

A case of sudden cardiac arrest in a young adult

Zaid B. Al Jebaje, John Elibol, Robbie Wall, Osman Saleem

ABSTRACT

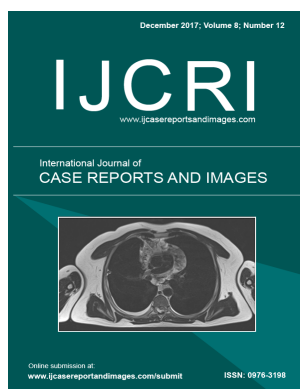
Introduction: Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited myocardial condition that primarily affects the right ventricle. The hallmark characteristic of this disease is continual loss and replacement of normal myocardium with fibrofatty tissue. This replacement of tissue can lead to life-threatening arrhythmias and potentially sudden cardiac death (SCD). Current diagnostic modalities include, electrocardiography, family history, echocardiogram, MRI scan, angiography and myocardial biopsy.

Case Report: A 27-year-old athletic female with no known past medical history collapsed while playing frisbee in the park. Upon emergency medical service (EMS) arrival, the patient was unconscious, pulseless, and in ventricular fibrillation. After a successful resuscitation, the patient was transferred to the emergency department and admitted to the ICU. Electrocardiography revealed a QT interval of 460 milliseconds and T-wave inversion in V1, V2, and V3. Transthoracic echocardiogram revealed a left ventricular ejection fraction (LVEF) of 30% along with moderate enlargement and reduced function of the right ventricle. Genetic testing showed the patient was heterozygous for a novel variant of uncertain significance in the DSC2 gene that codes for desmosomal protein desmocollin-2. Management at this time included a wearable defibrillator for 30 days, b-blockers, and abstaining from moderate and severe physical activity. The patient then received a single chamber subcutaneous intracardiac device (ICD) and was counseled on avoiding strenuous physical exertion. Six months later, she received an implantable ICD. The patient's first degree family members were all offered screenings.

Conclusion: This case demonstrates the complex workup involved as well as the therapeutic options for patients with ARVC. This case also highlights the importance of counseling, affected patients and unaffected carriers, as well as screening of first-degree relatives in hopes of preventing serious unwanted outcomes.



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CASE REPORT

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Keywords: Arrhythmia, Arrhythmogenic right ventricular cardiomyopathy, Cardiomyopathy, Sudden cardiac arrest, Sudden cardiac death

How to cite this article

Al Jebaje ZB, Elibol J, Wall R, Saleem O. A case of sudden cardiac arrest in a young adult. Int J Case Rep Images 2017;8(12):811–816.

Article ID: Z01201712CR10867ZA

doi:10.5348/ijcri-2017128-CR-10867

INTRODUCTION

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited myocardial condition that

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Received: 21 September 2017

Accepted: 23 October 2017

Published: 01 December 2017

primarily affects the right ventricle. The prevalence of ARVC is approximately 1 in 2000 to 1 in 5000 in the general population and generally affects men more than women at a ratio of 3:1 [1, 2]. The hallmark characteristic of this disease is continual loss and replacement of normal myocardium with fibrofatty tissue [3]. This replacement of tissue can lead to life-threatening arrhythmias and potentially sudden cardiac death (SCD). Despite the name, variations of ARVC can also affect the left side of the heart. In some cases, left-sided arrhythmias as well as left-sided heart failure may occur. Approximately 30–50% of cases of ARVC are considered familial and can be inherited in autosomal dominant or autosomal recessive forms. However, the autosomal dominant form is more common [4]. The pathogenesis is believed to be linked to over 30 genes that code for proteins involved in desmosomal structures between cardiac cells. The most known affected proteins are desmoplakin (DSP) [5], plakophilin 2 (PKP2) [6], desmoglein 2 (DSG2) [7], and desmocollin 2 (DSC2) [8]. These mutations cause a weakening in cell to cell

mediated adhesions within the myocardium and lead to fibro-fatty replacement of normal ventricular tissue. Consequently, intense physical activity is restricted in these patients because of the increased risk of arrhythmia and sudden cardiac arrest/death.

