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PHENOTYPE DIFFERENCE BETWEEN GENOTYPE OF PLAKOPHILIN-2 MUTATION AND DESMOGLEIN-2 MUTATION IN SYMPTOMATIC CHINESE PATIENTS WITH ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY-A REPORT FROM CHINESE ARVD REGISTRY

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Objectives Arrhythmogenic Right Ventricular Dysplasia/cardiomyopathy (ARVD/C) is an inherited heart muscle disease associated mainly with the mutations of desmosome. Plakophilin-2 (PKP2) and desmoglein-2 (DSG-2) are reported as the most two common ARVD-causing genes in western countries. In this study we aim to determine the prevalence of PKP2 and DSG2 mutations in Chinese ARVD/C patients, and their phenotype characteristics.

Methods Genotype and phenotype were investigated in a cohort of 23 symptomatic unrelated Han Chinese with a clinical diagnosis of ARVD. PKP2 and DSG2 genes were identified using PCR and direct sequencing. Clinical evaluation included family and personal medical histories, 12-lead electrocardiography, transthoracic echocardiography, signal average electrocardiography, 24-h ambulatory electrocardiography, and MRI in some patients.

Results Five novel heterozygous mutations (R158K, Q211X, L419S, A793D and N852fsX930) of PKP2 were identified in 30% (7/23) of ARVD patients; three mutations (R46G, D494A and F531C) in

DSG2 were identified 13% (3/23) of the patients. Among the positive patients initial symptoms occurred at 30±10 years. All of them documented VT. Symptoms of the patients with PKP2 mutation were severe than that of patients with DSG2 mutation, all of the patients with PKP2 mutation had syncope, but none of the patients with DSG2, they only had palpitation. most of the patients with PKP2 mutation (6/7) showed epsilon waves in ECGs but only one in patients with DSG2; 6 patients with PKP2 mutation showed inverted T wave in V1 to V3, but only one in patients with DSG2 mutation.

Conclusions Five novel heterozygous mutations (R158K, Q211X, L419S, A793D and N852fsX930) of PKP2 and three heterozygous mutations (R46G, D494A and F531C) of DSG2 were identified. The study has revealed a greater frequency of occurrence of PKP2 mutations when compared to DSG2 mutations. There are some difference between patients with PKP2 mutation and that with DSG2 mutation including clinical symptom and ECGs. It seems that phenotype of PKP2 mutation were severe than that of DSG2 mutation in this cohort.