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Case Report

Arrhythmogenic Right Ventricular Cardiomyopathy: From Clinical Presentation to Diagnostic and Therapeutic Challenges

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ABSTRACT

The diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC) remains challenging. Detailed echocardiography is a sensitive tool for identifying structural and functional when ARVC is suspected. A thorough assessment of cardiac magnetic resonance imaging is required to further establish the diagnosis. This case illustration aimed to broaden the awareness of right ventricular cardiomyopathy among physicians, establishing the appropriate diagnostic approaches, and sensible use of implantable cardioverter-defibrillators may help to prevent unnecessary deaths.

1. Introduction

Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) is an inherited myocardial disease that is characterized by fibro-fatty replacement of the right ventricular myocardium. The diagnosis of ARVC/D remains challenging due to the absence of particular testing and advanced genetic testing. The principal challenges for ARVC/D diagnosis include earlier detection of the disease, differential diagnosis from other arrhythmogenic diseases affecting the right ventricle, and the development of new objective electrocardiographic and imaging criteria for diagnosis. This case report will reveal our challenges in establishing ARVC/D diagnosis prior to deciding whether to choose the catheter ablation first or Implantable Cardioverter Defibrillator (ICD) as the definitive therapy.[1-3]

2. Case Illustration

A 61-year-old male patient was referred from a rural hospital because of frequent palpitation and documented ventricular tachycardia (VT). VT was LBBB morphology with a superior axis (Fig.1A) that suggested VT came from the apex of the right ventricle (RV). It could be converted by cardioversion 200 Joules and maintained by Amiodaron 3x200 mg. Palpitation had been going on for 1.5 years, indifferent to one another. Within the last 6 months, he had been admitted to the hospital eight times. His First, coronary artery disease was considered to be the main cause. Diagnostic coronary angiography was completed and the result was normal.

When he arrived at Saiful Anwar General Hospital, Malang, blood pressure was 134/78 mmHg and heart rate 77 bpm regular and strong. ECG showed sinus rhythm, left axis deviation, and inverted T waves in precordial leads (V1-V6, Fig.1B). Other physical examination was within normal limits.

This patient was suspected for arrhythmogenic right ventricular cardiomyopathy (ARVC) and undergoing several diagnostic approaches to establish the diagnosis. Echocardiography (Fig.2) demonstrated that the systolic left ventricular (LV) function was normal (LV ejection fraction 84.8% by Teich, and 81.8% by Simpsons), no LV dilatation (LVIDd 3.58 cm) with LV concentric hypertrophy. There was right atrium (RA) and RV dilatation, measured by basal diameter RV 4.31 cm and RA major/BSA 3.84; however, systolic RV function was good (TAPSE 1.74 cm). The dimension of proximal right ventricular outflow tract (RVOT) was 34 mm and distal RVOT 23 mm. No dyskinesia of basal RV and 47.7% of RV FAC (fractional area change). Echocardiography findings supported the minor criteria of ARVC diagnosis.

Cardiac magnetic resonance (CMR) imaging was highly suggested. We used 'black-blood' spin echo technique with Gadolinium

