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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 three-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, which were successfully ablated using an epicardial–endocardial approach.