Introduction: Catecholaminergic polymorphic ventricular tachycardia (CPVT) is an inherited genetic disorder. Ventricular tachycardia (VT) in CPVT typically occurs during exercise or at the time of emotional stress, and classically take the form of bidirectional VT or ventricular fibrillation (VF). QT interval is usually normal in CPVT patients. We report a case of an 8-year-old boy with a variant in RYR2 which is the major causative gene for CPVT. He presented both bidirectional VT during exercise and QT prolongation, overlapping clinical features of CPVT and Long QT syndrome (LQTS).

Methods: We performed genetic analysis for 46 genes using targeted gene panel sequencing methods by next generation sequencer and confirmed the detected mutations by Sanger methods. We defined a mutation which was not identified in ethnic matched healthy controls and was judged as pathogenic by multiple prediction software.

Result: Case presentation: An eight-year-old boy was referred to our hospital due to sinus bradycardia, atrial tachycardia, and exercise induced premature ventricular contractions (PVC). He did not have any episodes of syncope nor convulsions. He underwent the percutaneous pulmonary valvuloplasty when he was 4 months old. He did not have family history of any cardiac diseases or arrhythmias. At the age of 8 years, his resting 12 leads electrocardiogram showed the heart rate of 60 bpm and prolonged QTc (Bazzet QTc 513 msec). His Holter ECG showed supraventricular tachycardias (SVT) 57 times / day and bigeminal PVC provoking into bidirectional VTs. The exercise stress test exhibited bidirectional VTs and shortened QTc. He had the overlapping clinical features of CPVT and LQTS. A rare missense variant in RYR2 (c.5128C>A, p.H1710N) was detected without any other mutations or rare variants in known LQTS genes. After starting oral administration of nadorol and flecainide, the bidirectional VTs disappeared though SVT sometimes occurred. He has not experienced any syncopal episodes over the following 4 years.

Conclusion: A patient having a missense variant in RYR2 showed CPVT phenotypes and prolongation of QT intervals. It has been reported that approximately 30% of CPVT cases have been misdiagnosed as concealed LQTS. As shown in this case, prolonged QT interval does not necessarily exclude the possibility of CPVT. In the future, the mechanism of the QT prolongation and clinical features of overlap syndrome should be clarified.