Short Communication

iMedPub Journals
http://journals.imedpub.com

Journal of Genomics & Gene Study

2021
Vol. 4 ISS. 6

Gender effect on the genotype-phenotype correlation in congenital long QT syndrome

Hector Barajas-Martinez

Corporacion Genetica Global (Global Genetics Corporation), USA/MEXICO

Abstract

In long QT syndrome (LQTS), female shows a higher risk of a cardiac event compared with a male in previous studies. However, the real nature about the difference between male and female is still lacking. In this investigation, we sought to comprehensively compare the genotype-phenotype correlation between sexes in genotyped LQTS patients. We enrolled 603 congenital LQTS cases from 6 registered centers (65.8% females; 72.9% probands; average ages at diagnosis, 21.8±17.8 y/o). Participants provided written informed consent, and clinical characteristics were recorded. All patients underwent at least LQT1-3 gene screening. Seventeen reported LQTS and other inherited arrhythmia candidate genes were sequenced with next-generation sequencing (NGS) in 306 (51%) cases. Major cardiac events (MCE) were defined as (aborted) sudden cardiac death, and/or documented malignant arrhythmias. Cardiac symptom includes syncope and MCE. Multiple factors determined the QTc and repolarization reserve could affect the clinical manifestation and eventually predispose cardiac events in LQTS patients. The gender difference in QTc is probably caused by sex hormone, especially higher level of testosterone after puberty. During the menstrual cycle, pregnancy, postpartum period, or menopause, QTc and risk of cardiac events significantly fluctuate in females. Nevertheless, the effect of gender and its underlying genetic dominators on QTc and clinical consequences is still undetermined, and the type of genetic mutation may help to further distinguish the high-risk LQTS subgroups, especially in the female. Although our result is deduced from patients collected by multiple international centers, and it includes the largest Chinese and Mexican LQTS cohort by far, it may not be generalizable to LQTS of all ethnicities, because we have restricted access to Black or African. In conclusion, the present study systematically reports the gender differences of genotype-phenotype correlation in LQTS patients, which provides further guidance on risk stratification and precision intervention. Further studies are warranted to decipher the unique gender code and to improve outcomes of LQTS patients.

Biography: Hector, Barajas-Martinez (PhD in Human Genetics and Fellow of Heart Rhythm Society) is currently the CEO of Global Genetics Corporation and Scientific Chief Officer (SCO) of CAMDIA MEDICAL LLC in Ventura, California. He is a new Special Section Editor of Electrocardiology and Genetics/Inherited Arrhythmia Disorders below to peer review Journal of Electrocardiology. He was a Clinical Director of Molecular Genetics/ Research Scientist at Molecular Genetics & Experimental Cardiology, Masonic Medical Research Laboratory in Utica, New York USA. Over the last 20 years or more he has been fully committed to advancing translational research in the field of genetics in cardiac arrhythmias. His new role in the Molecular Genetics and Functional Genomics Programs is to establish new strategies for molecular genetic approaches to identified new genetic markers in Inherited Cardiac Death Syndromes, Neuronal and Cancer diseases. He played a key role in the discovery and characterization of more than 8 new genes related to Brugada, Early Repolarization, Short and Long QT Syndromes and Hypertrophic and Dilated Cardiomyopathies, which were published in top tier scientific journals. We are interested in helping to identify novel gene or multiple genes mutations linked to inherited cardiac, diabetes, obesity, cancer and neuropathology syndromes associated with ion channels and structural gene-diseases.