

## **Hypertrophic Cardiomyopathy, Catecholaminergic Polymorphic Ventricular Tachycardia. Does genetics always help?**

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### **INTRODUCTION**

Hypertrophic cardiomyopathy (HCM) is a common genetic cardiovascular disease (ie, 1/500) and is a major cause of sudden death. Nowadays, in more than 60% of cases is possible to identify a genetic mutation and the vast majority of genes responsible for HCM encode proteins associated with the sarcomere.

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is an hereditary disease characterized by the development of adrenergically-mediated ventricular tachycardia in individuals with an apparently normal heart. There are described mutations in 6 genes, most of them are caused by the mutations in the cardiac ryanodine receptor gene RyR2.

Mutations in Ca<sup>2+</sup>-handling proteins can contribute to the pathogenesis of HCM. We identify a mutation in RYR2 gene in a patient with HCM phenotype.

### **CLINICAL CASE**

Our patient is a 12 years old girl checked for the first time for nonspecific chest pain.

Family past history negative for cardiac problems.

Physical examination and baseline ECG were normal

An echocardiogram revealed a maximum left ventricular wall thickness of 20 mm at the basal segment without left ventricular outflow obstruction. This hypertrophic zone had an increase echogenicity appearing like brightness cardiac mass. The left ventricular function was normal

Subsequently the patient underwent a cardiac MRI that revealed hypertrophy of the basal segment with helicoidal appearance, with increase of signal intensity in T1 and T2 indicative of fibrosis and revealed late gadolinium enhancement in this area compatible with a patchy fibrosis.

Stress test was normal, neither isolated premature ventricular contractions nor VT was induced. 24-hour Holter monitoring was normal as well.

Full panel testing was ordered for HCM finding a mutation in RYR2 p.332R>W, possibly pathogenic. This variant has been associated with CPVT.

The family screening in her parents and her brother was normal

The patient was placed on beta-blockers without seeing arrhythmic events during follow-up

### **CONCLUSION**

We describe a case of a patient with HCM with a genetic mutation in RYR2 p.332R>W. This variant is associated with CPVT and it is not found in general population.

Although RYR2 has been associated with HCM, the molecular and cellular mechanisms that link a RyR2 mutation with the development of HCM are completely unknown. Perhaps mutations in the ryanodine gene, that result in a dysfunctional release of calcium, lead to pathological cardiac remodeling.