CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA IDENTIFIED BY 24 HOURS ECG MONITOR

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ABSTRACT

Catecholaminergic polymorphic ventricular tachycardia is a genetic arrhythmogenic disease, characterized by stress-induced, bidirectional ventricular tachycardia that may cause cardiac arrest and sudden death. We herein report the case of a 15-year-old male patient, rescued at home by firefighters after a convulsant syncope, occurred while he was at home, locked into the bathroom. An ECG Holter monitoring was in progress during the pathological event, because of patient’s history of minor symptoms.

Key words: Ventricular tachycardia, ECG.

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Introduction

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is a genetic malignant arrhythmogenic disease. The prevalence in the population is not known, but it has been estimated around 1:10,000. Genetic mutations in RyR2 or CASQ2 can be identified in 50% to 70% of patients. A mutation in the gene responsible for the long QT syndrome type 4, the gene encoding ankyrin B, was reported in a single patient with polymorphic ventricular tachycardia similar to CPVT. To date, more than 70 RyR2 mutations have been reported. Ryanodine receptor-related CPVT were predominantly males and became symptomatic earlier (8 ± 2 years). Calsequestrin-related CPVT is an uncommon variant of CPVT.

The differential diagnosis is between CPVT and long QT syndrome (LQTS). Often patients with CPVT are brought to clinical attention because the presence of LQTS has been suspected. Andersen-Tawil syndrome that is caused by loss of function mutations of the KCNJ2 gene encoding for the a channel that transports potassium ions into muscle cells. Genetic screening has crucial roles in establishing the appropriate diagnosis.

Case report

A 15-year-old male patient with an history of exercise-related brief syncopal episodes arrived at the emergency room, carried by firefighters after a self-limited convulsant syncope with a facial trauma, occurred into the home-bathroom during an ECG Holter monitoring.

Subsequent patient anamnesis revealed that the present syncope was related to sexual activity (masturbation).

There was a familial history of sudden death: both his father and an uncle (father’s brother) had suddenly died, in the previous last year during a strong emotional stress.

Three years ago the patient was admitted to hospital because of a brief syncopal episode. In
this occasion neither echocardiography, nor cardiac magnetic resonance revealed any structural abnormality, while the treadmill exercise test was interrupted for ventricular arrhythmias and it was recommended a beta-blocker therapy (nadolol 1.5–2 mg/kg/day).

Finally, the present 24-hour ECG Holter monitoring, that was in progress during the last syncopal attack, showed, at the onset of symptoms, a brief period of bigeminal ventricular rhythm and after a typical bidirectional ventricular tachycardia, two alternating QRS complexes morphologies with different polarity that suddenly degenerated into self-limiting polymorphic ventricular tachycardia with some “torsades de pointes” phases. (Fig 1)

**Figure 1**: ECG Holter monitor showed a typical bidirectional ventricular tachycardia, two alternating QRS complexes morphologies with different polarity that suddenly degenerated into self-limiting polymorphic ventricular tachycardia with some “torsades de pointes” phases.

An implantable automatic cardioverter/defibrillator was implanted (first episode occurred during betablocker therapy).

Genetic screening showed a pathological mutation into cardiac ryanodine receptor gene (RYR2) that affected the calcium release by the sarcoplasmic reticulum and caused catecholaminergic polymorphic ventricular tachycardia.

During a 26 months follow-up (continuing betablocker therapy), the patient presented no more recurrences. No ICD intervention until today.

**Discussion**

CPVT is a condition characterized by episodic syncope, life-threatening ventricular arrhythmias and sudden cardiac death in response to physical activity or emotional stress in patients without structural heart abnormalities.

The age of occurrence of first symptoms is between 7 and 9 years of age\(^6\), but sudden death may be the first presentation. Before the age of 40 there is a family history of sudden death in 30% of patients\(^7\) and if untreated CPVT is highly lethal.

CPVT is caused by two genetic mutations, RYR2 and calsequestrin (CASQ2). Patients with RYR2 mutation become symptomatic earlier, and men have a higher risk of cardiac events\(^8\). RYR2 is an autosomal dominant pattern and is responsible for excessive calcium release from the sarcoplasmic reticulum during sympathetic activation that generates depolarizing membrane currents required for myocardial contraction\(^9\).

A second genetic mutation involves cardiac CASQ2 with an autosomal recessive inheritance. The CASQ2 protein is responsible for the major calcium reservoir within the sarcoplasmic reticulum. The CASQ2 mutation may alter the calcium content within the sarcoplasmic reticulum, the function of the ryanodine receptor, or impair the calcium release process\(^9\). Molecular genetic testing for RYR2 and CASQ2 is important for clinical diagnosis.

CPVT is difficult to diagnose, because the resting electrocardiogram is usually normal and echocardiography shows no specific findings. The exercise treadmill testing is the most important diagnostic test and the arrhythmia must be recorded by Holter monitoring. A typical finding on ECG is the bidirectional tachycardia defined as a ventricular tachycardia with 180-degree alternation of the QRS axis on a beat-to-beat basis.

Our patient, ventricular arrhythmia appeared during masturbation, and then turned in the bidirectional ventricular tachycardia that finally degenerated into torsades de pointes. This is the typical course of CPVT, and it is caused by activity triggering by a burst in the sympathetic tone.

The focus of treatment is to suppress the adrenergic activity and beta-blockers are the most important drugs in the treatment of CPVT. Beta-blockers are effective for acute phase and maintenance treatment\(^10\). If the symptoms recur despite the administration of beta-blockers, an implantable cardioverter/defibrillator must be employed. In prevention of primary manifestations, beta-blockers are indicated for all clinically affected patients and probably for those with an RYR2 mutation with no history of cardiac events or ventricular arrhythmias on exercise stress testing.

In cases of cardiac arrest or ventricular arrhythmias in patients on beta-blocker therapy, an
implantable cardioverter defibrillator (ICD) may be necessary.

CPVT is a potentially fatal disorder that is usually observed in childhood. As in our case, patients presenting with sudden cardiac arrest can be mistaken as idiopathic VF and suffer from the sequelae of resuscitation. Therefore, clinicians should carefully analyze the triggering of factors VF in healthy individuals.

References


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