American Journal of Case Reports

ISSN 1941-5923
© Am J Case Rep, 2017; 18: 299-303
DOI: 10.12659/AJCR.901267

Received: 2016.08.26
Accepted: 2016.11.30
Published: 2017.03.24

# New Task Force Criteria Provide Evolution in Diagnosis of Arrhythmogenic Cardiomyopathy in Patients without Typical Progression of the Disease

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

ABDEF 1 Mariana S. Parahuleva
BCD 2 Jens Figiel
BCD 1 Holger Ahrens
BCDE 1 Bernhard Schieffer
ABDF 1 Dimitar Divchev
ABCDEF 1 Ulrich Lüsebrink

1 Department of Internal Medicine, Division of Cardiology and Angiology, University Hospital of Giessen and Marburg, Marburg, Germany
2 Department of Diagnostic and Interventional Radiology, University Hospital of Giessen and Marburg, Marburg, Germany

**Corresponding Author:** Mariana S. Parahuleva, e-mail: mariana.parahuleva@innere.med.uni-giessen.de
**Conflict of interest:** None declared

**Patient:** Male, 41
**Final Diagnosis:** ARVC
**Symptoms:** Recurrent palpitations and presyncope
**Medication:** β blockers
**Clinical Procedure:** CMRI • EP study • ICD implantation
**Specialty:** Cardiology

**Objective:** Unusual clinical course

**Background:** The original Task Force Criteria from 1994 for the clinical diagnosis of ARVC were highly specific and based on structural, histological, EKG, and familial features of disease. However, recommendations for clinical diagnosis and management of ARVC are sparse and lacked sensitivity for early disease.

**Case Report:** Ventricular electrical instability and sudden cardiac death are the hallmarks of ARVC, and are often present before structural abnormalities. In this case report, we describe a patient who had detectable electrical abnormalities and structural changes that remained unchanged for over 10 years.

**Conclusions:** The disease progression in this case was defined as the development of a new 2010 TFC, which was absent at enrolment in 1994 and in 2008.

**MeSH Keywords:** Arrhythmogenic Right Ventricular Dysplasia • Disease Progression • Electrocardiography • Magnetic Resonance Imaging

**Full-text PDF:** http://www.amjcaserep.com/abstract/index/idArt/901267

1404 — 2 2 7

## Background

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a slowly progressive disorder characterized by replacement of myocardial cells by fibro-fatty tissue, giving rise to ventricular tachyarrhythmias, in which electrical abnormalities precede detectable structural changes. Diagnosis is based on the presence of major and minor criteria from the Task Force Criteria (TFC) from 1994, which were revised in 2010 [1,2]. The new TFC include evaluation of findings from 6 different diagnostic categories: structural, histological, electrocardiographic, depolarization abnormalities, arrhythmic, and genetic features.

We describe a case with arrhythmogenic cardiomyopathy, in which structural and electrocardiographic abnormalities were the same for over 10 years. Based on revised TFC from 2010, and not on those from 1994, arrhythmogenic cardiomyopathy diagnosis in this case was possible.

## Case Report

A 41-year-old male presented in 2008 with recurrent palpitations and presyncope. Medical and family history was unremarkable. Initial 12-lead ECG showed sinus bradycardia (45 bpm), and terminal activation delay ≥55 ms in leads V1 through V3 in the presence of an incomplete right bundle-branch block (Figure 1A). On cardiac magnetic resonance imaging (CMRI) and echocardiography, the right ventricle (RV) was dilated (RVOT 35 mm) with an ejection fraction of 29% and dyskinesia of the right ventricular anterior wall (Video 1). The 24-h Holter monitoring revealed frequent sinoatrial arrests (up to 4.5 s) and a second-degree atrioventricular conduction block type I and II (<30 bpm) in the morning. He did not have any complaints nor did he ever have a syncope. Special electrophysiological (EP) investigation excluded an AV (node) reentrant tachycardia and pre-excitation syndrome as causes of palpitations, but demonstrated non-sustained ventricular tachycardia (NS-VT), which was inducible with programmed ventricular stimulation. Our patient had dilated RV and NS-VT and met 1 major and 1 minor criterion according to the TFC from 1994 for the clinical diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC) [1]. The patient was started on β blockers (Bisoprolol 5 mg daily).