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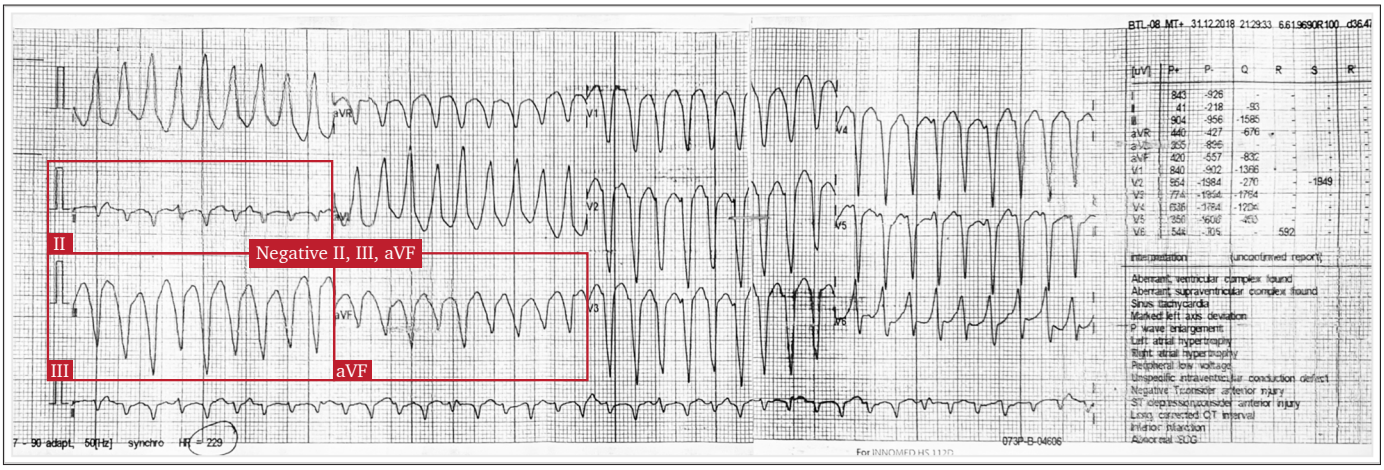
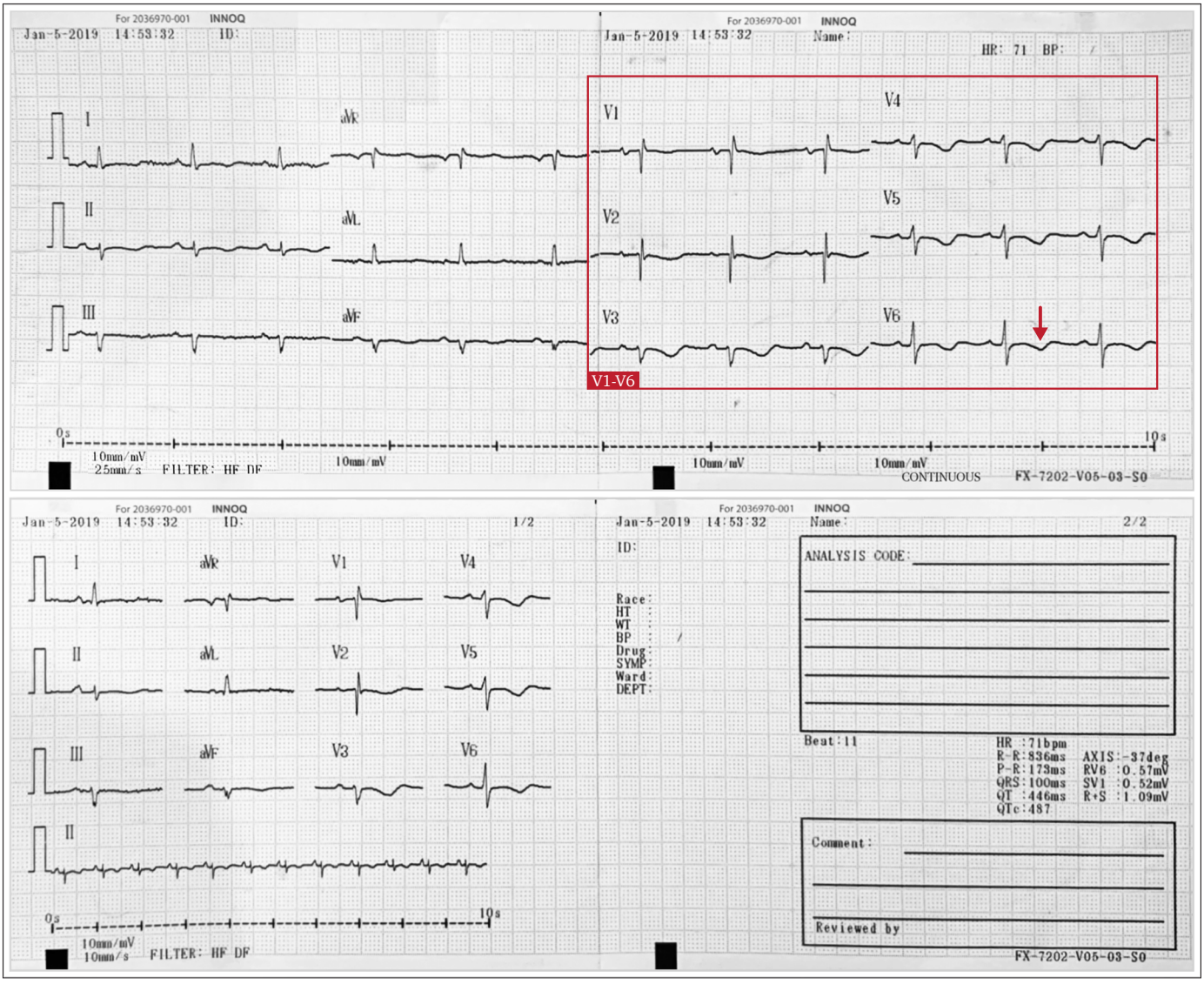
contrast. The results (Fig.3) included dilatation of RV without dilatation of RVOT (± 2.8 cm), aneurysmal outpouching with accordion sign at RV free wall during systolic and diastolic phase, and thinning of diaphragm wall of RV. There was fatty infiltration – with extensive late enhancement of Gadolinium at RV free wall and one segment of left ventricle (LV). Dyskinesia of RV free wall and diaphragm wall also showed. RV ejection fraction was 20.9%.

