DOI: 10.7860/JCDR/2022/53348.16329

Case Report

Others Section

Arrhythmogenic Right Ventricular Dysplasia Diagnosed in a Patient with Long-term Undiagnosed Syncope

AVI SHAH[1], RAMESH PATEL[2], SANDEEP AGGARWAL[3]

ABSTRACT

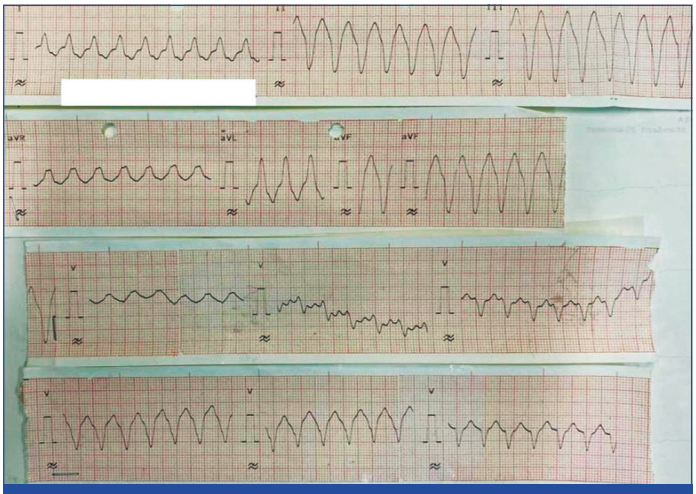
Arrhythmogenic Right Ventricular Dysplasia (ARVD) is an inherited cardiomyopathy which represents progressive replacement of right ventricular myocardium with fibrofatty tissue. In the present case, a 42-year-old male presented to the hospital as he fell down, due to syncope. He had a history of recurrent syncopal in past 4-5 years. His chest radiogram revealed cardiomegaly (CT ratio 0.64), repeat electrocardiogram showed symmetric T-wave inversions and epsilon waves in right precordial V1, V2 and V3 leads, and echocardiogram revealed mild left ventricular dysfunction (ejection fraction - 45%) and dilated right atrium and ventricle. Cardiac Magnetic Resonance Imaging (MRI) confirmed the diagnosis of ARVD.The patient was advised for the placement of an Automatic Implantable Cardioverter Defibrillator (AICD), but he denied for this treatment. Thus, he was managed with anti-arrhythmic drugs and was advised for regular follow-up. The patient was followed-up for six months where episodes of syncope had significantly reduced to one in six months but need of AICD implantation was still there to avoid sudden cardiac death.

Keywords: Cardiac, Defibrillator, Right ventricular cardiomyopathy

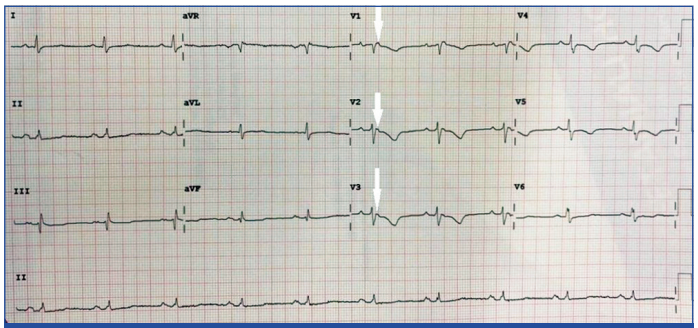
CASE REPORT

A 42-year-old male presented to Emergency Department with syncope (fell down due to syncope). He was a known case of hypertension and Type II diabetes mellitus and was on regular medical treatment. The patient's history also suggested several incidences of recurrent syncopal episodes in the last 4-5 years for which he consulted many neurologists and physicians, but condition remained undiagnosed.