He remains well with no further VT recurrence, but in 2015 he experienced a further episode of VT. Admission ECG showed VT with left bundle-branch block morphology, inferior axis, and late transition. On review of previous 12-lead ECGs, there still was sinus bradycardia (46 bpm) and terminal activation delay ≥55 ms in leads V1 through V3 in the presence of an incomplete right bundle-branch block (Figure 1B). Blood tests, including troponin, were normal and coronary angiography demonstrated no coronary artery disease. Additional cardiac evaluation included a cardiac imaging study showing a dilated RV with fibro-fatty infiltration, but no further structural changes compared to the CMRI performed in 2008. The CMRI showed a dilated RV (RVEDV/BSA: 117 mL/m$^2$) with an ejection fraction of 33.7% and dyskinesia of the right ventricular anterior wall (Video 2). An echocardiogram also found a dilated RV with fractional area change [FAC] 30.5% and PLAX RVOT 42.7 mm. Hence, the diagnosis of ARVC could now be made, as 1 major and 2 minor criteria were fulfilled [2]. Invasive electrophysiological assessment demonstrated clinical tachycardia (CL 280 ms) with left bundle morphology and superior axis, which was easily induced with 2 sensed extra stimuli, and successful direct ablation was made. Hence, the diagnosis of ARVC could now be made, as 2 major and 1 minor criteria were fulfilled [2]. Mapping using the electroanatomic mapping system (CARTO, Diamond Bar, CA) demonstrated a centrifugal pattern of wavefront activation from a central point on the anterior right ventricular free wall, where local electrograms were 60 ms before QRS onset, and irrigated ablation (35 W; 43°C) successfully terminated VT (Figure 2). The other 2 non-sustained arrhythmias were inducible with programmed ventricular stimulation, both with left bundle morphology and superior axis, and varying cycle length (380 ms and at 400 ms). Genetic analysis (plakophilin-2, PKP2) did not show any mutations. The patient was diagnosed with ARVC and continued to take β blockers. A dual-chamber implantable defibrillator (ICD) was implanted because of recently documented sustained ventricular tachycardia with varying cycle lengths (270 ms and 300 ms) in a LifeVest wearable cardioverter-defibrillator and sinus bradycardia [3]. During a 9-month follow-up, the patient remains on β blockers and had no documented ventricular arrhythmias.

## Discussion

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a genetic disease associated with progressive replacement of myocardial cells by fibro-fatty tissue and heart failure, initially only in the right ventricle [4,5]. The criterion standard for diagnosing ARVC is an endomyocardial biopsy demonstrating fibro-fatty replacement [1,2]. ARVC is associated with malignant ventricular arrhythmias, which originate from the right ventricle, and high risk of sudden death [1,2]. Electrical instability precipitating sudden cardiac death often presents before structural abnormalities. Therefore, early accurate diagnosis is of utmost importance. Our patient, however, presented with a sinus bradycardia (45 bpm), terminal activation delay ≥55 ms in leads V1 through V3 in the presence of an incomplete right bundle-branch block (Figure 1A), sinoatrial arrests (up to 4.5 s), and a second-degree atrioventricular conduction block type I and II (<30 bpm) in the morning in Holter monitoring, but no

