Author's response to reviews

Title: Identification of novel KCNQ1 and KCNH2 mutations and the protective effect of KCNH2 SNP K897T in Long QT Syndrome families

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Author's response to reviews:

Dr. Melissa Norton, MD
Editor-in-Chief
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Dear Dr. Norton

We would be grateful if you would kindly consider the enclosed manuscript entitled “Identification of novel KCNQ1 and KCNH2 mutations and the protective effect of KCNH2 SNP K897T in LQTS families” for BMC medical genetics

In this manuscript, we performed mutational analysis in a population of human patients with long QT syndrome that causes life-threatening arrhythmias and sudden death usually in the young, otherwise healthy, individuals.

The novel aspects of the manuscript include:


2. Most importantly, we used a family-based approach to show that variant K897T in KCNH2 is a modifying factor for QTc and the rare allele T confers a protective effect against prolongation of QTc in patients with long QT syndrome. Controversial results exist in the literature regarding the effect of KCNH2 variant K897. Our study is the first to investigate the effect of SNP K897T on another KCNH2 mutation located in cis, and provides convincing evidence that K897T confers a protective effect.

We respectfully request that Dan Roden, Al George, Jr, Peter J Schwartz, and
Jeff Towbin be excluded from reviewing this manuscript due to conflict of interest.

Thanks for your consideration.

Yours sincerely,

Qing Wang, Ph.D., M.B.A.
Director, Staff, and Professor.