Case Report

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Arrythmoenic Right Ventricular Dysplasia

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Abstract

We present a 45 year nonsmoker male presenting to the Cardiology OPD of AIIMS, Bhubaneswar presenting with effort dyspnea NYHA class IV with features of right heart failure with massive edema of both feet diagnosed to be Cor Pulmonale outside, EKG revealed typical epsilon wave in leads V_1 to V_3 and echo revealed fibrofatty infiltration of RV apex with reduced RV ejection fraction (35%), Cardiac MRI revealed the same and withdrawing all anti PAH drugs the patient was put on antifailure medications presently doing well. Being a rare entity arrythmogenic right ventricular cardiomyopathy (ARVC) stands out a prominent entity in young RV failure.

Keywords: Cardiomyopathy; Palpitation and Failure.

Introduction

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited myocardial disorder characterised by fibro-fatty infiltration affecting both the right ventricle (RV) and left ventricle (LV) with a wide phenotypic expression. Its true prevalence is unknown, with estimates of between 1 in 2000 and 1 in 5000. ARVC most commonly presents with palpitations, nonsustained ventricular tachycardia (VT) and syncope and sudden cardiac death, but many patients are initially asymptomatic [1]. Four stages of ARVC evolution have been described

- Latent,
- Symptomatic ventricular arrhythmias of RV origin,
- Isolated right heart failure,
- Dilated biventricular cardiomyopathy.

Triangle of dysplasia in ARVC constitutes outflow tract, apex and sub-tricuspid area where as in our case it was classical apical involvement. ARVC has been unusually prominent as a cause of death in young athletes in Italy of Veneto region. ARVC is a desmosomal disease, and this would accord with the thinner walled RV with morphologic change. ARVC is diagnosed with modified task force criteria [2] with RV ejection fraction ≤ 40% on MRI, nonsustained or sustained VT of LBBB morphology and superior axis, a pathologic mutation associated with ARVC and the presence of T wave inversion in right precordial leads. Late contrast enhancement of LV in cardiac MRI is an important radiological sign in diagnosis although plakoglobin deficiency once considered an important marker now it is also well observed in DCM as well. Being familial in nature we screened in one son and daughter of the patient and found no morphologic evidence of RV involvement in them with normal biventricular function. Our case is a rare entity of young heart failure with well evident fibrofatty infiltration of RV apex in echo with epsilon wave in precordial ECG although better appreciated in signal averaged ECG.

Case Report

Forty five year old male non smoker, non diabetic, nonhypertensive presented with effort dyspnea Class IV with ascites and massive pedal edema diagnosed to be Cor Pulmonale outside, ECG revealed classical epsilon wave in anterior precordial leads with T wave

inversion in inferolateral leads (Figure 1) suggestive of biventricular involvement of the disease, Echo revealed complete fibrofatty infiltration of RV almost more than half of the RV with septal encroachment (Figure 2) with severe RV dysfunction and IVC plethora with almost normal LV systolic function without evidence of PAH with mild TR. RV was dilated with mild dilation of RA secondary to nonhypertensive TR. ARVC was diagnosed which was further revealed by Cardiac MRI with late gadolinium enhancement also evident in LV apicolateral wall (Figure 3). Patient was subjected to 24 hour holter monitoring which revealed episodic runs of NSVT . Patient was treated with antifailure measures, fluid and activity restriction with oral amiodarone to take care of episodic NSVT as patient was unable to afford for ICD therapy and patient was symptomatically better in follow up with regression of ascites and pedal edema with improved symptomatic class. *Our case is unique in the fact that fibrofatty involvement of more than half of the RV was so beautifully elucidated in echo that Cardiac MRI was hardly necessary to make a big difference in diagnosis.*

