNQ1, c.1781G>A, p.Arg594Gln variant was NOT detected in this individual.

### Interpretation

MCR

The c.1781G>A (p.Arg594Gln) variant in the KCNQ1 gene was previously identified in a family member of this individual with features of long QT syndrome and is classified by our laboratory as pathogenic.

The absence of the pathogenic familial variant in the individual tested here suggests that this individual is at no greater risk than someone in the general population for having long QT syndrome.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

A genetic consultation may be of benefit for interpretation of this result.

### Method

MCR

DNA sequence analysis was used to test for the presence of the following familial variant:

KCNQ1 (NM_000218.2), c.1781G>A
(Chr11(GRCh37):g.2799254), p.Arg594Gln

Genbank accession number refers to build hg19.

### Disclaimer

1 MCR

**CAUTIONS:**

Rare variants may be present and could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

If the familial variant tested here was previously identified at an outside laboratory, and a positive control sample from a family member was not provided, the possibility of a false negative result should be considered since we did not have the opportunity to confirm our laboratory's ability to detect the variant.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. This assay does not exclude the presence of other variants in the genes tested, because analysis was directed specifically toward the familial variant(s) provided to our laboratory. Accurate result interpretation is dependent upon correct information provided to the laboratory, including clinical and family history, and biological relationships among family members. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Some genes 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.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic blood or marrow transplantation, a pre-transplant DNA specimen is recommended for testing. VARIANT EVALUATION



**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 |

MAYO CLINIC LABORATORIES

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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.

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, variants found deep in the intron or variants that do not result in an amino acid substitution are not reported.

For additional information regarding the evidence used by our laboratory to classify the familial variant, please contact the laboratory at 1–800–533–1710.

**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.

**Reviewed by** MCR

Mary Karow

**Received:** 16 Nov 2016 14:13 **Reported:** 17 Nov 2016 15:14



## 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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