

Accepted Manuscript

Utilizing the Genome Aggregation Database, Computational Pathogenicity Prediction Tools, and Patch Clamp Heterologous Expression Studies to Demote Previously Published Type 1 Long QT Syndrome Mutations from Pathogenic to Benign

Daniel J. Clemens, BS, Anne R. Lentino, Jamie D. Kapplinger, BA, Dan Ye, MD, Wei Zhou, MD, David J. Tester, BS, Michael J. Ackerman, MD, PhD

PII: S1547-5271(17)31419-4

DOI: [10.1016/j.hrthm.2017.11.032](https://doi.org/10.1016/j.hrthm.2017.11.032)

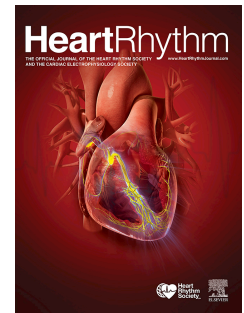
Reference: HRTM 7407

To appear in: *Heart Rhythm*

Received Date: 16 August 2017

Please cite this article as: Clemens DJ, Lentino AR, Kapplinger JD, Ye D, Zhou W, Tester DJ, Ackerman MJ, Utilizing the Genome Aggregation Database, Computational Pathogenicity Prediction Tools, and Patch Clamp Heterologous Expression Studies to Demote Previously Published Type 1 Long QT Syndrome Mutations from Pathogenic to Benign, *Heart Rhythm* (2017), doi: 10.1016/j.hrthm.2017.11.032.

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.



**Utilizing the Genome Aggregation Database, Computational Pathogenicity Prediction
Tools, and Patch Clamp Heterologous Expression Studies to Demote Previously Published
Type 1 Long QT Syndrome Mutations from Pathogenic to Benign**

Daniel J. Clemens, BS¹, Anne R. Lentino¹, Jamie D. Kapplinger, BA^{1,2}, Dan Ye, MD^{1,3}, Wei
Zhou, MD^{1,3}, David J. Tester, BS^{1,3}, Michael J. Ackerman, MD, PhD¹⁻⁴

Brief Title: Clemens – False Positive *KCNQ1* Variants

¹Mayo Clinic Graduate School of Biomedical Sciences, Department of Molecular Pharmacology & Experimental Therapeutics; Windland Smith Rice Sudden Death Genomics Laboratory, Mayo Clinic, Rochester, MN, 55905, USA

²Mayo Clinic School of Medicine, Mayo Clinic, Rochester, MN, 55905, USA

³Department of Cardiovascular Diseases/Division of Heart Rhythm Services, Mayo Clinic, Rochester, MN, 55905, USA

⁴Department of Pediatrics/Division of Pediatric Cardiology, Mayo Clinic, Rochester, MN, 55905, USA

Funding Sources:

This work was supported by the Mayo Clinic Windland Smith Rice Comprehensive Sudden Cardiac Death Program. JDK is supported by the NIH grant GM72474-08. JDK thanks the Mayo Clinic Medical Scientist Training Program for fostering an outstanding environment for physician-scientist training.

Disclosures:

MJA is a consultant for Audentes Therapeutics, Boston Scientific, Gilead Sciences, Invitae, Medtronic, MyoKardia, and St. Jude Medical. MJA, DJT, and Mayo Clinic have received sales-based royalties in the past from Transgenomic for their FAMILION-LQTS and FAMILION-CPVT genetic tests. MJA and Mayo Clinic have an equity/royalty relationship with AliveCor, Blue Ox Health, and StemoniX. However, none of these entities have contributed to this study in any manner.

Correspondence:

Michael J. Ackerman, MD, PhD
Mayo Clinic Windland Smith Rice Sudden Death Genomics Laboratory
Guggenheim 501, Mayo Clinic, 200 First Street SW, Rochester, MN 55905
507-284-0101 (phone), 507-284-3757 (fax)
ackerman.michael@mayo.edu

Total Word Count: 4,906 Words, 2 Tables, 2 Figures, 21 References

Download English Version:

<https://daneshyari.com/en/article/8660251>

Download Persian Version:

<https://daneshyari.com/article/8660251>

[Daneshyari.com](https://daneshyari.com)