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Arrhythmogenic Right Ventricular Dysplasia Presenting with Sustained Ventricular Tachycardia: A Case Report

F. Kaddari [a*], H. Rouam [a], Y. Eshaq [a], M. Eljamili [a] and M. Elhattaoui [a]

[a] Cardiology Department, Errazi Hospital, University Hospital Mohammed VI, Marrakesh, Morocco.

Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Arrhythmogenic right ventricular dysplasia (ARVD) is a cardiomyopathy characterized pathologically by fibrofatty tissue replacement of the myocyte of the right ventricle (RV) and clinically by life-threatening ventricular arrhythmias in young people**.** It is a major cause of sudden death**.** We present the case of a 60-year-old man with cardiovascular risk factors, was admitted for–unstable ventricular tachycardia (VT) treated immediately with synchronized cardioversion. After the stabilization of the patient, Electrocardiogram demonstrated an epsilon wave in precordial leads and diffuse T-wave inversions. Transthoracic echocardiography revealed a dilated, hypokinetic right ventricle with moderately reduced function and a focal area of dyskinesia. The diagnosis of arrhythmogenic right ventricular dysplasia (ARVD) was made and an implantable cardioverter defibrillator (ICD) was indicated for secondary prevention. This case report will present the clinical presentation, diagnosis and management of this rare disease.

**Corresponding author: E-mail: Kaddari.FZ@Gmail.com, kaddari.fz@gamil.com;*

Keywords: Arrhythmogenic right ventricular dysplasia; implantable cardioverter defibrillator; sudden death.

1. INTRODUCTION

Arrhythmogenic right ventricular Dysplasia (ARVD). Is a rare inherited disorder usually affecting the right ventricle (RV), characterized by fibro-fatty tissue replacement of the healthy ventricular myocardium. It often predisposes young patients to ventricular tachycardia, heart failure, and/or sudden cardiac death [1]. Its diagnosis is often difficult, based on a combination of clinical, electrical, morphological, and histological findings. Doppler echocardiography is a reliable and efficient investigation for the diagnosis of ARVD. It remains on the first intention to review a suspected ARVD [2].

2. CASE REPORT

A 60-year-old man with cardiovascular risk factors such as chronic smoking at 15 Pack years and dyslipidemia. He was admitted for hemodynamically unstable sustained ventricular tachycardia (VT); the mean ventricular rate was at 195 bpm with superior axis (Negative QRS in II, III, and aVF leads, and positive in aVL lead, mid -ventricular inferior-septal location using the KUCHAR algorithm [3] (Fig. 1). The patient was treated immediately with synchronized cardioversion. A coronary angiography was performed excluding the ischemic etiology of the ventricular tachycardia.

After the stabilization of the patient the electrocardiogram showed a normal sinus rhythm at 50bpm, suspected epsilon waves in V2, V3, and V4, intraventricular block, and negative T waves in the anterior, lateral, and inferior territories [4] (Fig. 2)

At the F -EKG realized to our patient, we found several patterns of epsilon waves; the wiggle waves pattern in the FIII lead, and small spike waves p in FI and FII lead (Fig. 3).