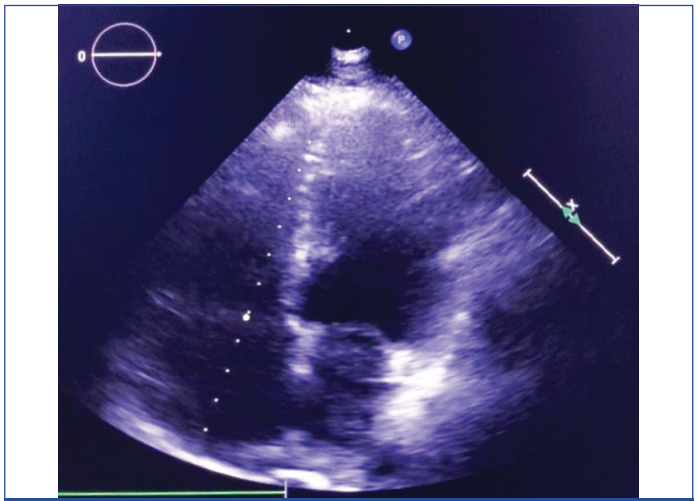
At presentation, patient's heart rate was 186 beats/min with sinus rhythm, blood pressure was 132/86 mmHg, respiratory rate was 17 breaths/min, body temperature was 98.8°F and oxygen saturation of 98% at room air was noted. Cardiopulmonary examination revealed no murmurs and normal chest and heart sounds on auscultation. Any neurological deficit or carotid bruits were not noted. Electrocardiography (ECG) suggested monomorphic ventricular tachycardia [Table/Fig-1] which was normalised to sinus rhythm via Direct Current (DC) shock cardioversion. Chest radiograph revealed cardiomegaly with Cardiothoracic (CT) ratio of 0.64. Repeat ECG after stabilisation showed symmetric T-wave inversions and epsilon waves in right precordial V1, V2 and V3 leads [Table/Fig-2]. Echocardiography revealed mild left ventricular dysfunction (ejection fraction- 45%) and dilated right atrium and ventricle [Table/Fig-3].



[Table/Fig-1]: Electrocardiogram showing monomorphic ventricular tachycardia.

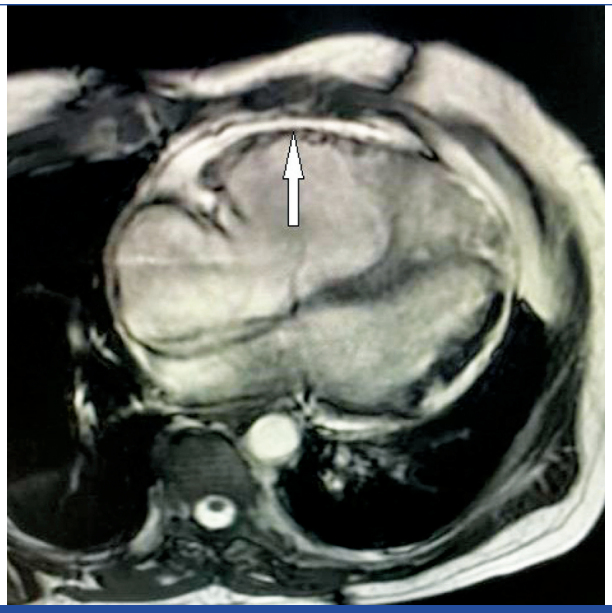


[Table/Fig-2]: Electrocardiogram showing Epsilon waves in leads V1,V2 and V3.



[Table/Fig-3]: Echocardiogram showing dilated right atrium and right ventricle.

Based on these findings, the patient was suspected to have ARVD, and to confirm the same, cardiac Magnetic Resonance Imaging (MRI) was performed. Cardiac MRI suggested notable dilatation of right ventricle and right atrium with significant hypokinesia associated with focal dyskinesia, fibrofatty tissue in the wall of right ventricle, mild thinning of free wall of right ventricle and mild tricuspid regurgitation [Table/Fig-4]. These structural abnormalities, observed on cardiac MRI, confirmed the diagnosis of ARVD. Further, coronary angiography was also performed but displayed normal coronary arteries.



[Table/Fig-4]: Cardiac MRI showing fibrofatty infiltration of free wall of right ventricle.

The patient was advised and explained about the need for the placement of an AICD but because of economic constraints, he denied undergoing AICD implantation. Thus, the patient was managed medically, with metoprolol 25 mg twice a day along with low dose frusemide. Genetic testing for desmosomal complex protein and electrophysiological study were advised to determine the reason for significant right ventricular dysfunction, but he refused to do so due to economical constraints. The patient was followed-up for six months, where episodes of syncope had significantly reduced to one with no fall or injury. But, the need for AICD and electrophysiological study in this patient still remained.

