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Case Report

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Asymptomatic Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) in a Young Adult: A Case Report and Comprehensive Review

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ABSTRACT
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) is a rare genetic heart disorder characterized by fibrofatty replacement of the myocardium, which can present as silent disease to ventricular arrhythmias, heart failure and sudden cardiac death. Here, we report a case of ARVC in a 38 years old male, who initially presented to us with complaints of dizziness. Confirmed diagnosis was made with the help of Cardiac MRI, though a provisional diagnosis was made via ECG and ECHO.

Keywords: ARVC, ECG, ECHO, CMR

Introduction

ARVC is a rare and under-recognized form of hereditary cardiomyopathy primarily affecting the right ventricle, characterized by structural and functional changes in the myocardium. Pathologically, there is an fibrofatty infiltration of the myocardium and clinically features of RV electrical instability can be observed [1]. Despite its name, there have been cases of ARVC where we can observe involvement of the left ventricle and causing left sided heart failure. The disease often remains asymptomatic, posing challenges in early detection, particularly in young individuals with no apparent cardiac symptoms with the prevalence of 1:2000 to 1:5000 [2].

Case Report

A 38 years old asymptomatic gentleman was referred to our outpatient department for necessary cardiac evaluation as the patient initial general health check-up revealed cardiomegaly. His family history was non-significant for any sudden cardiac events. Physical examination revealed no abnormal findings, his routine blood investigations were normal and his resting electrocardiogram (ECG) was within normal limits. His ECG (figure 1) shows normal sinus rhythm with right axis deviation, P pulmonale, and symmetrically inverted T waves (in precordial leads) (minor criteria).

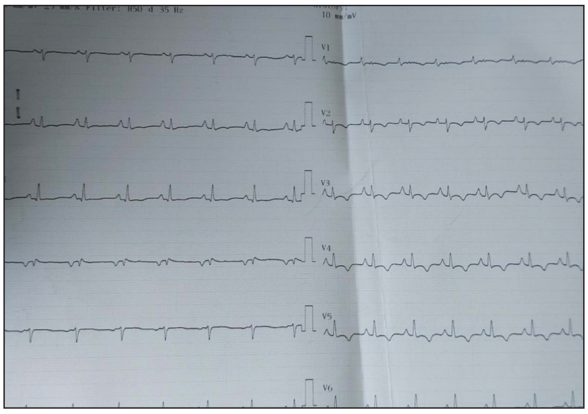

Figure 1: ECG showing Normal Sinus rhythm with P-Pulmonale and inverted T waves in precordial leads

Following this, we performed trans-thoracic echocardiography (figure 2a, 2b) and it revealed Non coapting free flow Severe TR, dilated RA, RV, dyskinetic RV apex, dilated RV outflow tract (in parasternal long axis view: 40mm and parasternal short axis view:44mm) (major criteria). With these findings, we suspected this case to be ARVC and the case was discussed with the radiology team and Cardiac MRI was planned. His CMR (figure 3) revealed dilated RA and RV with dyssynchronous RV wall motion and diffuse enhancement along the RV wall and RV outflow tract. It showed drastic reduction in right ventricular ejection fraction (RVEF:36%) (major criteria)

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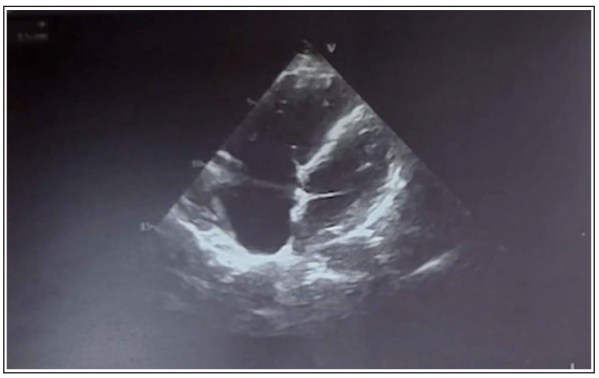