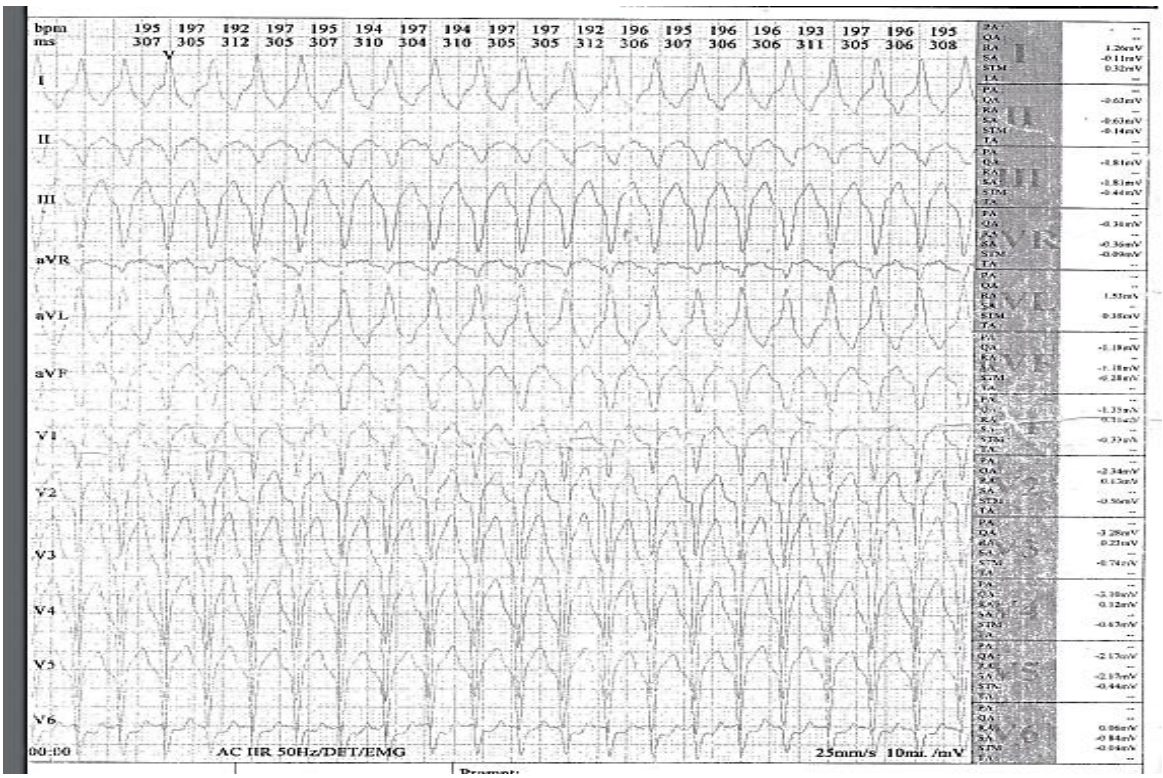


Fig. 1. 12-lead Electrocardiogram showing sustained Ventricular tachycardia

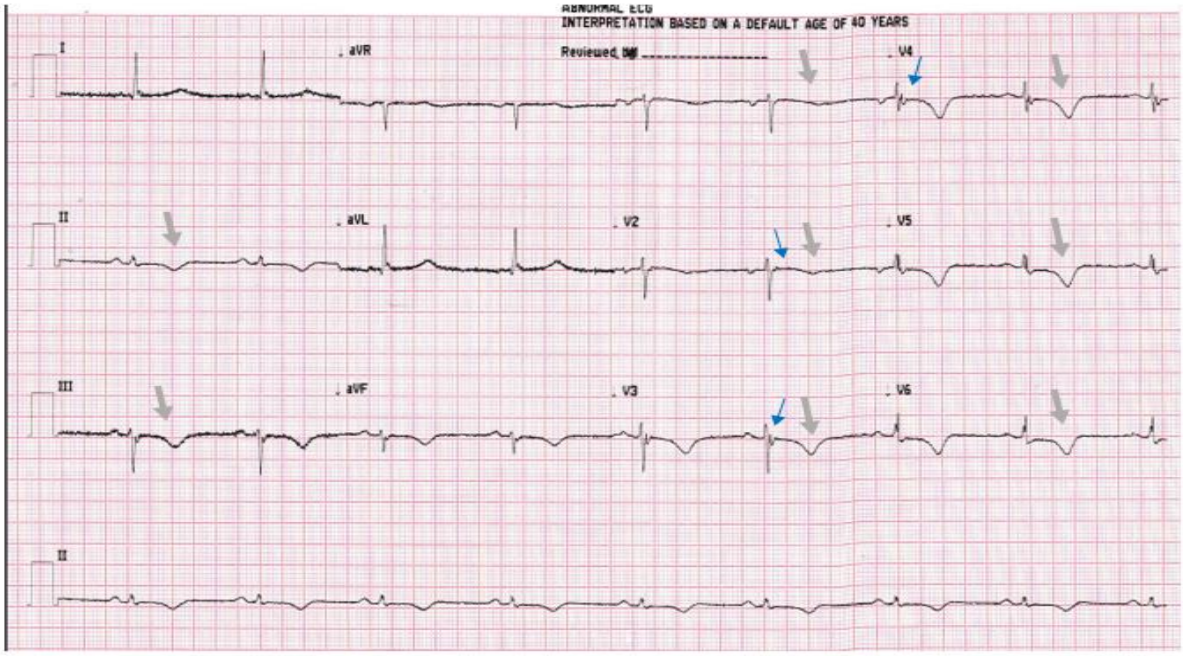


Fig. 2. 12-Lead Electrocardiogram showing Epsilon waves in V2, V3, and V4 (blue arrows) and negative T waves in the anterior, lateral, and inferior territories (grey arrows)

