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FAMILIAL SUDDEN CARDIAC DEATH CAUSED BY A DSG2 P.F531C MUTATION AS GENETIC BACKGROUND WHEN CARRYING WITH HETEROZYGOUS KCNE5 P.D92E/E93X MUTATION

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Background: Sudden cardiac death (SCD) induced by malignant ventricular tachycardia (MVT) among young adults with arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) is a devastating event. Parts of ARVC/D patients have a mutation in genes encoding components of cardiac desmosomes, such as desmoglein-2 (DSG2), plakophilin-2 and desmoplakin.

Case presentation: Here, we report a potentially pathogenic mutation in the DSG2 gene, which was identified in a family with ARVC/D using whole exome sequencing (WES) and Sanger sequencing. In all, Patient III: 1 with ARVC/D carried the compound heterozygous mutations of DSG2 p.F531C and KCNE5 p.D92E/E93X, which were both inherited from her mother (II: 2), who died of SCD. Carriers of DSG2 p.F531C showed various phenotypes, such as ARVC/D, SCD, MVT and dilated cardiomyopathy. For III: 1, there were significant low-voltage regions in the inferior-apical, inferior-lateral wall of the right ventricular epicardium and outflow tracts of the right ventricle. Under the guidance of a 3-Dimensional mapping system, MVT was successfully ablated with an epicardial-endocardial approach targeting for late, double or fragmental potentials after implantable cardioverter-defibrillator electrical storms. No VT recurrence was observed during the one year of follow-up.

Conclusions: When coexisting with heterozygous KCNE5 p.D92E/E93X, heterozygous DSG2 p.F531C as a genetic background was found to predispose to ARVC/D, SCD and MVT, epicardial-endocardial approach can be used successfully for ablation.

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