DISCUSSION

The ARVD is an inherited "desmosomal" cardiomyopathy characterised with gradual replacement of right ventricular myocytes by adipocytes and fibrosis. It is a rare and underdiagnosed disease of cardiac myocytes that is typically considered as right ventricular arrhythmias, dilation or aneurysm, sudden cardiac death, and heart failure. It accounts for 5% of sudden cardiac death in young adults and overall, 25% deaths in athletes. Around 30-50% patients represent family history of the ARVD. Several origins of ARVD have been anticipated such as inflammation, infection, apoptosis and degeneration, but lacks adequate evidence to confirm their role in ARVD [1-4].

The clinical manifestation of ARVD includes replacement of right ventricular myocardium with fibrofatty scar tissue, ventricular free wall thinning and hypertrophy, and right ventricular outflow tract enlargement [2]. In early stages of ARVD, left ventricle is usually spared, however it is predominantly involved in advanced stages of disease and may also causes left ventricular failure [5,6].

Diagnosis of ARVD is quite difficult as most patients may remain asymptomatic and if present with symptoms, then clinically mimic myocarditis, sarcoidosis, Brugada syndrome, idiopathic right ventricular outflow tract tachycardia and congenital heart diseases with right chambers overload. Thus, International Task Force (ITF) has given highly sensitive diagnostic criteria of ARVD based on the presence of major and minor criteria involving electrocardiographic, arrhythmic, morphological, histopathologic, and genetic factors [Table/Fig-5] [7]. In the present case, the patient was suspected of ARVD based on his ECG (symmetric T-wave inversions and epsilon waves in right precordial V1, V2 and V3 leads) and echocardiographic examination (dilated right atrium and ventricle). To confirm the same, cardiac MRI was performed.Cardia MRI is a powerful non-invasive diagnostic tool for ARVD as it effectively excludes other causes of right-sided heart failure and thus should be employed in confirmation of ARVD due to its high sensitivity and specificity [2,8,9].

I. Global and/or Regional dysfunction and structural alterations	
Major	• Severe dilatation and reduction of right ventricular ejection fraction with no (or only mild) left ventricular impairment • Localised right ventricular aneurysms (akinetic or dyskinetic areas with diastolic bulging) • Severe segmental dilatation of the right ventricle
Minor	• Mild global right ventricular dilatation and/or ejection fraction reduction with normal left ventricle • Mild segmental dilatation of the right ventricle • Regional right ventricular hypokinesia
II. Tissue characterisation of wall	
Major	• Fibrofatty replacement of myocardium on endomyocardial biopsy
III. Repolarisation abnormalities	
Minor	• Inverted T waves in right precordial leads (V2 and V3) in people aged >12 years, in absence of right bundle branch block
IV. Depolarisation/Conduction abnormalities	
Major	• Epsilon waves or localised prolongation (>110 ms) of the QRS complex in right precordial leads (V1-V3)
Minor	• Late potentials (signal-averaged ECG)
V. Arrhythmias	
Minor	• Left bundle branch block type ventricular tachycardia (sustained and non-sustained) by ECG, Holter or exercise testing • Frequent ventricular extra-systoles (>1000/24 hours) by Holter
VI. Family history	
Major	• Familial disease confirmed at necropsy or surgery
Minor	• Family history of premature sudden death (<35 years) due to suspected right ventricular dysplasia • Familial history (clinical diagnosis based on present criteria)

[Table/Fig-5]: Criteria for diagnosis of Arrhythmogenic Right Ventricular Dysplasia (ARVD).

There is no cure of ARVD. The goal should be to prevent sudden cardiac death by managing ARVD with antiarrhythmic medications, radiofrequency ablation, or defibrillator implantation. Generally, treatment of choice in patients with ARVD is electrophysiological study followed by radiofrequency ablation of fibrofatty tissue, however, AICD implantation is considered as a gold standard management option in high-risk patients. Cardiac transplant has been reserved as a last resort treatment [6,10]. In the present case, the patient was advised to implant defibrillator to prevent sudden cardiac death, but he rejected because of financial constraints and thus was conservatively managed with anti-arrhythmic drugs and advised for lifestyle modification to prevent severe consequences.