This patient was then advised for ICD implantation and we constantly follow-up his clinical progress.



(A)



(B)

Figure 1. (A) The ECG of ventricular tachycardia with LBBB morphology originating from RV apex; (B) during sinus rhythm with left axis deviation and T waves inversion in precordial leads (V1-V6); red arrow

contrast. The results (Fig.3) included dilatation of RV without dilatation of RVOT (± 2.8 cm), aneurysmal outpouching with accordion sign at RV free wall during systolic and diastolic phase, and thinning of diaphragm wall of RV. There was fatty infiltration – with extensive late enhancement of Gadolinium at RV free wall and one segment of left ventricle (LV). Dyskinesia of RV free wall and diaphragm wall also showed. RV ejection fraction was 20.9%.

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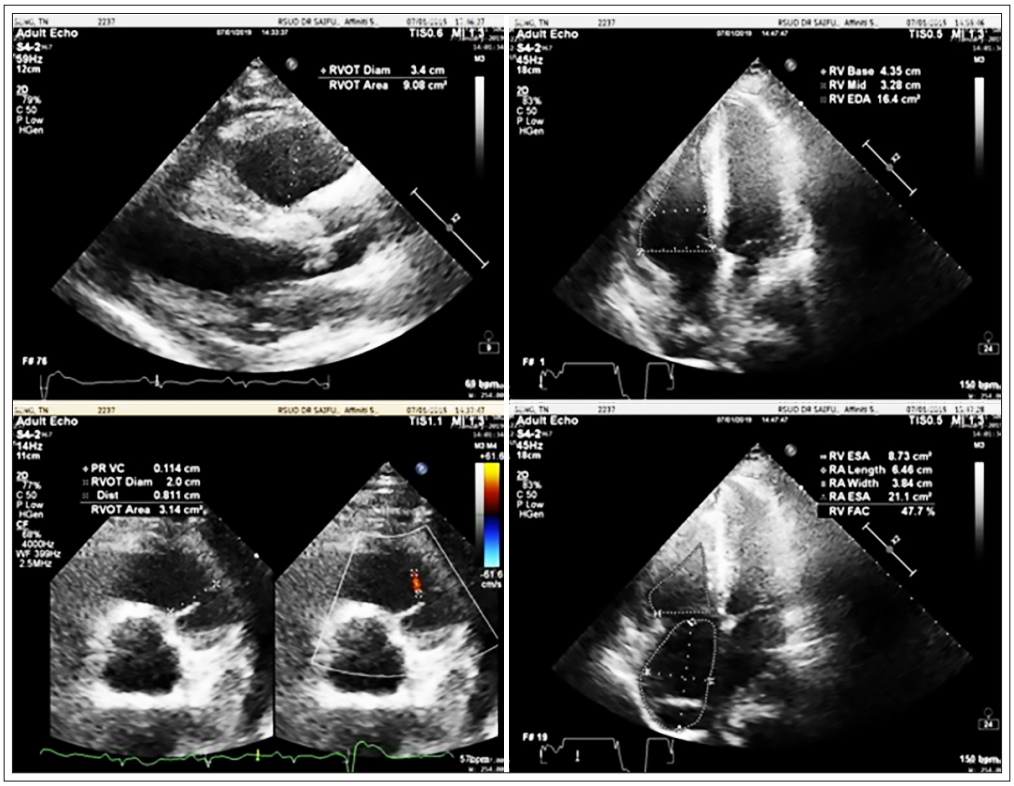