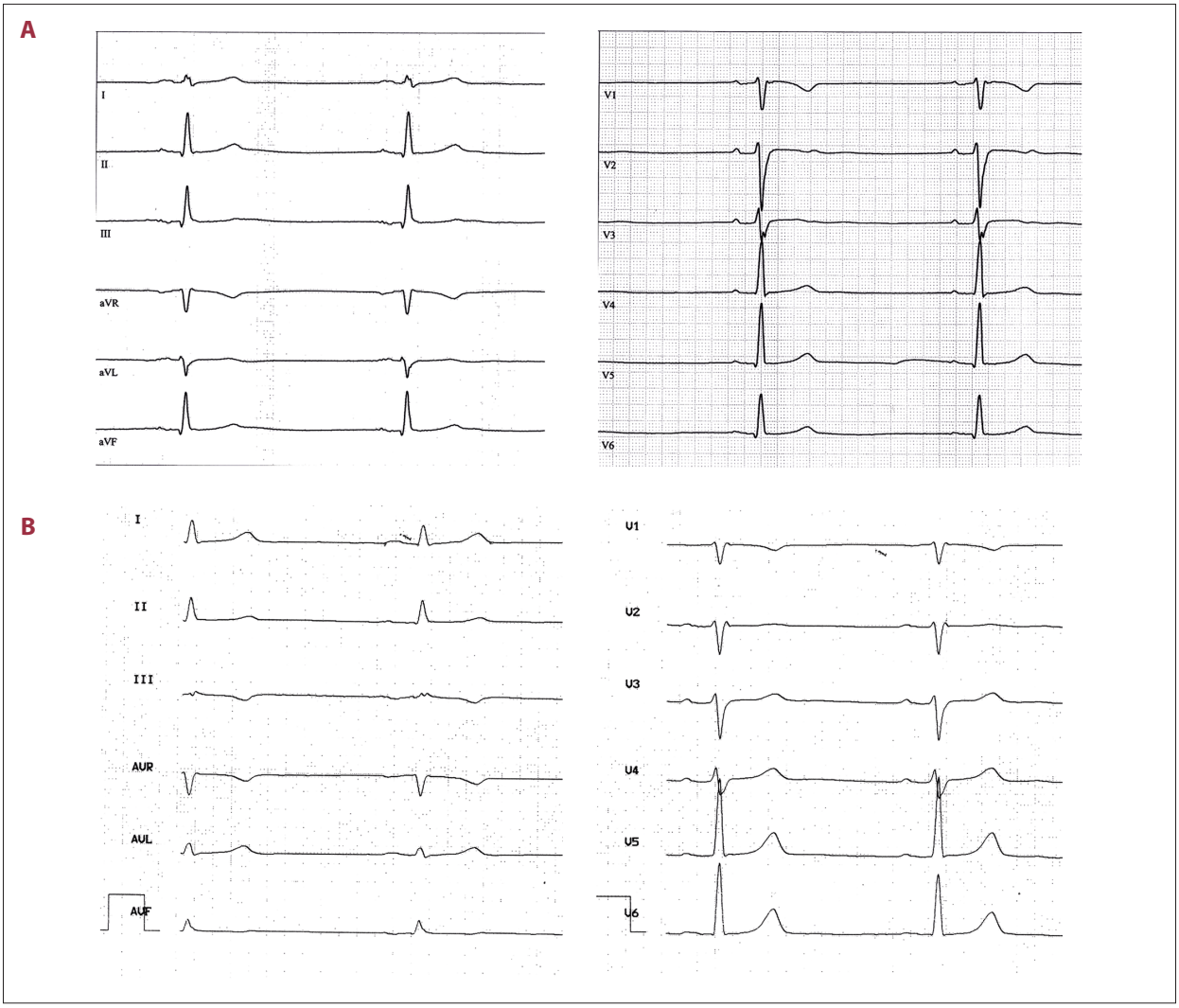
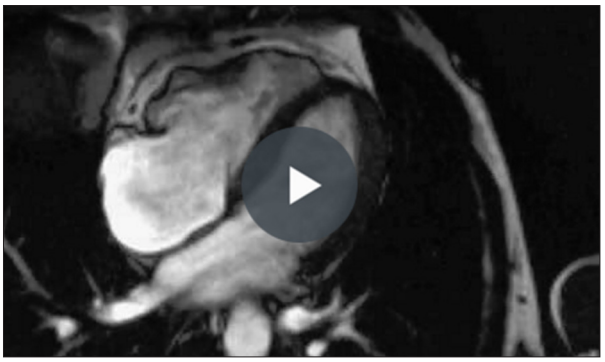
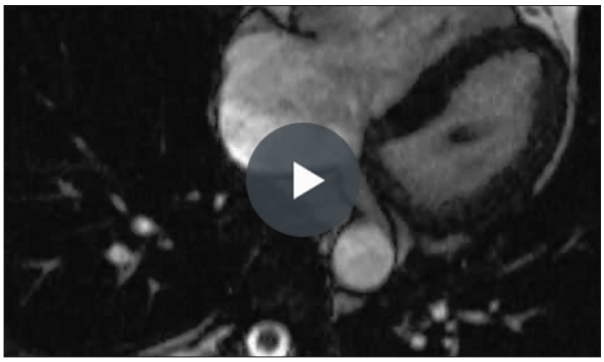



**Figure 1.** Late potentials (ECG). (**A**) Admission ECG (2008) showing terminal activation duration of QRS >55 ms in leads V1 through V3 in the presence of an incomplete right bundle-branch block. (**B**) Admission ECG, 7 years later (2015), showing the same late potentials.



