Patients characteristics of idiopathic ventricular fibrillation carrying SCN5A mutations without Brugada or long QT syndrome

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**Introduction**: The idiopathic ventricular fibrillation (IVF) is the main cause of sudden cardiac death (SCD) without any structural or metabolic heart disease, and some gene mutations are detected, which mainly code for ion channel units or its regulatory proteins. SCN5A mutations are one of the most common gene mutations in IVF patients as in Brugada syndrome (BrS) and long QT syndrome (LQT), but we can also detect the mutations in those who were not diagnosed as BrS and LQT. In this study, we evaluated the IVF patients’ characteristics with SCN5A mutations and without specific ECG pattern (Brugada like ST elevation and QT prolongation).

**Methods**: From our database about gene testing, firstly, we extracted the 36 patients with SCN5A mutations, who documented spontaneous VF events. Then, we checked their ECG, and excluded BrS (spontaneous or induced coved type ST-elevation), LQT (QT prolongation), or other heart diseases. Then, we recruited 7 IVF patients and evaluated their characteristics and clinical history.

**Result**: The median age was 19 years old (16, 16, 19, 38, 51, 60) and there were 6 males and 1 female. The genetic test about SCN5A mutations showed 5 pathogenic mutations (R433C, F1775LfsX15, R800H, Y68X, S329N) and 2 unknown pathogenic mutations (IVS21+17G>A, IVS7+5A>G). The detail of one female patient was not fully known. Three patients had the history of syncope events before IVF and only one male patient, who had received permanent pacemaker (PM) implantation for sick sinus syndrome, had familial history of IVF that his elder brother had the VF history and received implantable cardioverter defibrillator (ICD) therapy. Their other ECGs showed that one patient was only bradycardia, two patients had premature ventricular complex (one of two with couplet and another with inversion T wave in precordial leads), one patient with PM had atrial pacing rhythm and intraventricular conduction disturbance, and one patient showed complete right bundle branch block without ST elevation. Three patients underwent electrophysiology study, and only one of them could induce VF. The pilsicainide challenge test was important for excluding BrS, however, only 3 patients had undergone the test and all of their results were negative. As a treatment, all 7 patients received ICD and 2 patients took medical therapy.

**Conclusion**: We evaluated the characteristic of patients undetected any obvious heart disease with
SCN5A mutations, and most of them are male and young-onset without any familial history. More studies are needed to reveal this problem.