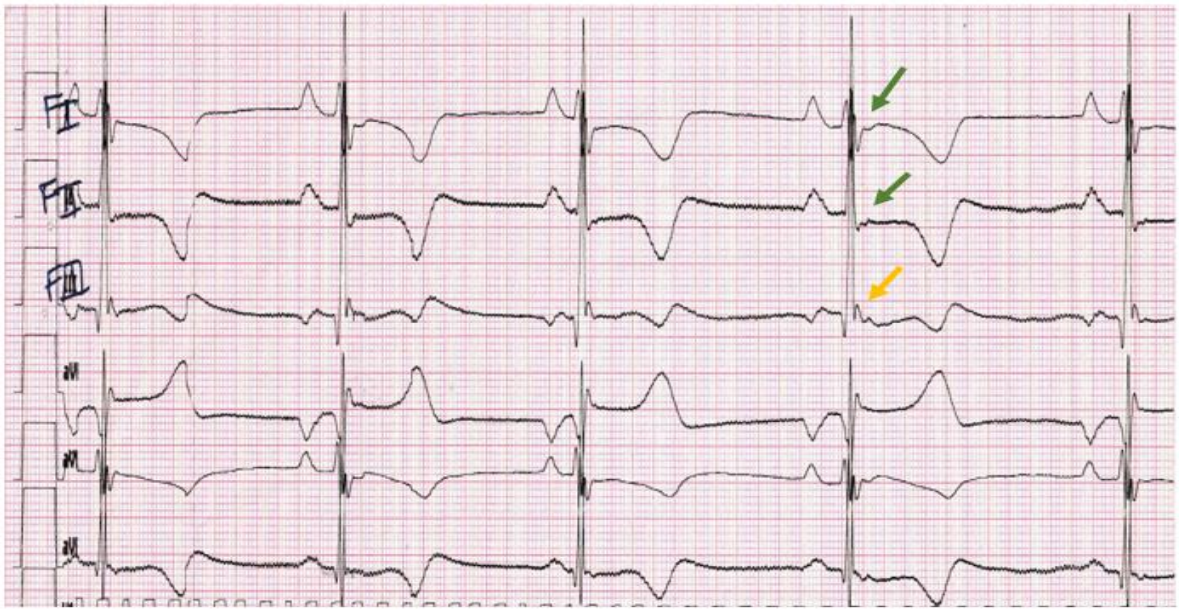


Fig. 3. Fontaine bipolar precordial leads (F-ECG) showing patterns of the epsilon Waves: Wiggle waves in FIII lead (yellow arrow) and small spike waves in FI and FII lead (green arrow)

Transthoracic echocardiography revealed moderate dilatation of the Right ventricle (RV), depressed RV systolic function with segmental kinetic disorders as anterior wall akinesia; lateral wall and apex hypokinesia (Fig. 4). However, the left ventricle size and systolic function were preserved.

24-Hour Holter Monitoring showed basic sinus rhythm. The average ventricular rate was at 50 beats per minute, with epsilon wave, presence of some Premature Ventricular Complex (PVC) with a left inferior axis delay without ventricular tachycardia or conduction disorder (Fig. 5). We found 3 patterns of epsilon waves: wiggle waves,

small spike waves, and smooth potential waves with the QRS duration in V_1 exceeding the QRS duration in V_3 by at least 25 msec. The small spike waves were divided into 2 subtypes, one upward and the other downward.

The diagnosis of ARVD was made based on EKG features and structural abnormalities seen on echocardiography according to Task Force Criteria [5] (Fig. 6).