Figure 2. Echocardiography supported the minor criteria of ARVC diagnosis. Note the dilatation of RV and RV FAC 47.7%

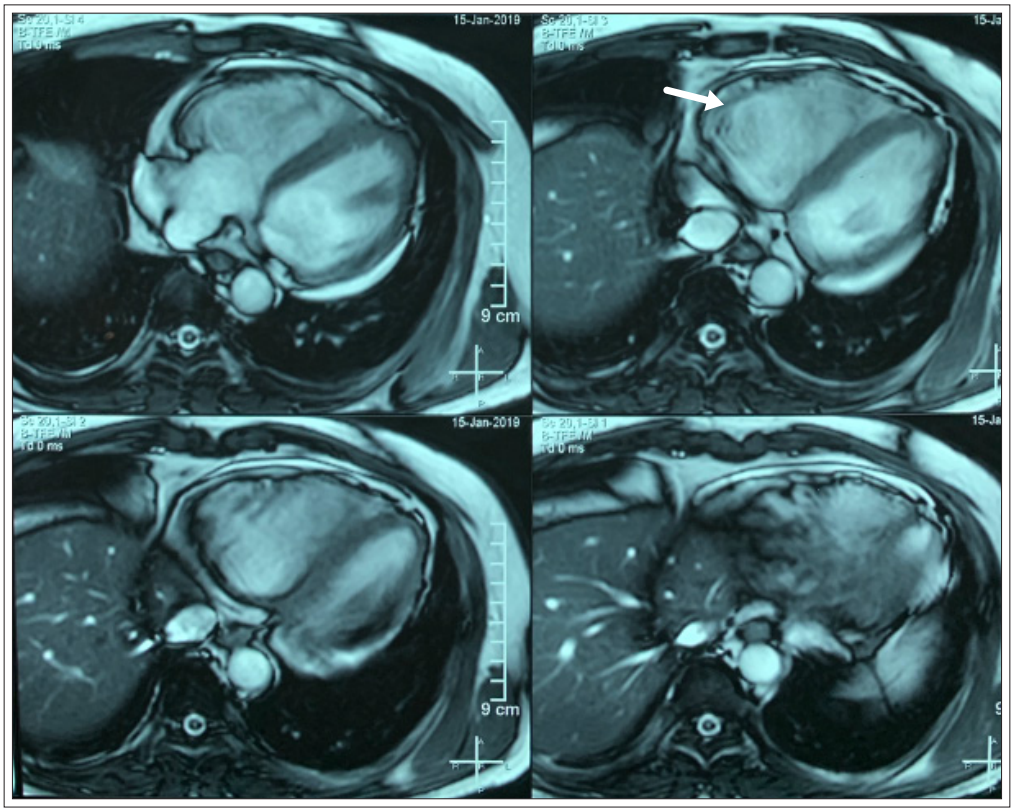

Figure 3. CMR findings in ARVC. Note the dilatation of right ventricle without dilatation of RVOT (white arrow)

Table 1. 2010 Task Force Criteria of ARVC diagnostic criteria[4]

