

# 20<sup>th</sup> European Cardiology Conference

October 16-18, 2017 | Budapest, Hungary

## Familial sudden cardiac death caused by DSG2 mutation as genetic backgrounds: Whole exome sequencing and valid therapy of catheter ablation

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**Objective:** We performed a clinical and genetic study in a family of arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) with history of sudden cardiac death (SCD).

**Materials & Methods:** Use emergency electrocardiograms and ICD monitoring to record. The Whole Exome and Sanger sequencing were performed using peripheral blood. The genes predisposing to cardiomyopathies and arrhythmias were screened for potential pathogenic mutations. Electrophysiological examination and catheter ablation for MVT induced repeat ICD electrical storms were performed.

**Results:** The patient III:1 of ARVC/D carried heterozygous mutations of DSG2 p.F531C and KCNE5 p.D92E/E93X, which were inherited from her mother (II:2) died of SCD. The carriers with DSG2 p.F531C were showed with aggravated phenotypes of ARVC/D and DCM. For III:1, there were significant epicardial and low-voltage regions in the inferior-apical, inferior-lateral and outflow tract of right ventricle. The MVT was successfully ablated with epicardial-endocardial approach targeting for late, double or fragmental potentials after ICD electrical storms, under the guidance of three-dimensional mapping system. No VT recurrence was observed during one year follow-up.

**Conclusions:** DSG2 pathogenic mutation detected by whole exome sequencing was found to be an important genetic background predisposing to SCD, ARVC/D and MVT, which was successfully ablated by epicardial-endocardial approach.

### Biography

Yubi Lin has completed his PhD 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 and has been serving as an Editorial Board Member of repute.

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