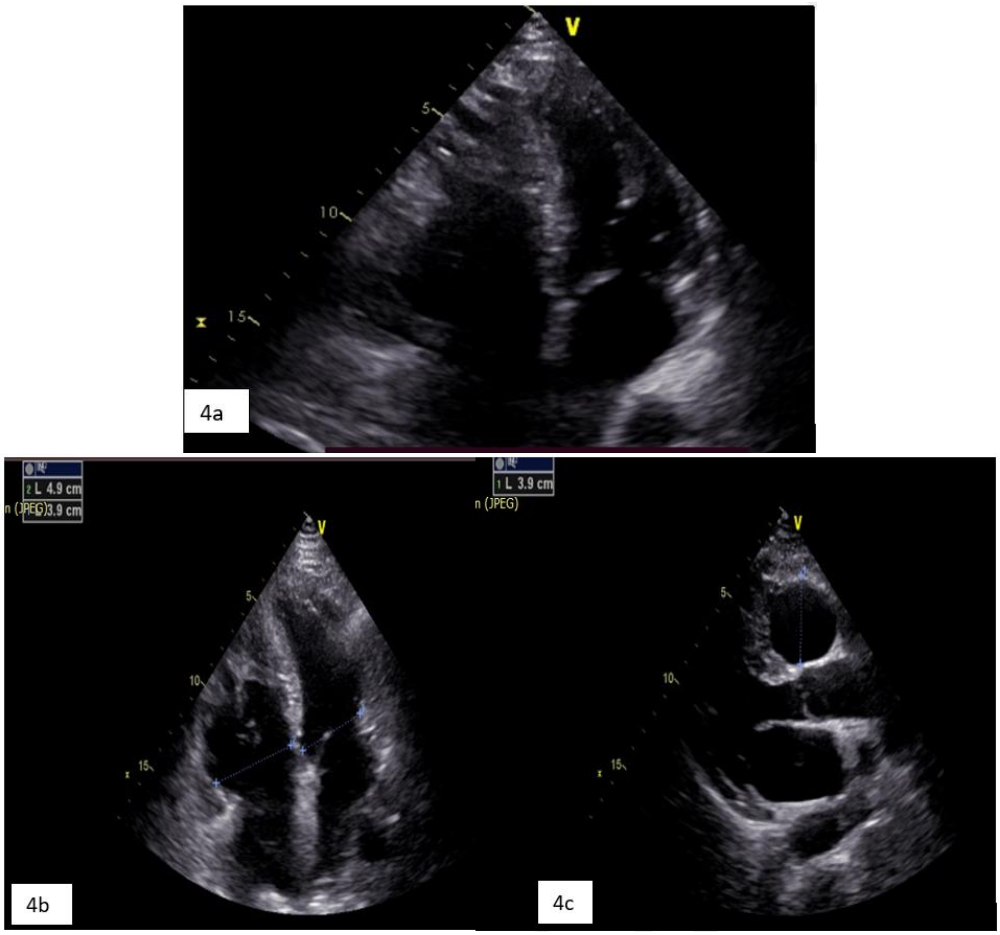
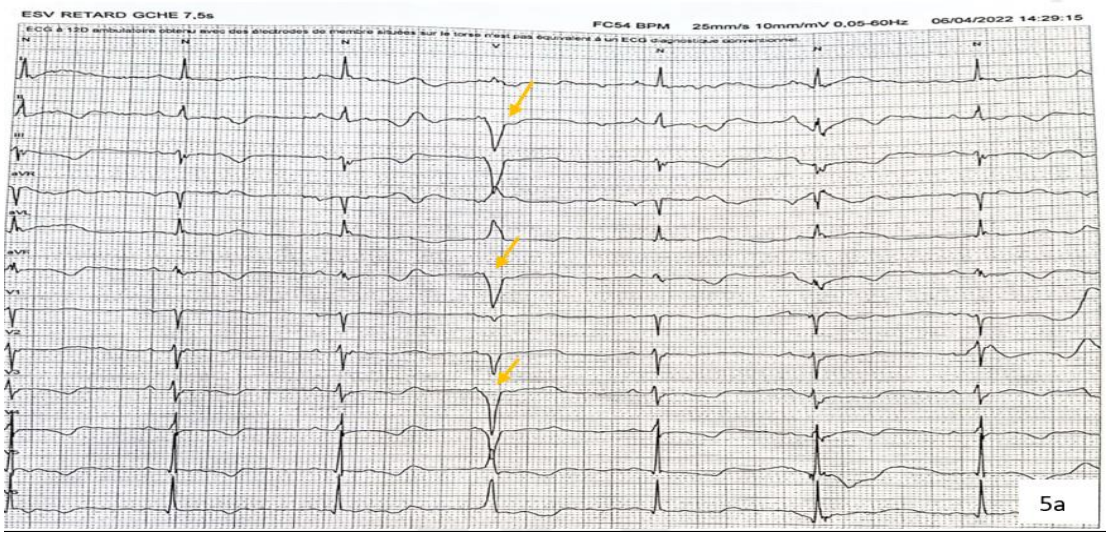