What makes this diagnosis challenging is that many of the patients suffering from ARVC are young, athletic individuals, who seem overtly healthy. The most common presenting symptoms can include syncope, cardiac ischemia, arrhythmia, sudden cardiac arrest or sudden cardiac death (SCD). The original diagnostic criteria, which was established in 1994, was revised in 2010 (Table 1). In 1994, a diagnosis of ARVC had to include either 2 major, 1 major + 2 minor, or 4 minor features. When the criteria were revised in 2010, three categories were introduced. They included, definite (2 major or 1 major + 2 minor), borderline (1 major + 1 minor or 3 minor), or possible (1 major or 2 minor) features for the diagnosis of ARVC. The purpose of this revision was to increase the sensitivity of the criteria

Table 1: Diagnostic criteria

1994 Task Force Criteria	2010 Task Force Criteria
2 major, 1 major + 2 minor, or 4 minor	Definite = 2 major or 1 major + 2 minor Borderline = 1 major + 1 minor or 3 minor Possible = 1 major or 2 minor
I. Global/regional dysfunction/structural alterations	
Major	By 2D echo
- Severe dilatation and reduction of RVEF with/without (or only mild) left ventricular impairment - Localized right ventricular aneurysms (akinetic or dyskinetic areas with diastolic bulging) - Severe segmental dilatation of the right ventricular	- Regional right ventricular akinesia, dyskinesia, or aneurysm - and one of the following (end diastole): - PLAX RVOT ≥ 32 mm (correct for body size [PLAX/BSA] ≥ 19 mm/m²) - PSAX RVOT ≥ 36 mm (correct for body size [PSAX/BSA] ≥ 21 mm/m²) - fractional area change $\leq 33\%$
	By magnetic resonance imaging scan
	- Regional right ventricular akinesia or dyskinesia or dyssynchronous right ventricular contraction - and one of the following: - Ratio of right ventricular end-diastole volume (RVEDV) to body surface area (BSA) ≥ 110 mL/m² (male) or ≥ 100 mL/m² (female) - RV ejection fraction $\leq 40\%$
	By right ventricular angiography
	Regional right ventricular akinesia, dyskinesia, or aneurysm
Minor	By 2D echo
- Mild global right ventricular dilatation and/or ejection fraction reduction with normal left ventricular - Mild segmental dilatation of the right ventricular - Regional right ventricular hypokinesia	- Regional right ventricular akinesia or dyskinesia - and one of the following (end diastole): - PLAX RVOT ≥ 29 to < 32 mm (correct body size PLAX/BSA ≥ 16 to < 19 mm/m²) - PSAX RVOT ≥ 32 to < 36 mm (correct body size [PSAX/BSA] ≥ 18 to < 21 mm/m²) - fractional area change $> 33\%$ to $\leq 40\%$
	By magnetic resonance imaging scan
	- Regional right ventricular akinesia or dyskinesia or dyssynchronous right ventricular contraction - and one of the following: - Ratio of RVEDV to BSA ≥ 100 to < 110 mL/m² (male) or ≥ 90 to < 100 mL/m² (female) - Right ventricular ejection fraction (RVEF) > 40–$\leq 45\%$

II. Tissue characterization of wall

Major

- Fibrofatty replacement of myocardium on endomyocardial biopsy

- Residual myocytes <60% by morphometric analysis (or <50% if estimated), with fibrosis replacement of right ventricular free wall myocardium in ≥ 1 sample, with or without fatty replacement of tissue on endomyocardial biopsy

Minor

- Residual myocytes 60–75% by morphometric analysis (or 50–60% if estimated) with fibrous replacement of the right ventricular free wall in ≥ 1 sample, with or without fatty replacement of tissue on endomyocardial biopsy

III. Repolarization abnormalities

Major

- Inverted T-waves (V_1 , V_2 , V_3) or beyond; >14 years; in absence of complete RBBB QRS ≥ 120 ms

Minor

- Inverted T-waves in right precordial leads (V_2 and V_3) (people age >12 years, in absence of RBBB)
- Inverted T-waves in V_1 and V_2 ; >14 years; in absence of complete RBBB or in V_4 , V_5 , or V_6
- Inverted T-waves in V_1 – V_4 ; >14 years; in presence of complete RBBB

IV. Depolarization/conduction abnormalities

Major

- Epsilon waves or localized prolongation (>110 ms) of QRS complex in right precordial leads (V_1 to V_3)
- Epsilon wave (reproducible low-ampere signals between the end of QRS complex to onset of T wave) in right precordial leads (V_1 – V_3)

Minor

- Late potentials signal-averaged electrocardiogram (SAECG)
- Late potentials by signal-averaged electrocardiogram in ≥ 1 of 3 parameters in absence of QRS duration of ≥ 110 ms on electrocardiography
 - Filtered QRS duration (fQRS) ≥ 114 ms
 - Duration of terminal QRS <40 μ V (low amplitude signal duration) ≥ 38 ms
 - Root mean square voltage of terminal 40 ms ≤ 20 μ V
- Terminal activation duration of QRS ≥ 55 ms measured from nadir of S wave to end of QRS, including R', in V_1 , V_2 , or V_3 , in absence of complete RBBB