	Major	Minor
I. Global or regional dysfunction and structural alterations	By 2D echocardiogram: Regional RV akinesia, dyskinesia, or aneurysm and 1 of the following (end-diastole): PLAX RVOT ≥32 mm (PLAX/BSA ≥19 mm/m^2) PSAX RVOT ≥36 mm (PSAX/BSA ≥21 mm/m^2) Or RFAC ≤33% By MRI: Regional RV akinesia or dyskinesia or dyssynchronous RV contraction and 1 of the following: RV end-diastolic volume/BSA ≥110 ml/m^2 (male) or ≥100 ml/m^2 (female) Or RV ejection fraction ≤40% By RV angiography: Regional RV akinesia, dyskinesia, or aneurysm	By 2D echocardiogram: Regional RV akinesia or dyskinesia, and 1 of the following (end-diastole): 29 ≤PLAX RVOT <32 mm (16 ≤PLAX/BSA <19 mm/m^2) 32 ≤PSAX RVOT <36 mm (18 ≤PSAX/BSA <21 mm/m^2) Or 33% < RFAC ≤40% By MRI: Regional RV akinesia or dyskinesia or dyssynchronous RV contraction and 1 of the following (end-diastole): 100 ml/m^2 ≤ RV end-diastolic volume/BSA <110 ml/m^2 (male) or 90 ml/m^2 ≤ RV end-diastolic volume/BSA <100 ml/m^2 (female) Or 40% < RV ejection fraction ≤45%
II. Tissue characterization of wall	Residual myocytes <60% by morphometric analysis (or <50% if estimated), with fibrous replacement of the RV free wall myocardium in ≥1 sample, with or without fatty replacement of tissue on endomyocardial biopsy	Residual myocytes 60% to 75% by morphometric analysis (or 50% to 65% if estimated), with fibrous replacement of the RV free wall myocardium in ≥1 sample, with or without fatty replacement of tissue on endomyocardial biopsy
III. Repolarization abnormalities	Inverted T waves in right precordial leads (V_1, V_2, and V_3) or beyond in individuals >14 yrs of age (in the absence of complete RBBB QRS ≥120 ms)	Inverted T waves in leads V_1 and V_2 in individuals >14 yrs of age (in the absence of complete RBBB) or in V_4, V_5, or V_6 Inverted T waves in leads V_1, V_2, V_3, and V_4 in individuals >14 yrs of age in the presence of complete RBBB
IV. Depolarization/conduction abnormalities	Epsilon wave in the right precordial leads (V_1 to V_3)	Late potentials by SAECG in ≥1 of 3 parameters in the absence of a QRS duration ≥110 ms on the standard ECG Filtered QRS duration ≥114 ms Duration of terminal QRS <40 μV (low-amplitude signal duration) ≥38 ms Root mean square voltage of terminal 40 ms ≤20 μV 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
V. Ventricular Arrhythmias	Nonsustained or sustained VT of LBBB morphology with superior axis (negative or indeterminate QRS in leads II, III, and aVF and positive in lead aVL)	Nonsustained or sustained RVOT VT of LBBB morphology with inferior axis (positive QRS in leads II, III, and aVF and negative in lead aVL) or with unknown axis >500 ventricular extrasystoles per 24 h (Holter)
VI. Family history	ARVC/D confirmed in a first-degree relative who meets current TFC ARVC/D confirmed pathologically at autopsy or surgery in a first-degree relative Identification of a pathogenic mutation categorized as associated or probably associated with ARVC/D in the patient	History of ARVC/D in a first-degree relative in whom it is not possible or practical to determine whether the family member meets current TFC Premature sudden death (<35 yrs of age) due to suspected ARVC/D in a first-degree relative ARVC/D confirmed pathologically or by current TFC in second-degree relative

Adapted with permission from Marcus et al. (4).

2D = two-dimensional; ARVC/D = arrhythmogenic right-ventricular cardiomyopathy/dysplasia; ECG = electrocardiography; BSA = body surface area; LBBB = left bundle branch block; MRI = magnetic resonance imaging; PLAX = parasternal long-axis view; PSAX = parasternal short-axis view; RBBB = right bundle branch block; RFAC = right fractional area change; RV = right ventricular; RVOT = right ventricular outflow tract; SAECG = signal-averaged electrocardiography; TFC = task force criteria; VT = ventricular tachycardia.

