

Breaking It Gently: A Rare Case of Arrhythmogenic Right Ventricular Cardiomyopathy Presenting as Sustained Ventricular Tachycardia

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Abstract We report a case of a 42-year-old female with Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) who presented with recurrent episodes of palpitations, dizziness and lightheadedness. On evaluation, she developed a new onset sustained ventricular tachycardia (VT) and the diagnosis was confirmed through genetic testing, cardiac imaging, and electrophysiological study. Insertion of automatic implantable cardioverter defibrillator (AICD), antiarrhythmic medication, and ventricular ectopy radiofrequency ablation afforded improvement of symptoms. In conclusion, combination of these treatment modalities abated ventricular tachycardia from ARVC.

Keywords: *arrhythmogenic right ventricular cardiomyopathy, ventricular tachycardia, arrhythmia, antiarrhythmic medications, automatic implantable cardioverter defibrillator, radiofrequency ablation*

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1. Introduction

Arrhythmogenic Right Ventricular Cardiomyopathy may present with symptomatic sustained ventricular tachycardia which may be fatal that could lead to sudden cardiac death. This would mean early specialized intervention to abate symptoms and most importantly to prevent life threatening arrhythmias that could progress to cardiac arrest.

2. Case Presentation

A 42-year-old female diagnosed with ARVC at the age of 36 when she presented with recurrent episodes of palpitations, dizziness and lightheadedness. The diagnosis was confirmed by genetic testing though with an unspecified genomic information, electrocardiography, cardiac imaging, and electrophysiological study. No history of the same disease or sudden cardiac death in the family though his father has had atrial fibrillation. Six years ago, she gradually experienced intermittent episodes of palpitations which progressed to recurrent attacks of lightheadedness and dizziness. She denied chest pain, dyspnea or orthopnea during this time. Though her initial electrocardiogram showed normal sinus rhythm with normal axis and first degree atrioventricular block, her inpatient telemetry captured several runs of sustained monomorphic ventricular tachycardia (Figure 1). Transthoracic echocardiogram revealed normal left atrium,

left ventricular cavity size and wall thickness with mildly reduced systolic function with an estimated ejection fraction of 40% and note of regional wall motion abnormalities. Cardiac catheterization showed normal coronary arteries, severe left ventricular dysfunction and elevated ventricular end diastolic pressure. Cardiac magnetic resonance imaging was highly suspicious of AVRC. Electrophysiology study demonstrated new morphology of inducible monomorphic VT, confirming that she did not have a single morphology thus more suggestive of arrhythmogenic right ventricular cardiomyopathy. She was initiated on sotalol and mexiletene and eventually had an AICD inserted. Throughout the years, she had several hospital admissions due to palpitations, dizziness, lightheadedness, and defibrillator firing. Her AICD has been interrogated as well as her antiarrhythmic medications have been cautiously adjusted which afforded improvement on her symptoms and diminished discharge of her cardiac device.

Her recent hospital admission was due to palpitations and intracardiac device firing which she felt. She denied chest pain however she had mild dizziness and lightheadedness. She was noted to have sustained VT (Figure 2). Her blood pressure has been within the range during this course. She was started on amiodarone and lidocaine drips which converted her ectopy to sinus rhythm (Figure 3). Due to the intermittent episodes of sustained VT despite antiarrhythmic medications and intracardiac device, radiofrequency catheter ablation of the ventricular ectopy was recommended. The procedure was done successfully with no recurrence of palpitations, dizziness, lightheadedness, and device firing to date.

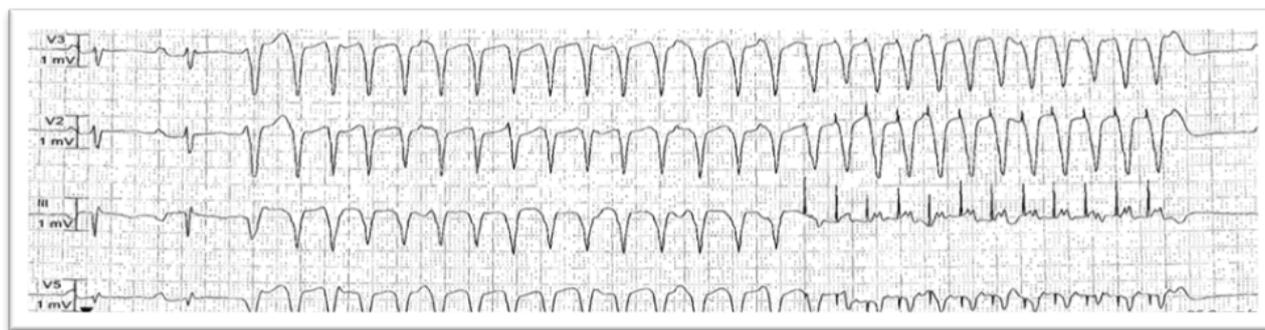


Figure 1. Patient's Telemetry Rhythm during her first hospitalization six years ago. The tracing revealed ventricular tachycardia with paced rhythm



