

# Catecholamine induced polymorphic ventricular tachycardia

## ABSTRACT

**Aim:** To describe a rare case of catecholamine induced polymorphic ventricular tachycardia.

**Presentation of case:** Intermittent attacks of Ventricular fibrillation and syncope.

**Summary:** Catecholaminergic polymorphic ventricular tachycardia (CPVT) is a familial cardiac arrhythmia that is associated with ryanodine receptor gene (*RYR2*) or calsequestrin 2 (*CASQ2*) gene mutation. It is a rare condition that occurs in patients with structurally normal heart and leads to exercise/emotion triggered syncope and cause fatal arrhythmias. We present the case of a 21- year old female patient who was diagnosed with this condition at the age of 10 years and continues to live a healthy life.

**Conclusion:** Patients with CPSVT require life-long beta-blockers and might need an implanted defibrator.

*Keywords: Catecholamine, Ventricular tachycardia, Sudden cardiac death*

## INTRODUCTION

“CPVT is found to cause syncope or sudden cardiac death, and the three characteristic features of CPVT were later described by Coumel and associates”.<sup>1</sup> “These features of CPVT are: 1) exercise or emotion-induced potentially fatal ventricular tachyarrhythmias; 2) a peculiar pattern of bidirectional ventricular tachycardia with a normal resting ECG; and 3) an anatomically normal heart”<sup>1</sup>. Recent reports suggest that “CPTV is a genetic disease related to the mutation of 2 genes: mutations in cardiac ryanodine receptor gene (*RYR2*) or calsequestrin 2 gene (*CASQ2*) which lead to an increased intracellular  $Ca^{++}$  concentration, leading to arrhythmia due to a cascade of delayed after depolarization and triggered activity”.<sup>2,3,4</sup>

## PRESENTATION OF CASE

A 21 year old female with an established diagnosis of catecholamine induced polymorphic ventricular tachycardia presented to the outpatient clinic for a regular follow up.

She denies any exertional chest pain, pressure or discomfort, shortness of breath.

She has a past medical history of an episode of unprovoked syncope at the age of 10 years which required the use of external defibrillator and CPR to resuscitate her. She was diagnosed with catecholamine induced ventricular tachycardia and a defibrator was implanted at the age of 10 years. She experienced another episode of syncope when she went to a local concert. The automated cardioverter discharged appropriately and she was resuscitated.

There is no family history of sudden cardiac death, syncope, or any similar occurrences.

In between her episodes, she has normal exam findings. She has stable vitals and her cardiac and respiratory examination is normal and she has no complaints. On ICD interrogation it was found that there was appropriate detection and discharge for Ventricular Fibrillation from the AICD and she has successfully been reverted to normal rhythm during her episodes.

Her ECG shows normal sinus rhythm and Echo shows normal EF and mild mitral regurgitation. She tested positive for the gene responsible for CPVT including the RYR2 gene and CASQ2 gene mutation, while her parents tested negative for the same.

She takes nadolol 40mg once a day and would require the same life-long.

## DISCUSSION

“CPVT is found to be a cause of syncope, ventricular arrhythmias and sudden cardiac death. It mostly presents as syncope between 7-9 years of age”,<sup>5</sup> but sudden cardiac death could be the first presentation in young patients. In about 30% of CPVT patients, there is a positive family history of sudden cardiac death generally before the age of 40.<sup>6</sup> “It is seen that patients with *RYR2* mutation become symptomatic earlier, and men are at an increased risk of cardiac events”.<sup>7</sup> “CPVT is found to be associated with two genetic mutations, *RYR2* and *CASQ2*. *RYR2* is inherited in an autosomal dominant pattern and mediates the release of calcium from the sarcoplasmic reticulum that is necessary for myocardial contraction”.<sup>8</sup> “The *RYR2* mutation increases calcium release and can trigger fatal ventricular arrhythmias. The other genetic form of CPVT has an autosomal recessive inheritance and involves *CASQ2*. The *CASQ2* protein serves as the major calcium reservoir within the sarcoplasmic reticulum. It has the capability to bind increased amounts of calcium. The mutated protein may alter the calcium content within the sarcoplasmic reticulum, leading to alteration in the function of ryanodine receptor, or impairing the calcium release process”.<sup>9</sup>

“It is challenging to establish the diagnosis as ECG is normal in the absence of symptoms and echocardiography shows no specific findings. A characteristic finding on ECG is ventricular tachycardia with 180-degree alternation of the QRS axis (bidirectional tachycardia). CPVT is not reproducible by programmed electrical stimulation”.<sup>5</sup> “In patients suspected to have this disease, the arrhythmia should be recorded by Holter monitoring/loop recorder or induced by exercise treadmill testing. Activities triggering a burst in the sympathetic tone is the pivotal mechanism for this process. Hence, the focus of treatment is to contain the adrenergic activity, therefore, beta-blockers are the first-line drugs used in the management of CPVT. Beta-blockers are effective for acute phase as well as the maintenance treatment”.<sup>10</sup> However, if the symptoms recur despite the administration of a sufficient dose of beta-blockers, an implantable cardioverter/defibrillator must be employed.

Other arrhythmias in childhood might also lead to syncope or sudden cardiac death. The differential causes would include arrhythmogenic right ventricular cardiomyopathy (ARVC), Brugada syndrome, long QT syndrome (LQT), pre-excitation syndrome, commotio cordis, and Andersen-Tawil syndrome (ATS).

## CONCLUSION

CPVT is a potentially fatal disorder that is usually diagnosed in childhood. Patients presenting with sudden cardiac arrest can be misdiagnosed as having idiopathic VF and suffer from the sequelae of resuscitation. Therefore, it is important that clinicians carefully analyse the factors triggering VF in healthy individuals.

Ethical Approval:

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

Consent

As per international standard or university standard, patients' written consent has been collected and preserved by the author(s).

## ACKNOWLEDGEMENTS

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## COMPETING INTERESTS

There are no conflicts of interest to disclose.

## AUTHOR CONTRIBUTIONS

All the authors contributed to the preparation of the manuscript.

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