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Case Report

Arrhythmogenic cardiomyopathy: from concealed phase to refractory failure: a nine-year case study illustrating diagnostic evolution and biventricular progression

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ABSTRACT

Arrhythmogenic cardiomyopathy (ACM) is a type of cardiomyopathy characterized by fibrofatty replacement of the right ventricular myocardium and associated with life-threatening arrhythmias. The criteria of the diagnosis of ACM are based on the estimates of structural changes in the right ventricular myocardium, electrocardiographic changes, fibrofatty dystrophy in the course of a histological investigation, and a family history of the disease. The diagnosis can be a challenging task as there are no pathognomonic clinical markers, which makes differential diagnosis of ACM with secondary myocardial pathology more difficult to perform. The article presents a nine-year-old case study of a young male patient with ACM, dominated by life-threatening ventricular arrhythmias and episode of resuscitated sudden cardiac death despite optimal medical therapy and cardioverter-defibrillator implantation.

Keywords: Arrhythmogenic cardiomyopathy, Coronary angiography, Ventricular fibrillation, Implantable cardioverter-defibrillator

INTRODUCTION

Arrhythmogenic cardiomyopathy (ACM) is a heritable, progressive cardiac muscle disease characterized by fibro-fatty replacement of the ventricular myocardium, leading to potentially fatal ventricular arrhythmias.[1] It was formally defined in 1982, with the term ACM now encompassing left, right, and biventricular forms due to advances in imaging and genetics. ACM is inherited in an autosomal dominant pattern in 30-50%, involving mutations mainly in desmosomal protein genes such as desmocollins, desmogleins, desmoplakin, and junction plakoglobin, which are vital for cell adhesion under mechanical stress.[1,4] Defects in these proteins cause myocyte loss and replacement by fibrous and fatty tissue, disrupting intercellular connections and ion channels, notably connexin43 and Nav1.5, leading to electrical instability.[5,7] Acquired factors like viral myocarditis and intense endurance sports participation may also contribute, especially in gene-elusive individuals, with exercise potentially acting as a trigger.[2,3] The disruption of the Wnt/β-catenin pathway promotes fibrofatty infiltration, chamber dilatation, aneurysm formation, and increases the risk of ventricular tachycardia and sudden death.[6,8] Overall, structural and electrical remodeling underpins the pathogenesis of ACM.

Adolescents and young adults with ARVC often present with symptoms such as palpitations, premature ventricular contractions (PVCs), ventricular arrhythmias, suspected cardiac syncope, or a history of aborted cardiac arrest. A characteristic ventricular tachycardia (VT) in ARVC typically exhibits a left bundle branch block pattern with a superior axis, although other VT morphologies can also be associated. The disease progresses through three stages: initially, patients are usually asymptomatic during the

"concealed phase," but remain susceptible to ventricular arrhythmias and sudden cardiac death. In the second, "electrical phase," symptomatic arrhythmias may occur, with right ventricular (RV) morphological abnormalities sometimes visible on standard imaging modalities.

Ultimately, the condition may advance to a diffuse, progressive stage causing right, left, or biventricular heart failure, often with ongoing ventricular arrhythmias, reflecting the widespread myocardial degeneration characteristic of ARVC. Disease progression, including potentially fatal ventricular arrhythmias, may manifest as "hot phases" and sporadic bursts rather than as a continuous process.[4,9]

The diagnosis of ACM is based on a comprehensive scoring system outlined in the 2023 international (Padua) criteria, integrating structural, functional, electrocardiographic, and tissue findings. The 2023 European Task Force criteria delineate markers across six categories for right and left ventricular involvement. Morpho-functional abnormalities include regional wall motion issues and dilation or systolic dysfunction, with right ventricular (RV) major criteria and left ventricular (LV) minor criteria. Structural myocardial abnormalities focus on tissue characterization, such as fibrous replacement and specific late Gadolinium enhancement (LGE) patterns like the ring-like pattern in the LV.