Figure 2. Patient's 12 Lead Electrocardiogram when she presented with palpitations and cardiac device firing on her latest hospital admission this year. The tracing revealed sustained ventricular tachycardia

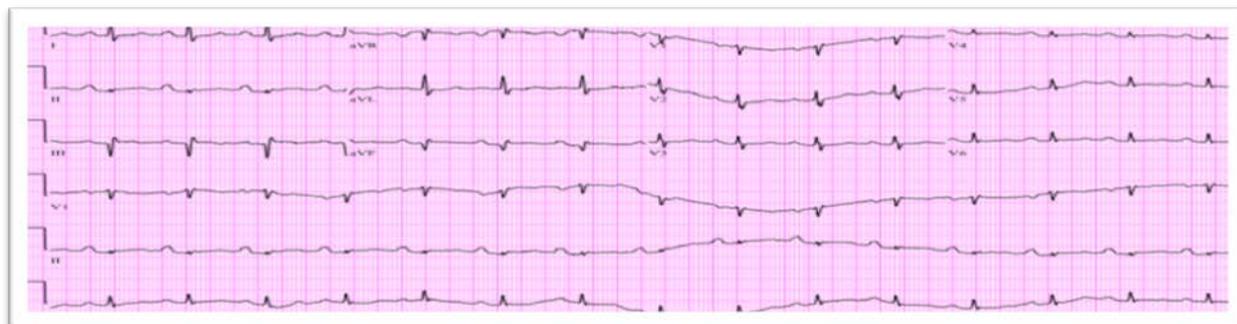


Figure 3. Patient's 12 Lead Electrocardiogram after the initiation of amiodarone and lidocaine drips. The tracing revealed normal sinus rhythm, left axis deviation

3. Discussion

ARVC is an inherited myocardial disease which predominantly affects the right ventricle (RV) and is characterized pathologically by gradual replacement of myocytes by fibrous tissue that can result in arrhythmia, sudden cardiac death (SCD), and heart failure.

ARVC was first described in 1977 in a report of six patients with sustained VT and enlarged RV [1]. It is most commonly inherited as an autosomal dominant trait with variable penetrance ranging from 20-25% of family members [2,3], with the first chromosomal locus identified at 14q23-q24 after clinical evaluation of a large Italian family [4]. An autosomal recessive variant (also called Naxos Disease) has been described in which there is a cosegregation of cardiac (ARVC), skin (palmoplantar keratosis), and hair (woolly hair) abnormalities and has been mapped on chromosome 17 (locus 17q21) [5]. The

etiology of this malignant form was found to be a two - base pair deletion in the gene encoding the desmosomal protein plakoglobin [6], a major component of cell adhesion junction. This stimulated research for the elucidation of desmoplakin gene which has been associated with the more common autosomal form [7]. Diminished level of plakoglobin showed a 91% sensitivity, 82% specificity, 83% positive predictive value and 90% negative predictive value, implying that a reduced immuno reactive signal of plakoglobin at the intercalated disk is a consistent feature in patients with ARVC [8]. Sixty eight percent of myocardial samples with histomorphologic manifestations of ARVC displayed decreased plakoglobin staining which might serve as an additional diagnostic marker of ARVC in forensic pathology [9].

The prevalence of the disease is estimated to affect 1 in 1000-5000 population and is more common in individuals of Greek and Italian origin [10]. In the United States, the median age at presentation was 26 years (51% were

males), the median time to diagnosis was one year from the initial presentation, and the median survival was 60 years [11].

