

A case of Arrhythmogenic right ventricular cardiomyopathy (ARVC) presenting with dizziness—a rare case report

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Keywords: Arrhythmogenic right ventricular cardiomyopathy (ARVC); Dizziness; Fibrofatty.



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Introduction

Arrhythmogenic right ventricular cardiomyopathy (ARVC), previously referred to as Arrhythmogenic right ventricular dysplasia (ARVD), is a genetic disorder which not only involves the right ventricular (RV) myocardium but also the left ventricle (LV)¹. It is characterized by fibrofatty replacement of myocardium, mainly in the regions of right ventricular (RV) outflow tract, area below the tricuspid valve and the right ventricular apex which are collectively referred to as triangle of dysplasia, however recent studies have shown that the RV apex is involved late in the disease course with commonly involved sites being basal inferior and anterior RV and posterolateral LV². Its prevalence ranges from 1 in 2000 to 1 in 5000.³

Abstract

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a genetic disorder. Its hallmark is the fibrofatty replacement of myocardium leading to cardiomyopathy. It can have various presentations which can range from silent disease to sudden cardiac death. Here, we report a case of ARVC in a 42-year-old female who presented to us with dizziness as her first clinical presentation. We clinched the diagnosis with the help of electrocardiogram, echocardiography and cardiac magnetic resonance imaging (CMR).

Case Report

A 42-year-old female presented to our outpatient department with history of intermittent dizziness, mainly on exertion, of 3 days duration. There was no family history of sudden cardiac death. Her clinical examination was unremarkable. Non-cardiac causes of dizziness were also ruled out. Her ECG (figure 1a) at presentation showed complete right bundle branch block with T wave inversion (TWI) in leads V1-4 (minor criterion). We immediately asked for transthoracic echocardiography (TTE) (figure 2a and 2b) which revealed dilated RV, dyskinetic RV apex, dilated RV outflow tract (RVOT) (in parasternal long axis view = 40mm and in parasternal short axis view = 44mm) (major criterion). Her LV function was normal. Her blood investigations including electrolytes were within normal limits. With such ECG and echocardiographic findings we

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suspected ARVC and asked for cardiac MRI (CMR). Her CMR (figure 3) revealed dilated RV along with fibrofatty replacement of RV apex with focal aneurysm/ dyskinetic segment in the upper part. The RV ejection fraction was reduced (RVEF=38%) (major criterion). Her 24 hours holter recording (figure 1b) revealed 4174 ventricular extrasystoles (minor criterion) per 24 hours. As per 2010 revised criteria for diagnosis of ARVC⁴, our patient met one major (from CMR or TTE findings) and two minor criteria (from repolarization abnormalities and arrhythmias) for ARVC, which means she had definite diagnosis of ARVC. The patient was later on advised for electrophysiological study in other cardiac centre.

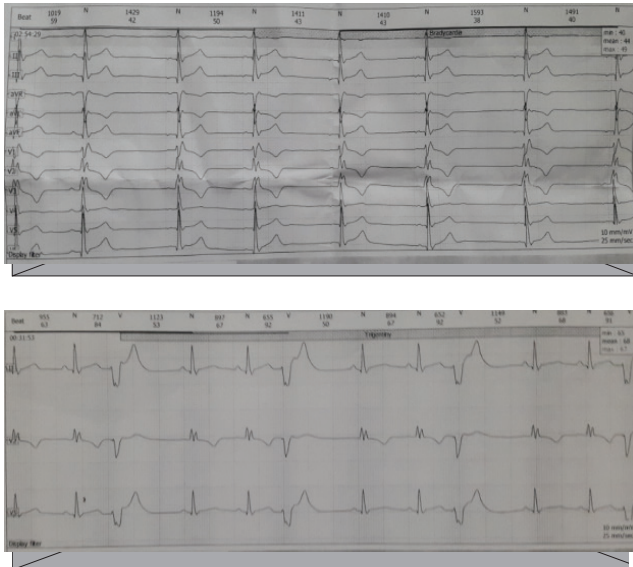


Figure 1a -Baseline ECG showing complete RBBB with T wave inversion in leads V1-4 and Figure 1b-Holter recording showing ventricular extrasystoles