ECG findings involve repolarization and conduction abnormalities, including T wave inversions, epsilon waves, and low QRS voltages. Arrhythmia criteria encompass frequent ventricular extrasystoles (>500/24 hours) and VT morphologies, with LBBB pattern indicating RV involvement and RBBB for LV. Family history and genetics consider pathogenic variants and premature sudden death in relatives as major criteria.[10,11]

Several conditions can imitate ACM, thereby complicating the diagnostic process. Conditions that mimic ACM, along with their key distinguishing features is given in Table 1.[4,12]

The primary aim in managing ACM is to reduce arrhythmic death, prevent disease progression, and enhance quality of life by controlling symptoms and heart failure. A multidisciplinary team conducts family screening and genetic counseling. Treatment includes lifestyle modifications, β-blockers as first-line therapy, antiarrhythmic drugs (e.g., amiodarone), catheter ablation, implantable cardioverter defibrillators (ICD), and heart transplantation for severe cases.[13,14]

Amiodarone effectively prevents ventricular arrhythmias but its impact on sudden cardiac death remains unproven.[15] Physical activity is a key trigger, and ICDs are recommended for high-risk patients, with transplantation reserved for refractory heart failure or uncontrollable arrhythmias.[1,14,15]

Table 1: Condition and criteria.

Condition	Key features	Distinguishing diagnostic criteria
Idiopathies RVOT tachycardia	Ventricular tachycardia originating from the right ventricular outflow tract, often benign	Typical ECG morphology, response to ablation, absence of structural heart disease
Brugada syndrome	ECG pattern with coved-type ST elevation in right precordial leads	Type 1 Brugada ECG pattern, genetic testing, provocation tests
Myocarditis	Inflammatory myocardial process, can cause arrhythmias	Cardiac MRI, biopsy showing inflammatory infiltrates, clinical history
Sarcoidosis	Granulomatous infiltration of myocardium, can cause arrhythmias	Biopsy showing non-caseating granulomas, hilar lymphadenopathy, PET scan uptake
Dilated cardiomyopathy (DCM)	Ventricular dilation, systolic dysfunction	Echocardiography, MRI, exclusion of other causes, histology
Uhl's anamoly	Congenital absence of RV myocardium	Echocardiography, cardiac MRI, clinical presentation
Pulmonary hypertension	Elevated pulmonary pressures causing right ventricular strain	Right heart catheterization, echocardiography

CASE REPORT

In 2016, the male patient of age twenty-four, initially presented to the Arrythmology department of the Grodno Regional Carpological Centre with palpitations detected during a routine medical evaluation.

The primary diagnostic workup revealed a significant electrical disturbance. Patient's ECG showed a regular sinus rhythm with a heart rate of 69 beats per minute. The electrical axis of the heart was normal. The T wave in the right precordial leads (V1-V3) was inversed. QRS complex in V2 was fragmented (Figure 1).

A 24-hour Holter monitor revealed a very high burden of ventricular ectopy with 4,221 PVCs, 6 runs of non-sustained VT were also recorded (Figure 2).

An exercise stress test proved particularly provocative, inducing short paroxysms of VT with a heart rate up to 170 beats per minute and an episode of paired bidirectional PVCs at peak exertion. However, concurrent structural

assessment by echocardiography showed only minor abnormalities: grade I mitral valve prolapse with trivial regurgitation and an anomaly of the left ventricular chords. The right ventricle appeared structurally normal. Given the disparity between severe electrical instability and the absence of overt cardiomyopathy, the initial diagnosis centered on ventricular arrhythmias in the setting of mitral valve prolapse, though arrhythmogenic right ventricular dysplasia was considered as a possible underlying cause. Management was initiated with antiarrhythmic medication (sotalol 160 mg 2 times per day).

