Investigate clinical characteristics and genetic mutations of CPVT patients in Taiwan and risk stratification

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Background: Catecholaminergic polymorphic ventricular tachycardia (CPVT) is a rare and lethal arrhythmia in children and young adults. RYR2 mutation accounts for 60% of CPVT patients. While many articles about CPVT are published in the Caucasian population, only a few studies are based on Asian ethnicity. A CPVT scorecard proposed using Caucasian patients is proven effective in reducing the burden of variants of uncertain significance (VUS) of RYR2 mutations is not yet tested in the Asian population.

Purpose: Identify mutations among Taiwanese CPVT patients and compare genetic and clinical results with other ethnicities. We also tested the efficacy of the CPVT scorecard in Taiwanese CPVT patients.

Methods: 40 clinically diagnosed CPVT patients and their family members were consecutively enrolled from multi-centers. We compared clinical characteristics and genetic results with other ethnicities, and applied the Caucasian CPVT scorecard to test whether it is applicable in Taiwanese CPVT patients.

Results: This is the first nationwide CPVT cohort in Taiwan. Among the 29 patients with CPVT-related gene mutation, 12 RYR2 variants was identified in our cohort with 5 of them unreported in previous studies. The RYR2 yield rate was 55%, similar to other ethnicities. Exercise-induced symptoms including syncope and cardiac arrest were more severe in Taiwanese CPVT cohort, compared to Japanese and Caucasian cohorts. The CPVT scorecard also successfully reduced the VUS rate for the Taiwanese cohort.

Conclusions: Our study demonstrated genetic difference in CPVT patients across ethnicities, suggesting the importance of genetic testing in Taiwan. The CPVT scorecard is applicable to Taiwanese CPVT patients and is a helpful tool in interpreting genetic test results in clinical practice.

Clinical characteristics of the cohort

Gender (male, %)	15 (52%)
Mean age of onset (years)	18 ± 15
Exercise-related syncope or cardiac arrest (n, %)	23 (79%)
Exercise-related cardiac arrest (n, %)	14 (48%)
Family history of unexplained sudden cardiac death (n, %)	5 (17%)
Heart rate (bpm)	73 ± 16
Exercise stress test or Holter positive (n, %)	14 (48%)
Ventricular arrhythmia (n, %)	23 (79%)
Beta-blockers only (n, %)	10 (35%)
Intracardiac defibrillator implantation (n, %)	15 (52%)
CPVT score	6 ± 3