CONCLUSION(S)

Diagnosis of ARVD, an inherited condition, is a critical and challenging task for every cardiologist as it represents high frequency of misdiagnosis. Proper evaluation of syncope is important along with 24-hour holter or event loop recorder as a necessity tool in management of syncope. Cardiac MRI, due to its both qualitative and quantitative analysis, plays a very important role in the diagnosis of ARVD. In this patient, intracardiac defibrillator implantation and fibrofatty tissue radiofrequency ablation might have provided significant results but could not be possible without patient's consent and thus ARVD was managed with conservative treatment with an advised of regular clinical follow-up.

REFERENCES

[1] Maithili D, Pamuru PR, Mohiuddin K, Remersu S, Calambur N, Oruganti SS, et al. Clinical picture of arrhythmogenic right ventricular dysplasia/cardiomyopathy patients from Indian origin. Indian Pacing and Electrophysiology Journal. 2009;9(1):05-14.
[2] Jain A, Tandri H, Calkins H, Bluemke DA. Role of cardiovascular magnetic resonance imaging in arrhythmogenic right ventricular dysplasia. J Cardiovasc Magn Reson. 2008;10(1):01-14.
[3] Khan A, Mittal S, Sherrid MV. Arrhythmogenic right ventricular dysplasia: From genetics to treatment. Anatolian Journal of Cardiology/Anadolu Kardiyol Derg. 2009;9(Suppl 2):24-31.
[4] James CA, Calkins H. Update on arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C). Curr Treat Options Cardiovasc Med. 2013;15(4):476-87.

[5] Rao U, Agarwal S, Gilbert T. Arrhythmogenic right ventricular cardiomyopathy (ARVC): Case report and review of literature. Heart Asia. 2014;6(1):145-49.
[6] Corrado D, Wichter T, Link MS, Hauer R, Marchlinski F, Anastasakis A, et al. Treatment of arrhythmogenic right ventricular cardiomyopathy/dysplasia: An international task force consensus statement. Eur Heart J. 2015;36(46):3227-37.
[7] Marcus FI, McKenna WJ, Sherrill D, Basso C, Bauce B, Bluemke DA, et al. Diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia: Proposed modification of the task force criteria. Circulation. 2010;121(13):1533-41.
[8] Ortiz PD, Agarwal R, Reddy R, Lenge VV, Civitello AB, Flamm SD. Arrhythmogenic right ventricular dysplasia: Initial presentation in a middle-aged woman. Tex Heart Inst J. 2006;33(3):386-88.
[9] Rastegar N, Te Riele AS, James CA, Bhonsale A, Murray B, Tichnell C, et al. Fibrofatty changes: Incidence at cardiac MR imaging in patients with arrhythmogenic right ventricular dysplasia/cardiomyopathy. Radiology. 2016;280(2):405-12.
[10] Dalal D, Nasir K, Bomma C, Prakasa K, Tandri H, Piccini J, et al. Arrhythmogenic right ventricular dysplasia: A United States experience. Circulation. 2005;112(25):3823-32.

PARTICULARS OF CONTRIBUTORS:
1. Fellow, Department of Medicine, Geetanjali Medical College and Hospital, Udaipur, Rajasthan, India.
2. Associate Professor, Department of Cardiology, Geetanjali Medical College and Hospital, Udaipur, Rajasthan, India.
3. Fellow, Department of Cardiology, Geetanjali Medical College and Hospital, Udaipur, Rajasthan, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Ramesh Patel,
Associate Professor, Department of Cardiology, Geetanjali Medical College and Hospital, Udaipur, Rajasthan, India.
E-mail: drrameshpatel1982@gmail.com

PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Nov 08, 2021
- Manual Googling: Feb 22, 2022
- iThenticate Software: Apr 04, 2022 (19%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Nov 17, 2021**
Date of Peer Review: **Feb 08, 2022**
Date of Acceptance: **Feb 23, 2022**
Date of Publishing: **May 01, 2022**