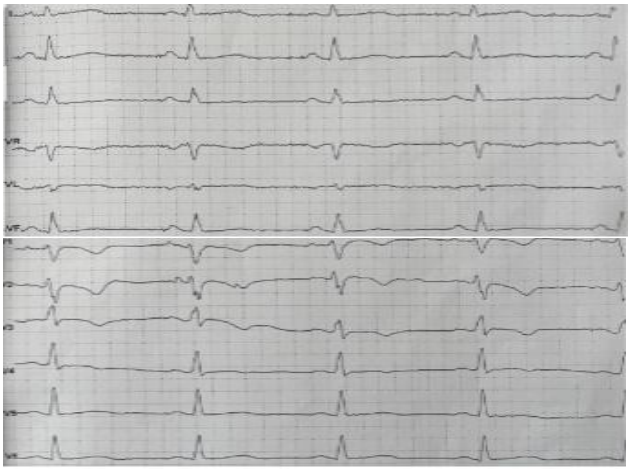

Figure 1: Patient's ECG upon admission.

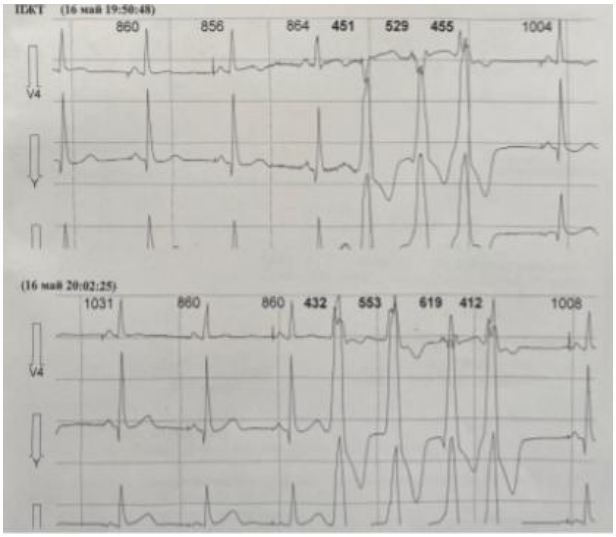

Figure 2: Fragment of patient's Holter monitor.

The clinical course over the next several years was characterized by persistent ventricular arrhythmias managed medically with sotalol, and further with beta-blocker (metoprolol).

A pivotal diagnostic shift occurred in 2023 following a cardiac MRI study, which raised strong suspicion for a right-dominant form of ACM.

A gadolinium-enhanced cardiac MRI revealed micro aneurysmal protrusions in the lateral and inferior walls of the RV, along with areas of dyskinesis of the RV inferior wall. RV EF was reduced to 42%.

Endomyocardial biopsy was not performed due to the presence of convincing evidence for ACM based on non-invasive cardiac examination methods. This suspicion was confirmed later that year when the patient suffered a sudden cardiac arrest during physical activity. Emergency medical services documented VF, requiring defibrillation. The post resuscitation period was complicated by a type 2 myocardial infarction (troponin I level of 50,000 ng/l) and severe hypoxic brain injury. Coronary angiography excluded obstructive disease, revealing only a myocardial bridge (Figure 3).

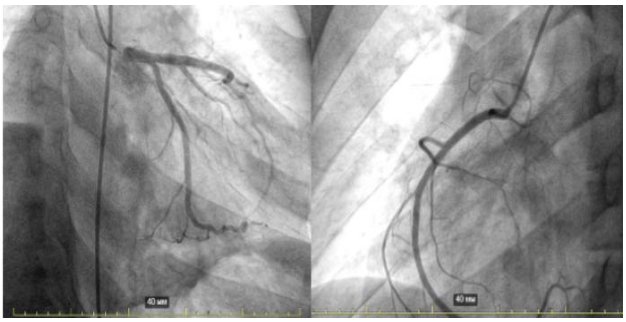

Figure 3: Patient's coronary angiography.

Post-arrest echocardiography showed new focal left ventricular hypokinesis with a mildly reduced ejection fraction of 53%. This sentinel event solidified the diagnosis of arrhythmogenic right ventricular dysplasia, and a secondary prevention ICD was subsequently implanted (Figure 4).