Fig. 4. (4a) Apical Four -chamber view showing apical trabeculations. (4b) Apical four-chamber and (4c) Parasternal long axis echocardiographic view (PLAX) showing RV dilatation



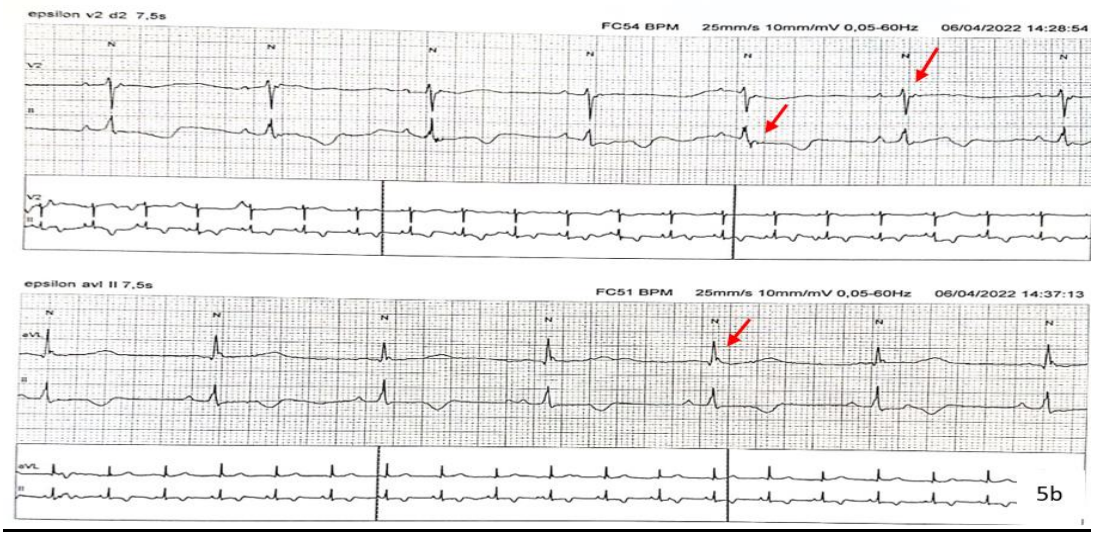


Fig. 5. 24-Hour Holter Monitoring with PVC (Yellow arrows) (5a) and Epsilon Waves (5b) (Red arrows)

	Diagnostic criteria
1-Morpho-functional ventricular abnormalities	**Major criteria** **-** Akinesia associated with dilatation of the RV and global RV dysfunction PLAX RVOT =38 mm , PSAX RVOT= 39 mm, Fractional area change =19% **Minor criteria** **-** Segmental akinesia of the RV
2-Repolarisation anomalies	**Major criteria:** - Inverted T waves in the right pre-cordium (V1, V2 and V3) **Minor criteria:** -Inverted T waves in the left precordium (V4-V6) (LEFT VENTRICLE)
3-Depolarisation abnormalities	**Minor Criteria** **-** Epsilon wave in the right precordial leads of V1 to V3
4- Arrhythmias	**Major criteria** -Sustained ventricular tachycardia (VT) with superior axis (Negative QRS in II, III and aVF leads and positive in aVL lead).

Fig. 6. The diagnostic criteria of ARVD in our case

PLAX: Parasternal Long Axis Echocardiographic
RVOT: View Right Ventricular Outflow Tract
PSAX: Parasternal short-axis views

The patient was treated with sotalol 80 mg and an implantable cardioverter defibrillator (ICD) was recommended for secondary prevention. In addition, molecular genetic analysis was proposed, and a cardiological evaluation for his first-degree relatives. The patient was fine at a three-month follow-up; no sustained VT reoccurred.

3. DISCUSSION

Histologically, the characteristic of ARVD is the replacement of the right ventricular myocardium by fibrous and adipose tissue. The fibrous cicatricial tissue progresses from the epicardium to the endocardium and principally involves the free ventricular wall, leading to thinning of the free ventricular wall and aneurysmal dilatation. The left ventricle (LV) is less affected than the right ventricle (RV). The origin of these structural changes is genetic. The most common clinical presentations are palpitations and syncope. These symptoms are in turn linked to the presence of non-sustained or sustained ventricular arrhythmias. Up to 19% of ARVD patients present as cardiac arrest [6]. However, it is a progressive disease, ultimately leading to RV and late-stage biventricular failure [7,8].