V. Arrhythmias

Major

- NSVT or sustained ventricular tachycardia of LBBB morphology with superior access (negative or indeterminate QRS in leads II, III, and aVF and positive in lead aVL)

Minor

- LBBB sustained or NSVT (ECG, Holter, Exercise tolerance test)
- >1000 ventricular extrasystoles per 24 hours (Holter)
- NSVT or sustained ventricular tachycardia of right ventricular outflow configuration, LBBB morphology with inferior access (positive QRS in leads II, III, and aVF and negative in lead aVL) or of unknown axis
- >500 ventricular extrasystoles per 24 hours (Holter)

VI. Family History

Major

- Familial disease confirmed at necropsy or surgery
- ARVC/D confirmed in false discovery rate who meets task force criteria
- ARVC/D confirmed pathologically at autopsy or surgery in false discovery rate
- Pathogenic mutation (associated or probably associated with ARVC/D) in patient under evaluation

Minor

- Family history of sudden death (<35 years) due to suspected ARVC/D
- History of ARVC in false discovery rate in whom not possible or practical to determine if family member meets task force criteria
- Premature sudden death (<35 years) due to suspected ARVC/D in false discovery rate
- ARVC/D confirmed pathologically or by current task force criteria in second-degree relative
- Familial history (clinical diagnosis based on present criteria)

Source: With permission from Hopkinsmedicine.org

Abbreviations: RVEF: Right Ventricular Ejection Fraction, LBBB: Left Bundle Branch Block, RBBB: Right Bundle Branch Block, PLAX: Parasternal Long Axis, RVOT: Right Ventricular Outflow Tract, NSVT: Nonsustained Ventricular Tachycardia, ECG: Electrocardiography; ARVC/D: Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia

and to increase the likelihood affected family members being identified before a cardiac event. The criterion was also revised in part to change in genetic testing and technological advances in medicine. If a patient survives the initial event, current diagnostic modalities include electrocardiography, family history, echocardiogram, magnetic resonance imaging scan, angiography and myocardial biopsy.

CASE REPORT

A 27-year-old athletic female with no known past medical history collapsed while playing frisbee in the park. Upon emergency medicine services (EMS) arrival, the patient was unconscious, pulseless, and in ventricular fibrillation. Cardiopulmonary resuscitation efforts were initiated and lasted 30 minutes. The patient received 8 DC shocks as well as on-scene intubation. Resuscitation attempts were successful and the patient was brought to the emergency department and transferred to the ICU.

Initial electrocardiography (Figure 1) showed a QT interval of 460 ms and T-wave inversion in V1, V2, and V3. Transthoracic echocardiogram revealed a left ventricular ejection fraction (LVEF) of 30% along with moderate enlargement and reduced function of the right ventricle. The patient was successfully extubated four days after admission. Physical examination at that time was unremarkable. Follow-up transthoracic echocardiogram revealed a preserved LVEF of 55%. Cardiac catheterization was negative for coronary disease. The initial read of the cardiac MRI scan focused specifically on the left ventricle and was unremarkable. Genetic testing showed the patient was heterozygous for a novel variant of uncertain significance in the DSC2 gene that codes for desmosomal protein desmocollin-2, the pathogenic variants of which are found in autosomal dominant forms of ARVC. After the genetic study was performed, cardiac MRI scan with and without contrast (Figure 2) was read for a second time with a focus on the right ventricle. This revealed mild dilatation of the right ventricle, an elevated right ventricular end diastolic volume index of 111 mL/m² (normal range 60–100 mL/m²), a decreased right ventricular ejection fraction (RVEF) of 36% (normally 60%), and no evidence of regional wall defects. Electrocardiography changes as well as the mutation in the DSC2 gene are consistent with two of the major criteria of ARVC and support a definitive diagnosis. Management at this time included a wearable defibrillator for 30 days, β -blockers, and abstaining from moderate and severe physical activity. The patient then received a single chamber subcutaneous intracardiac device (ICD) and was counseled on avoiding strenuous physical exertion. Six months later, she received an implantable ICD and has been following-up with our cardiac clinic. The patients first degree family members were all offered screenings.