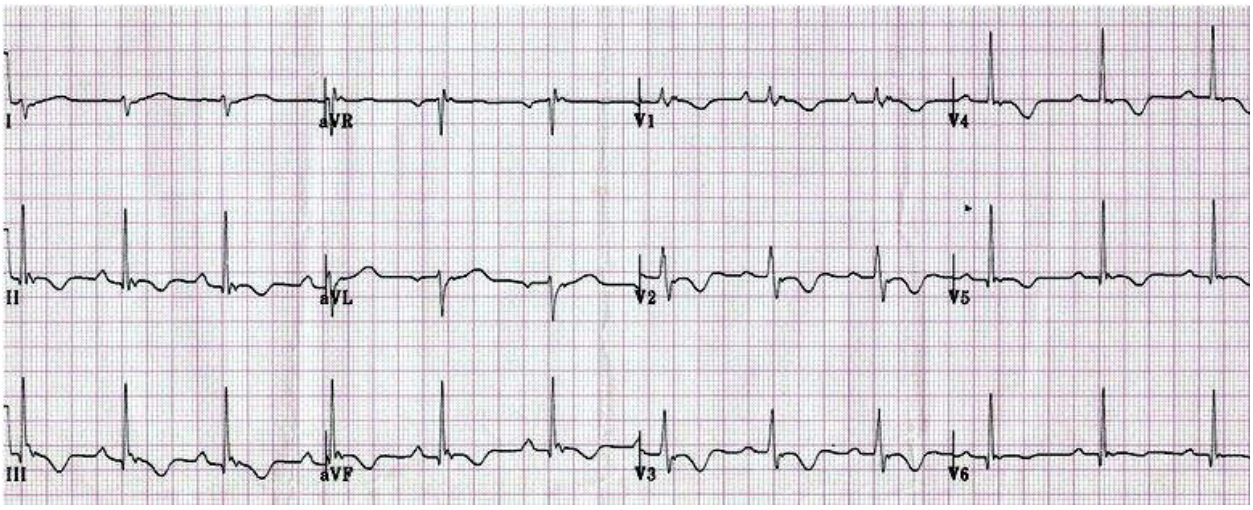

Fig. 1: Epsilon wave in V_1 to V_3 with evidence of same in inferolateral leads

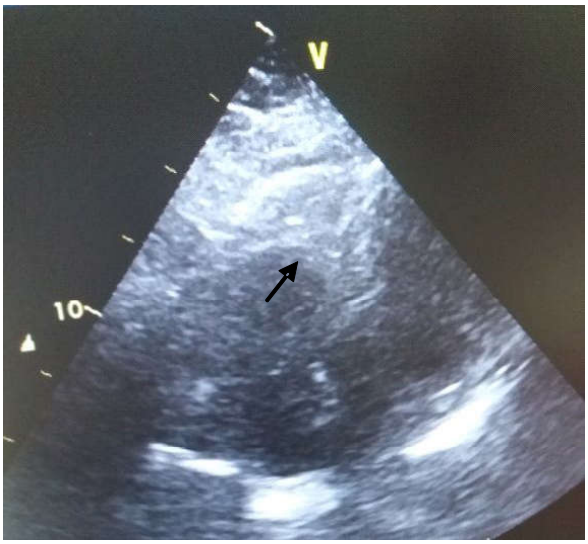

Fig. 2: Fibrofatty infiltration of RV Apex in Echo

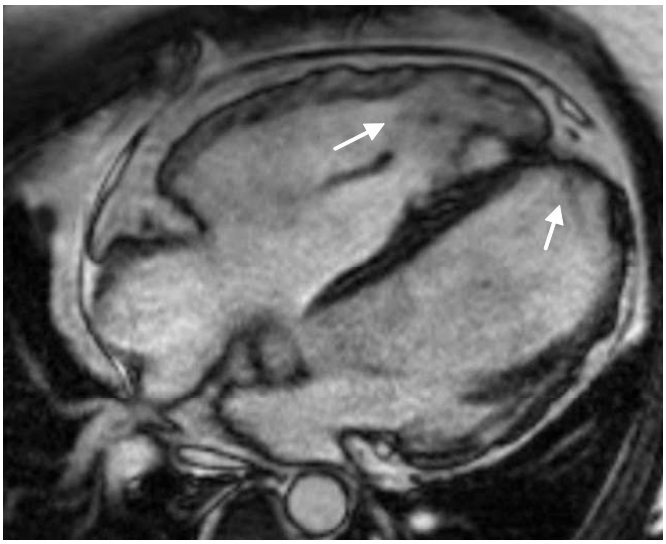

Fig. 3: Fibrofatty infiltration of RV apex and LV apicolateral wall in cardiac MRI

Discussion

Genetic screening is commercially available for ARVC but the patient could not afford for it. Five desmosomal genes inherited in autosomal dominant manner are associated with ARVC; plakophilin-2 (PKP2), desmoplakin (DSP), desmoglein (DSG-2), desmocollin (DSC2), and plakoglobin (JUP) [3].

Plakophilin mutations reported to be nearly 50 percent present at an earlier age and the survival is lower. Plakoglobin mutations are associated palmoplantar hyperkeratosis and woolly hair. Desmoglein and Desmocollin mutations have been associated with predominantly left ventricular involvement. Desmoplakin mutations have been identified in two autosomal recessive syndromes i.e. Carvajal and a "Naxos like" syndrome. The patient was refrained from strenuous exercise (e.g. basketball, squash, skiing, soccer, singles tennis, cycling, running and windsurfing) as exercise might be deleterious with the thinner walled right ventricle more vulnerable to cellular disruption. Beta-blockers, sotalol and amiodarone have been used to take care of NSVT or VT in ARVC. There is consensus that cardiac arrest survivors or patients with sustained VT should have an ICD. Radiofrequency ablation is moderately successful but recurrence rates is higher than for outflow tract VT because of the generalised nature of the disease and ICD treatment is necessary regardless of outcome [4] although patient could not afford for it due to economic constraint. Intractable heart failure needs transplantation in rare cases. Our case is a rarest case of well evident fibrofatty infiltration of RV apex so that MRI could help us a little although necessary as per modified task force criteria. We thought to put an ICD in this patient with antifailure measures but the patient was from remote district with low financial background. Most of the ARVC patients are misdiagnosed as Corpulmonale or DCM although a close look at the Echo can solve the mystery many a time. Close scrutiny of the family members with advice not to undergo strenuous exercise is always advisable as it unmasks RV thinning and genetic screening is not always possible to perform in all family members. MRI scanning is also recommended for adults and older children beginning age 10-12 years and should be repeated every 5 years until age 30, and once more at age 40 [5]. If the MRI is minimally abnormal (mild segmental or global enlargement) the scan should be repeated after 3 years.

Conclusion

We represent a rare case of ARVC with well evident epsilon wave, ECG evidence of biventricular involvement and well seen fibrofatty infiltration of more than half of the RV in Echo. Close scrutiny of a routine echo can bring out this mysterious disease and save the life of the suffering and family members. *Notwithstanding the potential for sudden death in a minority, it is important to emphasize that most diagnosed patients can lead full and normal*.

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