Figure 2(a): Echocardiogram showing dilated RA and RV

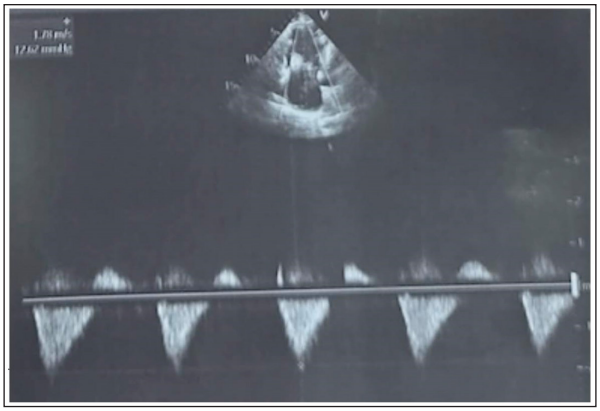

Figure 2(b): Echocardiogram showing Severe TR

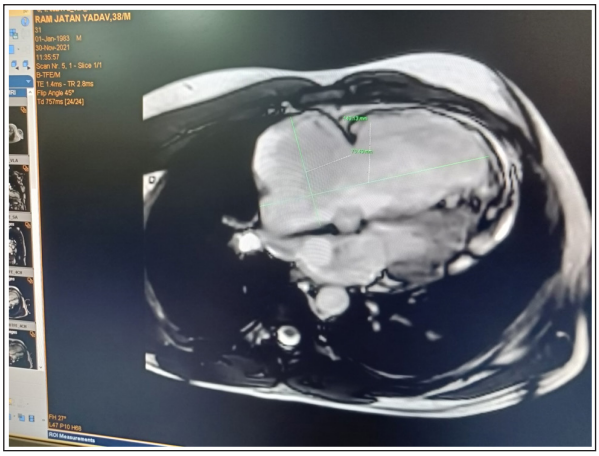

Figure 3: MRI showing dilated cardiac chambers and on calculation of RVEF<30%

In this compelling case, our asymptomatic young adult patient underwent a comprehensive evaluation based on the 2010 revised criteria for the diagnosis of Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC). The cardiac magnetic resonance imaging (CMR) and TTE findings fulfilled the major criteria, providing a significant diagnostic clue. Additionally, the presence of a repolarization abnormality on the electrocardiogram (ECG) fulfilled one of the minor criteria, further supporting the definitive diagnosis of ARVC. We discussed the unpredictable nature of ARVC and the potential risks it posed, particularly concerning the occurrence of life-threatening arrhythmias. Considering these circumstances, we recommended the placement of an Implantable Cardioverter-Defibrillator (ICD) as a proactive measure, should symptoms arise in the future.

Discussion

In 1982, Dr. Frank Marcus first reported Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), an inherited heart condition with a progressive nature and increased risk of ventricular arrhythmias and sudden cardiac death (SCD). ARVC is characterized by fibro-fatty replacement of the right ventricular (RV) myocardium, leading to RV dilation and systolic dysfunction. This genetic disorder significantly contributes to sudden cardiac death, particularly among young individuals and athletes, with most cases diagnosed between ages 10 and 50 [3]. Research suggests a connection between aberrant WNT signaling of desmosomal proteins and direct plakoglobin signaling, which transforms myocytes into adipocytes as the disease progresses, leading to fibro-fatty replacement of myocardium in ARVC [4]. Additionally, multiple genetic mutations in one individual may complicate the disease course [5].

The symptom complex in patients with ARVC can manifest in a variety of ways, as observed in one study. Among them, 67% reported experiencing palpitations, while 32% presented with syncope, indicating a temporary loss of consciousness. Additionally, 27% reported atypical chest pain, 11% mentioned experiencing dyspnea, a feeling of breathlessness or shortness of breath, and 6% had symptoms of right ventricular (RV) failure. These findings illustrate the diverse symptomatology of ARVC [6].

Diagnosis

Initial diagnosis can be made with the ECG findings of typical monomorphic ventricular tachycardia with left bundle branch block in a superior axis. Also, the Epsilon wave in the right precordial lead is highly specific for ARVC [7]. However, a final diagnosis of ARVC can only be reached when it fulfills the 2010 revised task force criteria [8].

I). Global or regional dysfunction and structural alterations in CMRI or ECHO
II). Tissue characterization of wall (fibrous replacement and percentage of residual myocytes in right ventricle)
III). Repolarization abnormalities on ECG (T wave inversion in V1, V2 and V3)
IV). Depolarization/ conduction abnormalities on ECG (epsilon wave in V1, V2 and V3)
V). Arrhythmias (VT with LBBB morphology and superior axis)
VI). Family history (ARVC in a first degree relative confirmed with Task Force criteria or at autopsy

Each of these criteria are further subdivided into major and minor criteria. We can reach a final definitive diagnosis when patient satisfies two major criteria or one major and two minor criteria or four minor criteria from different categories. Similarly, a borderline diagnosis can be made with one major and one minor or three minor criteria from different categories. Also, a possible diagnosis can be made with one major or two minor criteria from different categories.

In our case, we made the final diagnosis as our patient met one major and 2 minor criteria.

Management

The primary goal in treating ARVC is to prevent fatal ventricular arrhythmias and sudden cardiac death (SCD). It is crucial for patients diagnosed with definite ARVC to refrain from engaging in athletic activities, as this can exacerbate the condition and increase the risk of adverse outcomes [9,10].

Betablockers play a pivotal role in the management of ARVC by slowing the progression of ventricular dysfunction and effectively suppressing ventricular arrhythmias. Their use has been shown to be beneficial in enhancing patient outcomes and ensuring better control of the condition [10].

For patients at high risk of developing ventricular arrhythmias, and as a means of primary prevention, ICD therapy is considered essential. By providing prompt treatment during life-threatening arrhythmic events, ICDs can effectively reduce the risk of SCD. Moreover, patients with a history of sustained ventricular tachycardia (VT) or previous instances of aborted cardiac arrest are also candidates for ICD therapy, as a means of secondary prevention [11]. Overall, adherence to expert consensus guidelines on ICD therapy and the appropriate use of beta-blockers form the cornerstone of therapeutic approaches in ARVC management.

Conclusion

This case highlights the importance of considering ARVC in patients who has minimal symptoms and their initial cardiac workup suggests ARVC. Prompt diagnosis should be carried out through comprehensive evaluation of ECGs and ECHO along with Cardiac MRI. This would be very beneficial to carry out appropriate management strategies, thus reducing the risk of life-threatening arrhythmias, heart failure and/or sudden cardiac death. Regular follow-up and discussion with the electrophysiology team for placement of ICDs should also be considered. Screening of family members should also be carried out so that appropriate management strategies can be devised.

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