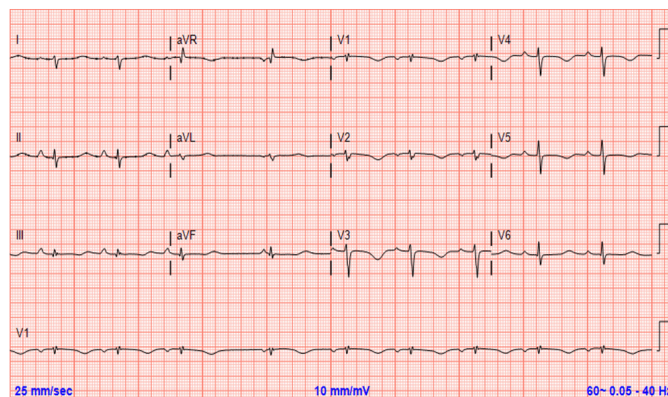


Figure 1: Electrocardiography showing T-wave inversion in V1, V2, and V3.

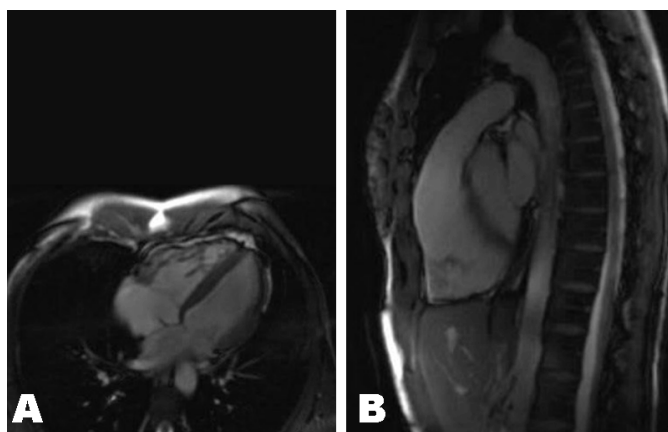


Figure 2 (A–B): Cardiac magnetic resonance imaging scan showing mild dilatation of the right ventricle, an elevated right ventricular end-diastolic volume index of 111 mL/m² (normal range 60–100 mL/m²), a decreased right ventricular ejection fraction of 36% (normally 60%), and no evidence of regional wall defects.

DISCUSSION

The diagnosis of arrhythmogenic right ventricular cardiomyopathy can be extremely challenging. This is because many patients are young and healthy adults before a symptomatic event ensues, usually precipitated by exercise. Unfortunately, by the time an event occurs, it may be too late because the most common presenting symptoms are cardiac ischemia, syncope, arrhythmia, or sudden cardiac arrest/death. ARVC accounts for 17% of cases of sudden cardiac death in the United States [9]. By definition our case represented a definitive diagnosis of ARVC due to two characteristics of the major criteria being met. This included T-wave inversion in V1–V3 with absence of right bundle branch block and mutation in desmocollin-2, which has a genetic association with ARVC [8, 10]. Restricting strenuous physical activity is imperative for both affected individuals as well as healthy genetic carriers in order to prevent exercise induced cardiac events and progression of disease [11, 12]. Additionally, clinical surveillance every two years and abstinence from intense physical activity is recommended

in individuals with either unknown genotypes or in individuals who are known carriers of the diseases. Although there is no cure for ARVC, the name of the game is symptom management and prevention of arrhythmia or sudden cardiac death. This can be achieved with medical therapy that includes beta-blockers, diuretics, ace-inhibitors or angiotensin II receptor antagonists. Other therapies include endo/epi cardiac ablation or an intracardiac device. If these regimens prove to be suboptimal in symptom control, cardiac transplant is an alternative option. Ultimately, ARVC is a serious and complex cardiomyopathy that can result in ventricular arrhythmias and sudden cardiac death. The ARVC has an underestimated prevalence and is a challenging diagnosis that requires a high index of suspicion. Although many patients including the one discussed in this case do not show evidence of right ventricular damage, eventually all patients will. As such, diligent follow-up is imperative when treating patients with ARVC. Many aspects of ARVC have yet to be discovered and unfortunately no cure has been found to date.

CONCLUSION

This case demonstrates the complex workup involved in cases of (ARVC) as well as the therapeutic options for these patients. This case also highlights the importance of counseling, affected patients and unaffected carriers, as well as screening of first-degree relatives in hopes of preventing serious unwanted outcomes.

Author Contributions

Zaid B. Al Jebaje – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
John Elibol – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Robbie Wall – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Osman Saleem – Substantial contributions to conception and design, Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor of Submission

The corresponding author is the guarantor of submission.

Source of Support

None

Conflict of Interest

Authors declare no conflict of interest.

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