

of 10. Exon 8 where R169 resides encodes thirty seven amino acids of the RyR2 N-terminal which are highly conserved among many species.

Conclusions R169Q has been reported first in an 18-year-old Taiwanese woman by Hsueh in 2006. The present study identified R169Q on RyR2 gene again in a CPVT patient of China Mainland suggesting that this position might be a mutation hotspot in Chinese CPVT patients. The study laid foundation for further deepened study and specific management of this disease.

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A HOTSPOT MUTATION RYR2-R169Q FOR CHINESE PATIENTS WITH CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA

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Objectives Background: Catecholaminergic polymorphic ventricular tachycardia (CPVT), a malignant inherited arrhythmias, is characterised by stress-induced bidirectional or polymorphic ventricular tachycardia in absence of structural cardiac abnormalities. Unexplained syncope or sudden death in young individuals may be ascribed to CPVT. Mutations of RyR2 gene were proved to cause autosomal dominant CPVT, while mutations of CASQ2 gene cause rare autosomal recessive CPVT. To date, 155 RyR2 mutations have been reported to cause CPVT, but no mutation is identified in mainland of China. We intend to study the variants and prevalence of RyR2 gene mutations in the Chinese CPVT patients.

Methods The clinical characteristics of three CPVT families provided by the National Channelopathy Registry Study were investigated, including family and personal medical histories, 12-lead electrocardiography and 24-h ambulatory electrocardiography. DNA samples of the probands and their parents were extracted from serum leukocyte. 45 exons where the known mutation clusters located were first identified by direct DNA-sequencing of PCR-amplified DNA fragment. The primers used for PCR were designed with Primer3 or Oligo6 software. The remaining 60 exons would be identified if those results were negative.

Results A novel heterozygous mutation was found in exon 8 at the 506th nucleotide (506G>A) on RyR2 in a 9-year old Chinese female child. This substitution resulted in an amino acid change from arginin to glutamin at the 169th position (R169Q). This mutation was absent in her parents and 50 control subjects (100 alleles) indicating that there is a de novo mutation occurred on the proband only. The first syncope occurred with bidirectional and polymorphic ventricular tachycardia recorded at the age of seven. A heart rate of 84 beats per minute and a QTc of 349 ms with right bundle branch block were observed. Despite the use of full-dose beta-blockade (metoprolol), stress-induced syncope re-occurred once a year during the following three years. Although radiofrequency current ablation was performed successfully at the age of eight, an unexpected sudden death still occurred at the age