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NOVEL COMPOUND HETEROZYGOUS MUTATIONS OF KCNQ1 IN LONG QT SYNDROME WITH FAMILIAL HISTORY OF UN-EXPLAINED SUDDEN DEATH: IDENTIFIED BY ANALYSIS OF WHOLE EXOME SEQUENCING AND PREDISPOSING GENE

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Objective: This study aimed to identify the pathogenic mutation in a Chinese family with long QT syndrome (LQTs) and unexplained sudden death (USD).

Methods & Results: Whole exome sequencing was conducted for the proband. The genetic data was screened using the 1000 genomes project and SNP database (PubMed), and the identified mutations were assessed for predicted pathogenicity using the SIFT and Polyphen-2 algorithms. We identified the compound heterozygous mutations in the KCNQ1 gene at c. G527A (p.W176X) and c.G1765A (p.G589S) predicted as "damaging". The *in-silico* analysis showed that when compared to the characteristics of mRNA and protein of wild-type KCNQ1, the mRNA of c.G527A mutation was significantly different in the centroid secondary structure; the subunit coded by W176X would lose the transmembrane domains S3-S6 and helices A-D; the protein secondary structure of G598S was slightly shortened in helix structure; the protein physics-chemical parameters of W176X and G589S significantly and slightly changed, respectively.

Conclusions: The compound heterozygous mutations of W176X and G589S coexisting in KCNQ1 gene of homologous chromosomes, resulting in more severe phenotype, are the likely pathogenic and genetic risks of LQTs and USD in this Chinese family.

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