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Catecholaminergic Polymorphic Ventricular Tachycardia and Left Ventricular Noncompaction Cardiomyopathy: a case report and review of the literature.

Tavera M.C.(1), Bloise R. (3), Napolitano C.(3), Monteforte N.(3), Stara R.(1), Tola G.(2), Giardina A.(2), Scorcu G.P.(2), Porcu M.(2), Priori S.(2), Tumbarello R.(1)
1. Division of Pediatric Cardiology, Cardio-Thoracic Department, Brotzu Hospital, Cagliari, Italy
2. Division of Cardiology, Cardio-Thoracic Department, Brotzu Hospital, Cagliari, Italy
3. Division of Cardiology and Molecular Cardiology, Fondazione Salvatore Maugeri, Pavia, Italy.

Introduction:
The case of a 14 year old girl affected by catecholaminergic polymorphic ventricular tachycardia (CPVT), sick sinus syndrome, left ventricular noncompaction cardiomyopathy (LVNC) and slight cognitive impairment will now be described.

Case report:
In 2006, a 9 year old girl experienced exertional syncope. Investigations revealed polymorphic ventricular tachycardia on exertion, sinus disfunction with pauses up to 3.59 sec and atrial tachycardia. Cardiac RM performed in 2006 was normal. A dual chamber epicardic pace-maker was implanted and anti-arrhythmic prophylaxis with propranolol and sotalol was started with little benefit.
In 2010, the patient was revalued in a reference center, and therapy was switched to nadolol and flecainide with good antiarrhythmic control. Genetic testing revealed a “de novo” hRyR2 gene mutation.
In 2011, she experienced monomorphic ventricular tachycardia without hemodynamic impairment. ECG revealed negative T waves in lateral precordial leads and echocardiogram showed a noncompaction-like morphology of the left ventricle with normal size and function. Plasmatic levels of flecainide were on therapeutic range. Dual chamber ICD with monocoil, active-fixation ventricular lead and 3830 active-fixation atrial lead (Medtronic, Inc.) was implanted. Flecainide was increased with better control of exertional atrial arrhythmias including atrial fibrillation. The evolution of cardiomyopathy will be monitored by close echocardiographic follow-up.

Discussion:
CPVT is a genetically determined disorder associated with syncope and sudden death that manifests predominantly in children and teenagers with a structurally normal heart. RyR2 mutations are the first reported gene alterations associated with the autosomal dominant form of CPVT. Whether or not ryanodine receptor mutation may induce structural abnormalities has been under debate since the first description of the ryanodine receptor involvement. Genetic mutations that give rise to CPVT phenocopies have been reported in the context of other arrhythmia syndromes with structural heart disease, such as arrhythmogenic right ventricular dysplasia and dilated cardiomyopathy. We describe a CPVT phenotype associated with sick sinus syndrome, LVNC and slight cognitive impairment, related to a RyR2 mutation. In our knowledge, no association between CPVT, sinus disfunction and LVNC has previously been reported . Further follow-up is required in order to assess evolution of the cardiomyopathy and arrhythmic profile.