**Video 1.** (2008): Cardiac magnetic resonance long axis view of the dilated right ventricle (RV) shows the transmural diffuse bright signal and dyskinesia of the RV free wall due to massive myocardial atrophy with fatty replacement.



**Video 2.** (2015): Cardiac magnetic resonance long axis view of the dilated right ventricle (RV) shows the transmural diffuse bright signal and dyskinesia of the RV free wall due to massive myocardial atrophy with fatty replacement.

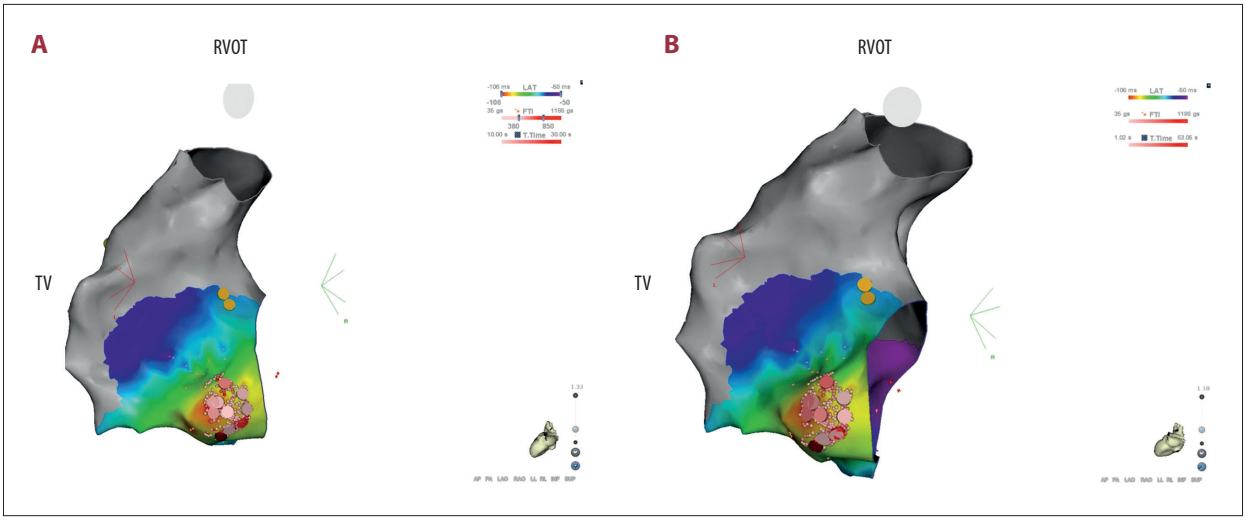



**Figure 2.** (**A, B**) Anteroposterior image of the right ventricle (RV) created using the electroanatomic mapping system (CARTO, Diamond Bar, CA). Red indicates early and blue indicates late activation (see color bar). The positions of the tricuspid (TV) and right ventricular outflow tract (RVOT) have been annotated. Activation can be seen originating in the inferoseptal/basal section of the RV free wall and spreading in a centrifugal fashion away from this central point. Radiofrequency ablation (red dots) at the site of earliest activation was successful in terminating ventricular tachycardia.