Modified Task Force Criteria for ARVC – Diagnostic Categories Major and Minor Criteria		
Definite: 2 major OR 1 major and 2 minor, OR 4 minor criteria from different categories Borderline: 1 major and 1 minor, OR 3 minor criteria from different categories Possible: 1 major, OR 2 minor criteria from different categories		
	Major	Minor
Global or regional dysfunction and structural alterations determined by echo, MRI, or RV angiography:		
Echo	Regional RV akinesia, dyskinesia, or aneurysm and 1 of the following (end diastole): a) PLAX RVOT ≥32 mm (PLAX/BSA ≥19 mm/m^2) b) PSAX RVOT ≥36 mm (PSAX/BSA ≥21 mm/m^2) c) Fractional area change ≤33%	Regional RV akinesia, dyskinesia, or aneurysm and 1 of the following (end diastole): a) PLAX RVOT ≥29 mm to <32 mm (PLAX/BSA ≥16 to <19 mm/m^2) b) PSAX RVOT ≥32 to <36 mm (PSAX/BSA ≥18 to <21 mm/m^2) c) Fractional area change >33 to ≤40%
MRI	Regional RV akinesia or dyskinesia or dyssynchronous RV contraction and 1 of the following: a) Ratio RVEDV/BSA ≥110 mL/m^2 (male), ≥100 mL/m^2 (female) b) RVEF ≤40%	Regional RV akinesia or dyskinesia or dyssynchronous RV contraction and 1 of the following: a) Ratio RVEDV/BSA ≥100 to <110 mL/m^2 (male), ≥90 to 100 mL/m^2 (female) b) RVEF >40 to ≤45%
RV angiography	Regional RV akinesia, dyskinesia, or aneurysm	
Tissue characterization of wall		
Endomyocardial biopsy showing fibrous replacement of the RV free wall myocardium in ≥1 sample, with or without fatty replacement and with:	Residual myocytes <60% by morphometric analysis (or <50% if estimated)	Residual myocytes 60% to 75% by morphometric analysis (or 50% to 65% if estimated)
Repolarization abnormalities		
ECG	Inverted T waves in right precordial leads (V_1, V_2, and V_3) or beyond in individuals >14 years of age (in the absence of complete RBBB QRS ≥120ms)	I. Inverted T waves in leads V_1 and V_2 in individuals >14 years of age (in the absence of complete RBBB) or in V_4, V_5, or V_6. II. Inverted T waves in leads V_1, V_2, V_3, and V_4 in individuals >14 years of age in the presence of complete RBBB
Depolarization/conduction abnormalities		
ECG	Epsilon wave (reproducible low-amplitude signals between end of QRS complex to onset of the T wave) in the right precordial leads (V_1 to V_3)	I. Late potentials by SAECG in ≥1 of 3 parameters in the absence of QRS duration of ≥110ms on the standard ECG: a) Filtered QRS duration (fQRS) ≥114 ms b) Duration of terminal QRS <40 µV (low-amplitude signal duration) ≥38 ms c) Root-mean-square voltage of terminal 40 ms ≤20 µV II. Terminal activation duration of QRS ≥55 ms measured from the nadir of the S wave to the end of the QRS, including R' in V_1, V_2, or V_3 in the absence of complete RBBB
Arrhythmias		
	Nonsustained or sustained VT of LBBB with superior axis (negative or indeterminate QRS in leads II, III, and aVF and positive in lead aVL)	I. Nonsustained or sustained VT or RV outflow configuration, LBBB morphology with inferior axis (positive QRS in II, III and aVF and negative in lead aVL) or of unknown axis II. >500 ventricular extrasystoles per 24 hours (Holter)
Family history		
	I. ARVC confirmed in a first-degree relative who meets current Task Force Criteria II. ARVC confirmed pathologically at autopsy or surgery in a first-degree relative III. Identification of a pathogenetic mutation categorized as associated or probably associated with ARVC in the patient under evaluation	I. History of ARVC in a first-degree relative in whom it is not possible or practical to determine whether the family member meets current Task Force Criteria II. Premature sudden death (<35 years of age) due to suspected ARVC in a first-degree relative III. ARVC confirmed pathologically or by current Task Force Criteria in second-degree relative

Fig. 7. Modified Task Force Criteria for arrhythmogenic right ventricular cardiomyopathy (ARVC) showing the diagnostic categories for major and minor criteria according to the 2010 ARVC Task Force Criteria

BSA: body surface area; ECG: electrocardiogram; echo: echocardiogram; MRI: magnetic resonance imaging; PLAX: parasternal long-axis; PSAX: parasternal short-axis; RBBB: right bundle branch block; RV: right ventricle; RVEDV: right ventricular end-diastolic volume; RVEF: right ventricular ejection fraction; RVOT: right ventricular outflow tract; SAECG: signal-averaged electrocardiogram; VT: ventricular tachycardia.