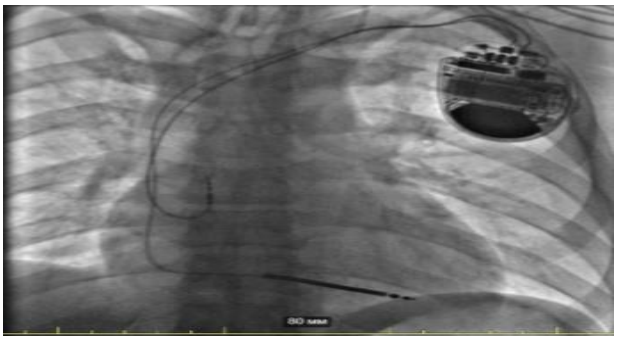

Figure 4: Chest X-ray revealing implanted ICD.

The patient's recovery was protracted, necessitating tracheostomy and extensive neurological rehabilitation.

Patient's diagnosis upon discharge was ACM. Myocardial bridge in the LAD according to coronary angiography. Grade 1 MVP with grade 1 mitral regurgitation. VF, clinical death, restoration of effective cardiac activity 10/08/2023. Frequent episodes of monomorphic VT. Frequent PVCs. ICD implantation 11/01/2023. Post

resuscitation disease. Post hypoxic encephalopathy. Amnestic syndrome. Partial kinetic apraxia, more pronounced in the hands. Mild volitional disorder.

Following ICD implantation, the patient's condition remained unstable. A hospitalization in early 2024 for recurrent palpitations demonstrated ongoing electrical instability despite therapy, with Holter monitoring confirming persistent polymorphic PVCs. The disease entered a more aggressive phase in 2025, manifesting as an electrical storm. The patient presented after receiving two appropriate ICD shocks. ICD interrogation revealed 74 episodes of ventricular tachycardia over a short period, including one sustained VT terminated by anti-tachycardia pacing and one VF episode terminated by a shock. Subsequent ambulatory monitoring, performed while on high-dose combination therapy with metoprolol and amiodarone, captured an extreme burden of ventricular ectopy: over 12,000 single PVCs, hundreds of paired complexes, and dozens of non-sustained VT runs within 24 hours (Figures 5 and 6).

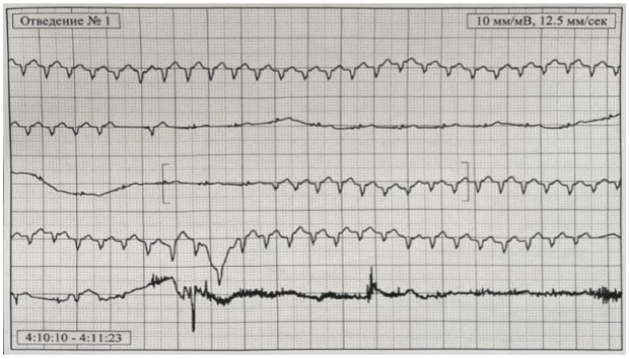

Figure 5: Episode of non-sustained VT (22 seconds) on patient's Holter monitor.

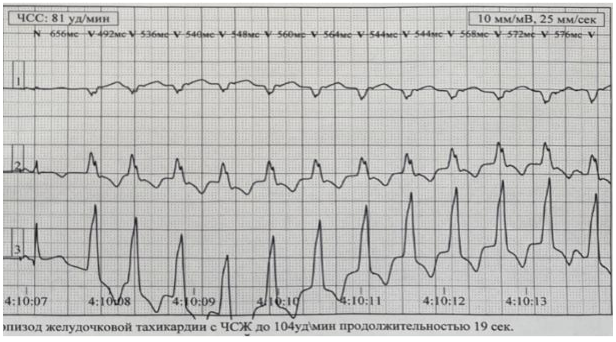

Figure 6: Episode of non-sustained VT (19 seconds) on patient's Holter monitor.

