Mutations of the Cardiac Ryanodine Receptor (RyR2) Gene in Catecholaminergic Polymorphic Ventricular Tachycardia

Mihoko Kawamura1, Iori Nagaoka1, Kenichi Dohchi1, Yukiko Nishio2, Hideki Itoh1, Hiromi Kimura1, Akashi Miyamoto1, Yuka Mizusawa1, Yuko Jito1, Katsuya Ishida1, Makoto Ito1, Takeru Makiyama2, Seiko Ohno2, Naokata Sumitomo3, Kotaro Oyama4, Minoru Horie1

1Department of Cardiovascular and Respiratory Medicine, Shiga University of Medical Science, Japan, 2Department of Cardiovascular Medicine, Kyoto University Graduate School of Medicine, 3Department of Pediatric and Child Health, Nihon University School of Medicine, 4Department of Pediatric Cardiology, Iwate Medical University Memorial Heart Center

In 20 cases with clinically-diagnosed catecholaminergic polymorphic ventricular tachycardia (CPVT) from 12 unrelated Japanese families, we conducted genetic testing on RyR2, a gene encoding the cardiac ryanodine receptor. The correlation between RyR2 - mutations and clinical phenotypes was investigated. The RyR2 mutations were found in 9 cases from the 20 probands (incidence: 45.0%) and in 3 from the 12 family members manifesting CPVT (incidence: 25.0%). Both bidirectional ventricular tachycardia (bVT) and atrial arrhythmias were significantly more frequent in RyR2 -positive compared to RyR2 -negative CPVT patients. The findings suggested that RyR2 mutations are closely related with bVT and atrial arrhythmias of early onset.

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