ventricular tachycardias. Non-sustained ventricular tachycardia (NS-VT) was inducible with programmed ventricular stimulation. On CMRI and echocardiography, the right ventricle (RV) was dilated (RVOT 35 mm) with an ejection fraction of 29% and dyskinesia of the right ventricular anterior wall (Video 1), but endomyocardial biopsy was negative for ARVC. Our patient met 1 major and 1 minor criterion according to the Task Force Criteria (TFC) from 1994 for the clinical diagnosis of ARVC, and was classified as borderline ARVC [1]. There was no suspicion of ARVC until he became symptomatic and had ventricular tachycardia (VT) with left bundle-branch block morphology, inferior axis, and late transition in 2015 with the same clinical and cardiac parameters. The other 2 non-sustained arrhythmias were inducible with programmed ventricular stimulation, both with left bundle morphology and superior axis as a major criterion for the diagnosis of ARVC. (Non-) sustained VT with left bundle morphology and superior axis as a major criterion were diagnostically superior in this case for the early diagnosis of ARVC, probably minimizing ventricular arrhythmias and delaying the progression of disease. The defining of EKG specificity, as well as electrophysiological studies by (non-) sustained VT in such patients, could help in early diagnosis of ARVC [6]. Electrophysiological studies can be indicated and they could be useful in differential diagnosis and suitable for automatic analysis and early diagnosis in patients with monomorphic VT with left bundle-branch block, who are susceptible to ARVC. However, induction of VT during electrophysiological study is not predictive for the future occurrence of VT in patients with ARVC [7]. Furthermore, the revised 2010 TFC provide additional cardiac evaluation for this patient, including a signal-averaged ECG and cardiac imaging, which are important for the early diagnostic and prognostic management of ARVC [2,3]. The signal averaged ECG was positive, as 1 (minor criterion) of the 3 parameters was abnormal, including a terminal activation delay ≥55 ms in leads V1 through V3 in the presence of an incomplete right bundle-branch block (Figure 1). Imaging is important for the clinical diagnosis of ARVC, included both echocardiography and CMRI [2,3]. Technical evolution in CMRI and echocardiography improved the sensitivity and specificity of the right and left ventricles images that provide both diagnostic and prognostic information. The CMRI showed a dilated RV (RVEDV/BSA: 117 mL/m$^2$) with an ejection fraction of 33.7% and dyskinesia of the right ventricular anterior wall (Video 2). Hence, the diagnosis of ARVC could now be made, as 2 major and 1 minor criteria were fulfilled [2]. The revised criteria from 2010 provide specificity of the diagnostic of ARVC, especially in patients previously classified as having borderline ARVC according to the TFC from 1994. The sensitivity of the revised criteria is not perfect, but in our case, in which the detectable electrical abnormalities and structural changes were the same for over 10 years, the diagnosis of ARVC was defined as the development of a new 2010 TFC, which was absent at enrolment in 2008 [1,2].

## Conclusions

The results of this case show a better diagnostic correlation between disease criteria assessed by TFC from 2010 compare to those from 1994, which is important for the early diagnosis and prognostic management of ARVC. Furthermore, electrophysiological studies with electroanatomic mapping systems

can be a useful technique to evaluate and specify VT with left bundle morphology in patients with borderline ARVC. The modifications of the TFC from 2010 represent recommendations for the early diagnosis of ARVC, which could minimize ventricular arrhythmias and delay the progression of disease.

## References:

1. McKenna WJ, Thiene G, Nava A, et al. Diagnosis of arrhythmogenic right ventricular dysplasia/cardiomyopathy. Task Force of the Working Group Myocardial and Pericardial Disease of the European Society of Cardiology and of the Scientific Council on Cardiomyopathies of the International Society and Federation of Cardiology. Br Heart J. 1994;71: 215-8.
2. Marcus FI, McKenna WJ, Sherrill D, et al. Diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia: proposed modification of the Task Force Criteria. Eur Heart J. 2010;31: 806-814.
3. Priori SG, Blomström-Lundqvist C, Mazzanti A et al: Task Force for the Management of Patients with Ventricular Arrhythmias and the Prevention of Sudden Cardiac Death of the European Society of Cardiology (ESC). 2015 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death: The Task Force for the Management of Patients with Ventricular Arrhythmias and the Prevention of Sudden Cardiac Death of the European Society of Cardiology (ESC) Endorsed by: Association for European Paediatric and Congenital Cardiology (AEPC). Europace, 2015; 17: 1601–87
4. Quarta G, Muir A, Pantazis A et al: Familial evaluation in arrhythmogenic right ventricular cardiomyopathy: Impact of genetics and revised task force criteria. Circulation, 2011; 123: 2701–09
5. Saberniak J, Hasselberg NE, Borgquist R et al: Vigorous physical activity impairs myocardial function in patients with arrhythmogenic right ventricular cardiomyopathy and in mutation positive family members. Eur J Heart Fail, 2014; 16: 1337–44
6. Batchvarov VN, Bastiaenen R, Postema PG et al: Novel electrocardiographic criteria for the diagnosis of arrhythmogenic right ventricular cardiomyopathy. Europace, 2016; 18: 1420–26
7. Corrado D, Leoni L, Link MS et al: Implantable cardioverter-defibrillator therapy for prevention of sudden death in patients with arrhythmogenic right ventricular cardiomyopathy/dysplasia. Circulation, 2003; 108: 3084–91

**Conflict of Interest Disclosures**

None.