Management was intensified with maximized pharmacotherapy, including metoprolol 50 mg, 1 tablet 3 times a day (08.00-14.00-19.00), amiodarone 200 mg, 1 tablet at 14.00, and potassium and magnesium supplements, highlighting the refractory nature of the arrhythmias in advanced ACM.

During the ICD function check as of 07 May 2025 no abnormalities were detected. However, six episodes of non-sustained VT were recorded despite ongoing therapy.

Patient was consulted by the Head of the angiography room of the Arrhythmology department in the Republican Scientific and Practical Center "Cardiology" (Minsk) on 07/07/2025. The patient was indicated for radiofrequency ablation of the arrhythmia substrate. At the time of consultation, there were no technical conditions for performing radiofrequency ablation of the VT due to the lack of ablation electrodes. The ongoing treatment in accordance with the clinical protocols was continued, and patient was put to the ablation waiting list. The patient has now been followed for nine years (2016–2025), with progressive disease from concealed arrhythmias to electrical storm despite maximal therapy.

DISCUSSION

This nine-year course allows interpretation of the patient's findings in the context of established ACM literature.

The initial ECG showed T-wave inversions in V1–V3 and a fragmented QRS in V2. As Haugaa et al have emphasized, these repolarization abnormalities are among the earliest electrocardiographic markers of ACM, often preceding structural changes by years.[4] The 24-hour Holter revealing 4,221 PVCs and six runs of non-sustained VT with left bundle branch block morphology points to an RV origin of arrhythmias. According to the 2023 Padua criteria, more than 500 PVCs per 24 hours with LBBB morphology constitutes an arrhythmia criterion for ACM.[10,11] Unlike idiopathic RVOT tachycardia, which follows a benign course without structural progression, this patient's escalating PVC burden and eventual electrical storm strongly support ACM as the correct diagnosis.[4] The cardiac MRI in 2023 revealed microaneurysmal protrusions, dyskinesis of the RV inferior wall, and an RVEF of 42%. Graziano et al describe these morpho-functional abnormalities as major diagnostic criteria for ACM.[10] The decision to forego endomyocardial biopsy is consistent with Pilichou et al, who noted that a combination of ECG, Holter, and CMR can achieve diagnostic certainty without invasive biopsy when typical findings are present.[1]

The patient's sudden cardiac arrest with VF during physical activity reflects the natural history described by Romero et al, who reported that aborted sudden death is a frequent first manifestation of ACM, particularly in young, physically active individuals.[2] Zorzi et al further emphasized that intense physical activity acts as a key trigger for arrhythmias.[3] The post-arrest myocardial bridge was incidental; the absence of obstructive coronary disease confirms that the dominant mechanism was re-entrant VT within RV scar.[15]

Despite secondary prevention ICD implantation and maximized pharmacotherapy (metoprolol 150 mg/day plus

amiodarone 200 mg/day), the patient developed an electrical storm with 74 VT episodes. This refractoriness is characteristic of advanced ACM, where progressive fibrofatty replacement creates a substrate resistant to antiarrhythmic drugs. Quarta et al described that many patients with advanced ACM require escalation to combination therapy or catheter ablation.[9] The progression to biventricular involvement (LVEF 53%) is a poor prognostic sign; Corrado et al documented that left ventricular involvement is associated with worse arrhythmia-free survival.[11] The indication for catheter ablation is clear. For patients with electrical storm despite optimal medical therapy, ablation offers the best chance of arrhythmia control. The international task force consensus statement strongly recommends ablation in such patients, and standard protocols prioritize it when pharmacotherapy fails.[13-15] The absence of genetic testing is a limitation; however, the clinical and imaging findings alone were sufficient for diagnosis under the 2023 Padua criteria.[10,11]

CONCLUSION

This case illustrates the progressive natural history of ACM and highlights the challenges of managing refractory ventricular arrhythmias despite ICD therapy and maximal pharmacotherapy. It underscores the value of early cardiac MRI and the need for timely catheter ablation in advanced disease.

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