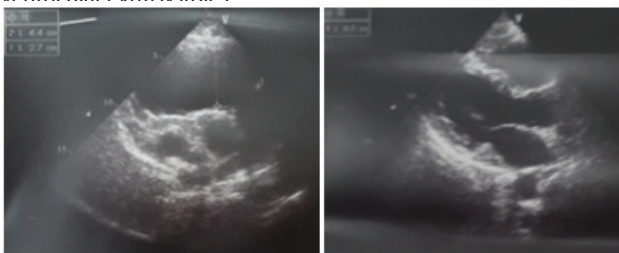


Figure 2a and 2b showing, in TTE, dilated RVOT in PSAX and PLAX views respectively.

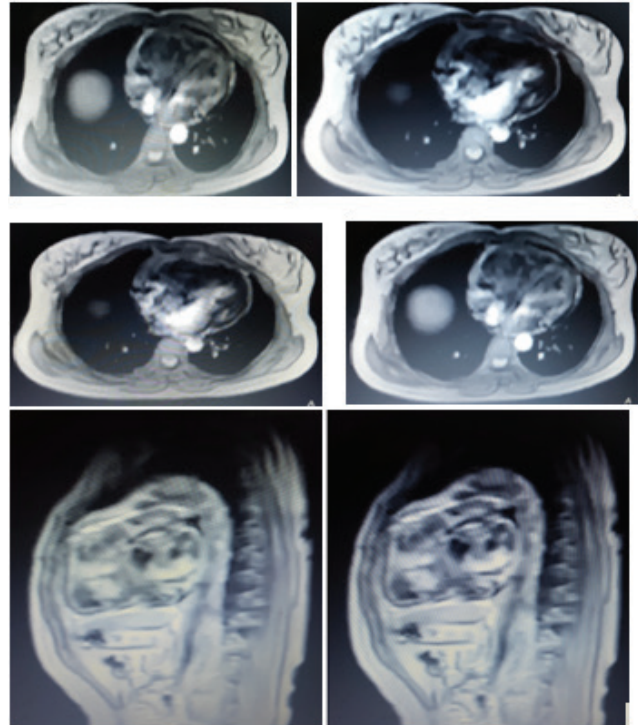


Figure 3-showing various CMR images depicting dilated RV, focal RV aneurysm in the upper part and fibrofatty infiltration of RV apex.

Discussion

ARVC is an important genetic cause of sudden cardiac death in young individuals and athletes. The common age at presentation ranges between 10 to 50 years⁵. Males tend to have complicated disease course⁶.

Aberrant Wnt signaling of desmosomal proteins and direct plakoglobin signaling transforming myocytes into adipocytes with disease progression have been related to fibrofatty replacement of myocardium in ARVC⁷. Multiple genetic mutations in an individual may lead to complicated disease course⁸.

Patients with ARVC can present with various symptoms. In one study, 67% presented with palpitations, 32% with syncope, 27% with atypical chest pain, 11% with dyspnea and 6% with RV failure⁹.

Epsilon wave in the right precordial leads is not a sensitive finding but is specific. These patients may present with typical monomorphic ventricular tachycardia with left bundle branch block pattern with a superior axis¹⁰.

Diagnosis is made according to 2010 revised task force criteria for the diagnosis of ARVC⁴. In our case, we made the diagnosis of definite ARVC after our patient met one major and two minor criteria.

Treatment of ARVC focuses on prevention of fatal ventricular arrhythmias and sudden cardiac death. Patients with definite ARVC should not participate in athletic activity¹¹⁻¹². Beta-blockers decrease the rate of progression of ventricular dysfunction and suppress ventricular arrhythmias as well¹².

Implantable cardioverter defibrillator (ICD) is recommended for those with history of syncope, high risk of sudden cardiac death, life threatening arrhythmias despite drug therapy¹³.

Conclusion

ARVD is a genetic disorder of cardiac myocytes. Its diagnosis is a challenging task. Young patients can be saved from sudden cardiac death if diagnosis is made promptly in suspected cases. Diagnosis should always be suspected in young patients who have history of resuscitated cardiac arrest or aborted sudden cardiac death, syncope and ventricular arrhythmias in the past.

Conflict Of Interest:

None

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