3. Discussion

We learned from this case that ARVC is an uncommon inherited cardiac disease characterized by progressive RV dysfunction due to fibro-fatty replacement of myocardium and associated with high-risk of ventricular arrhythmias and sudden cardiac death (SCD).[1] Our ARVC case occurred in males with young age, which corresponds to Kayser and Van der Wall[2] who said that predominantly occurs at a younger age, although any ages can be affected. The male-to-female ratio is 3:1.

The main pathological feature or ARVC/D is the replacement of myocytes by fibrous tissue in the RV free wall. In the early stage of the disease, structural changes may be absent or subtle and confined to a localized region of the RV, typically the inflow tract, outflow tract, or the apex of the RV – called the triangle of dysplasia.[3] Progression to diffuse RV disease and LV involvement, typically affecting the posterior lateral wall, is common.

We referred to the 2010 Task Force criteria[4] (Table 1) in order to establishing the diagnosis. Sustained VT of LBBB morphology with superior axis and inverted T waves in precordial leads from this patient's ECG were consistent to major criteria. Electrophysiological study (EPS) will be necessary to differentiate ARVC from idiopathic right ventricular outflow tract tachycardia.

Although these two points fulfilled a definite diagnosis of ARVC, we still think it is important to perform CMR for this patient.[5,6] Literature mentioned that cardiac imaging should be part of a comprehensive evaluation of ARVC diagnosis.[7]

A CMR assessment of the RV is often challenging, partly because it requires a significant learning curve due to low prevalence of the disease. Compared to echocardiography, CMR provides more accurate and reproducible measurements of chamber dimensions, volumes and function. In addition, it can provide non-invasive tissue characterization with the use of late gadolinium enhancement (LGE).[8] We personally approach the radiology team and prescribed the requirement about cardiac MRI protocol, as it is critical to know what signs to look for to diagnose ARVC. CMR findings of this patient included RV dilatation, fatty infiltration at RV free wall and one segment of LV, dyskinesia of RV free wall and diaphragm wall, and reduced RV EF (20.9%). There was also extensive LGE at free wall of RV which correlated to major criteria of ARVC.

Corrado et al[9] stated that this patient was stratified as

Corrado et al[9] stated that this patient was stratified as high-risk category, i.e. experienced cardiac arrest or sustained VT; therefore, he will benefit for ICD therapy despite his anti-arrhythmic medication. This patient had been advised for undergoing ICD implantation and at our closest monitoring, as he and his family still discuss about it.

ICD implantation is known for primary prevention of SCD in ARVC patients. However, the device is not free of short- and long-term complications. Some guidelines discourage the placement of defibrillators in patients with incessant VT, VF, or with significant psychiatric illness.[10]

Radiofrequency ablation has been proposed for patients with recurrent ventricular arrhythmias despite treatment with antiarrhythmic drugs. It is considered a complementary therapy for ICD, useful for management of symptoms but may not be sufficient to prevent SCD.[10] A case report by Santangeli et al[11] mentioned that endocardial and epicardial substrate ablation of VT in ARVC has a good long-term outcome. Most patients have complete VT control without amiodarone and limited antiarrhythmic drugs.

This case emphasized on cardiac magnetic resonance that is uniquely suited for ARVC/D evaluation as it is the most highly reproducible and specific technique for assessing morphology and segmental wall motion of RV. Hence, in our case, helping to identify the which patients will be benefit for ICD therapy in comparison to pharmacological and non-pharmacological approaches.

4. Declarations

4.1. Ethics Approval and Consent to participate
Patient has provided informed consent prior to involve in the study.

4.2. Consent for publication
Not applicable.

4.3. Availability of data and materials
Data used in our study were presented in the main text.

4.4. Competing interests
Not applicable.

4.5. Funding source
Not applicable.

4.6. Authors contributions
Idea/concept: OH. Design: OH. Control/supervision: AR, AFR. Data collection/processing: OH. Extraction/Analysis/interpretation: OH. Literature review: AR, AFR. Writing the article: OH. Critical review: AR, AFR. All authors have critically reviewed and approved the final draft and are responsible for the content and similarity index of the manuscript.

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