The diagnosis of the disease is established on the proposed standardized criteria based upon the identification of structural, histological, electrocardiographic, arrhythmic, and familial features, which were further subclassified into major and minor criteria [12] that would categorize it further into definite, borderline or possible diagnosis. Biomarkers have been studied including 1.64 fold rise in heat shock protein 70 (HSP70) in ARVC failing hearts compared with non-failing hearts [13], phasic elevation of troponin I with normal coronary angiogram in an ARVC patient who presented with symptomatic VT terminated by ICD discharges [14], and a recently proposed early biomarker MiR-320 which is significantly down-regulated in ARVC patient's plasma [15].

A large series at a tertiary referral center reported the frequency of the principle symptoms associated with ARVC as follows: palpitations (67%), syncope (32%), atypical chest pain (27%), dyspnea (11%), and right ventricular failure (6%) [16]. Patients typically present with palpitations or syncope as the manifestation of ventricular arrhythmias which can range from frequent ventricular premature beats to sustained VT [11]. Sustained or nonsustained monomorphic VT is the most common ventricular arrhythmia and it originates in the RV and therefore has a left bundle branch block (LBBB) pattern [11,17].

SCD exist in patients with ARVC and can be its initial presentation [11,17,18,19]. A forensic autopsy review for SCD revealed 10.4% was associated with ARVC with 75% occurred during routine daily activities, 10% percent during the perioperative period, and 3.5% while participating in sports [20].

An indispensable initial approach is to obtain a 12-lead electrocardiogram (ECG) and transthoracic echocardiography in all patients with a suspected diagnosis of ARVC. Additional testing had been recommended based on the clinical scenario or when the results of initial testing are non-diagnostic and may include one or more of the following: signal-averaged ECG (SAECG), 24-hour ECG monitoring, exercise ECG testing, cardiac magnetic resonance imaging, right ventriculography, endomyocardial biopsy, electrophysiological study, and genetic testing [21].

The presence of arrhythmia (nonsustained ventricular tachycardia, VT or frequent and complex premature ventricular contractions) or symptoms would entail treatment initiation with beta-blockers or implantable cardioverter-defibrillator (ICD) with additional medical therapy to prevent defibrillator discharge [22]. Sotalol prevented VT during programmed ventricular stimulation in 68% of the patients, whereas class Ia and Ib drugs were effective in only 5.6% and class Ic drugs in only 2% of patients [23].

The 2006 ACC/AHA/ESC guidelines for the management of ventricular arrhythmias and SCD recommended that sotalol or amiodarone may be effective therapies for the treatment of sustained VT or VF in patients with ARVC in whom ICD implantation is not feasible [24]. Radiofrequency ablation is not a definitive therapy due to the variable and progressive nature of ARVC however successful treatment

of some of the arrhythmogenic foci can be effected by it [25,26,27].

Both the 2006 ACC/AHA/ESC guidelines for the management of ventricular arrhythmias and the prevention of SCD and the 2012 ACCF/AHA/HRS guidelines on device-based therapy for cardiac rhythm abnormalities agreed that an ICD should be implanted in patients with ARVC and documented sustained VT or VF for the primary prevention of SCD [24,28].

A meta-analysis of AVRC patients with ICD for either primary or secondary prevention determined an annual rate of cardiac death, noncardiac death, and heart transplantation as 0.9 percent, 0.8 percent, and 0.9 percent respectively [29]. Most of the AVRC patients appear to do relatively well though they do carry a risk of developing heart failure and ventricular arrhythmias.

4. Conclusion

Arrhythmogenic right ventricular cardiomyopathy is progressively regarded as an etiology of malignant ventricular arrhythmias especially among healthy young adults and in those individuals hooked to vigorous exercise. It should be taken into account by physicians in young patients with cardiac arrhythmias or unexplored cardiomyopathy. Treatment involves the abolishment of malignant arrhythmias with antiarrhythmic agents, but now, is geared towards automatic implantable defibrillator placement as the most effective modality to arrest sudden cardiac death.

List of Abbreviations

ARVC=Arrhythmogenic Right Ventricular Cardiomyopathy; VT=Ventricular Tachycardia; AICD= Automatic Implantable Cardioverter Defibrillator; RV=Right Ventricle; SCD=Sudden Cardiac Death; LBBB=Left Bundle Branch Block; ECG=Electrocardiogram; SAECG=Signal-Averaged Electrocardiogram; Implantable Cardioverter-Defibrillator(ICD); VF=Ventricular Fibrillation; ACC=American College of Cardiology; AHA=American Heart Association; ESC=European Society of Cardiology; HRS=Heart Rhythm Society.

Conflict of Interests

The authors declare that they have no conflict of interest.

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