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Whole exome sequencing identified a pathogenic mutation in *RYR2* in a Chinese family with unexplained sudden death

Yubi Lin[1,2]
[1]Guangdong General Hospital, China
[2]Medical School of South China University of Technology, China

Objective: This study aimed to identify the pathogenic mutation in a Chinese family with unexplained sudden death (USD) or occasional syncope.

Materials & Methods: Whole exome sequencing and gene chip sequencing were respectively conducted for two related patients. 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.

Results: We identified a heterozygous mutation in the RYR2 gene at c.490C>T (p. P164S), highly conserved across all species, in three members of this family, while another heterozygous de novo mutation in SCN5A at c.5576G>A p. R1859H was detected in one family member. Both variants were verified by Sanger sequencing. Importantly, *RYR2* p. P164S is associated with the risk of sudden cardiac death, such as in Catecholaminergic polymorphic ventricular tachycardia.

Conclusions: A pathogenic mutation in *RYR2* (p. P164S) is the expected cause of USD in a Chinese family associated with malignant ventricular arrhythmias. Whole exome and chip gene sequencing can be useful for discovering the genetic causes of USD.

Biography

Yubi Lin has completed his PhD degree from Jinan University and Postdoctoral studies from Guangdong Cardiovascular Institute, Medical School of South China University of Technology. He is the Chief Expert of Guangdong Province Family Doctor Association Telemedicine and the expert committee member of CMIA Remote Heart Monitoring Professional Committee of China. He has published more than 27 papers in reputed journals.

linyubi88@qq.com

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