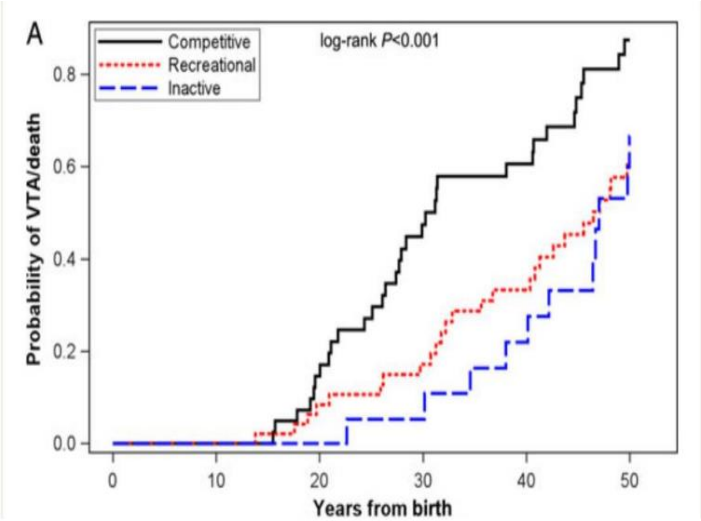


Fig. 8. Cumulative probability of VT/death since birth in probands with ARVD according to level of sport [11]

Electrocardiogram reveals T-wave inversion in the precordial leads (V1 to V4) and ventricular arrhythmias with a right bundle branch block pattern. Ventricular arrhythmias vary from premature ventricular complex to ventricular tachycardia or ventricular fibrillation (VF). The Doppler echocardiography is of great help in the study of morphological abnormalities. However, the evaluation of the segmental kinetics of the RV is subjective and difficult. The presence of focal hypokinesia associated or not with parietal thinning is specific to ARVD [9]. Then, The MRI can be used to evaluate the degree of dilatation and contractile function of the RV, to look for abnormal areas of contraction as well as contraction and the presence of fibrosis.

The diagnosis of ARVD is based on a combination of arguments. A diagnostic score has been established, including major (2 points) and minor (1 point) criteria, considering EKG, paraclinical, personal, and family parameters [10]. The table (Fig. 7) summarizes modified Task Force Criteria for arrhythmogenic right ventricular cardiomyopathy (ARVD) [5].

A molecular genetic analysis must be proposed to a proband in whom the diagnosis is confirmed or in case of high clinical suspicion (definite or probable diagnosis according to the task force criteria), as well as a cardiological evaluation for first-degree relatives. Then, genotyping is recommended to family members to identify genetically affected individuals in the preclinical phase [9].

The evolution is characterized by the emergence of arrhythmic events that can lead to sudden death and by an alteration of the bi-ventricular systolic function that can also lead to death by heart failure. The goal of treatment is to reduce the risk of sudden death and improve quality of life by improving the symptoms of arrhythmia and heart failure. Restricting intensive sports activities is imperative for patients with non-symptomatic mutations and patients with clinical symptoms [11]. Ruwald et al. found that competitive and recreational sports participation are associated with cardiac events in patients with arrhythmogenic right ventricular cardiomyopathy (Fig. 8) [11,12]. For a patient with ARVD and non-tolerated sustained VT, an ICD is recommended (Class I recommendation) [5]. Prophylactic anticoagulation for the primary prevention of thromboembolism based on global or regional ventricular dilatation/dysfunction, as in our patient's case, is not recommended (class III recommendation) [5].

4. CONCLUSION

The diagnosis of ARVD is difficult due to the lack of specific single diagnostic criteria. Once the diagnosis is made a stratification of the rhythmic and thromboembolic risk as part of prevention as well as the genetic counselling that should be proposed to the patient.

CONSENT

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

ETHICAL APPROVAL

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

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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