Yubi Lin, J Clin Exp Cardiolog 2017, 8:9(Suppl)
DOI: 10.4172/2155-9880-C1-077

Whole genome sequence identified a rare homozygous pathogenic mutation of DSG2 gene in a familial arrhythmogenic cardiomyopathy involving both ventricles

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

Background: This study was designed to identify the pathogenic mutation in a Chinese family with arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) using the whole genome sequencing (WGS).

Methods and results: Probands II: 1 and II: 2 underwent routine examinations for diagnosis. Genomic DNA was extracted from the peripheral blood of family members and analyzed using WGS. The potentially pathogenic mutations that occurred in genes *DSG2, PKP4, PRKAG2, FOXD4, CTTN* and *DMD,* which were identified by SIFT or Polyphen-2 software as "Damaging", were validated using Sanger sequencing. Probands II:1 and II:2 shared an extremely rare homozygous mutation in the *DSG2* (p. F531C) gene, which was also demonstrated using intersection analysis of WGS data from the Probands II:1 and II:2. Electron microscopy and histological staining of myocardial biopsies showed widened and destroyed intercalated discs; interrupted, atrophic and disarranged myocardial fibers; and hyperplastic interstitial fibers, collagen fibers and adipocytes were infiltrated and invaded.

Conclusions: A homozygous mutation of DSG2 p. F531C was identified as the pathogenic mutation in patients with ARVC/D involving both ventricles, as the result of widened and impaired intercalated discs; interrupted myocardial fibers; and abnormally hyperplastic interstitial fibers, collagen fibers and adipocytes.

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

Notes: