A Case of Arrythmogenic Right Ventricular Cardiomyopathy (ARVC)

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ABSTRACT

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a common cause of sudden cardiac death in young adults. It is a disease that has a wide spectrum of presentation. Early identification of the disorder is of paramount importance considering the fatal complications. Here we report one such case.

Introduction :

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Arrhythmogenic right ventricular cardiomyopathy (ARVC) is characterized by fibrofatty degeneration mainly in the right ventricular myocardium.[1] The prevalence of ARVC is reported to be 0.02-0.1%[2,3] Progressive right ventricular dysfunction, ventricular arrhythmias, and sudden cardiac death are the clinical features.

Case Report

A 38-year-old male, presented with sudden onset palpitations, chest pain and dizziness since one hour. Patient was a treated case of Pulmonary tuberculosis ten years ago and a smoker since last five years. Patient was non alcoholic. There was no family history of sudden cardiac death; or any arrhythmias detected in past. Patient was brought to the emergency department. On examination his pulse was not palpable and blood pressure was not recordable. Heart rate - 250/min. Patient was given cardio version of 200J after which ECG changes reverted. He was then started on amiodarone infusion and oral Metoprolol.

Patient was investigated. Haemoglobin - 11 gm/dl, Total leucocyte count - 7000/mm^3, platelets 2.2 lakhs/mm^3. Serum electrolytes, calcium and magnesium levels were normal. Kidney function test, liver function test, Thyroid function test were normal. Chest radiograph was normal. 2D Echocardiography was not suggestive of any abnormality. Coronary angiography was normal. Right ventricular angiogram revealed increased volume of RV (Right Ventricle) with generalized hypokinesia and mild RV dysfunction. RV outflow tract was normal in size and dimension.

Electrophysiological studies were done. On incremental ventricular pacing there was evidence of central conduction with no evidence of accessory pathway. On programmed stimulation from right ventricle with isoprenaline infusion sustained monomorphic VT (ventricular tachycardia) was inducible. VT had Left bundle branch block morphology with left axis deviation suggesting origin from RV. IV adenosine produced ventriculoatrial dissociation with ongoing tachycardia confirming VT. VT could not be terminated with RV pacing. Patient was advised Automated implantable cardioverter defibrillator (AICD) implantation and was discharged.

Discussion :

ARVC is a genetic disorder that affects areas on surface of heart muscle cells which link the cells together, desmosomes[4]. ARVC is inherited as an autosomal dominant disorder, and hallmark is replacement of normal myocardium with fibrofatty infiltrates that render it hypokinetic leading to a multitude of arrhythmias with right ventricular origin. Autosomal recessive variants are also seen (Naxos disease) with mutation in similar protein, plakoglobin[5]. Rarely nongenetic diseases such as congenital anomalies and viral or inflammatory myocarditis may produce fibrofatty infiltration of myocardium.

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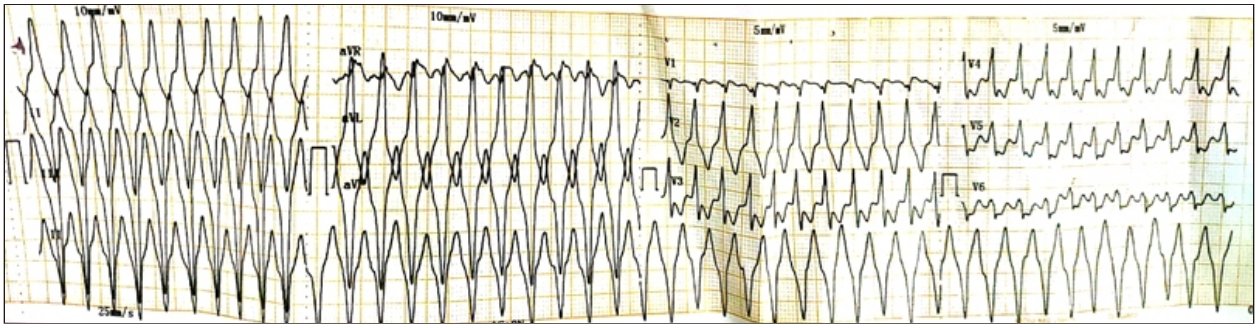

Figure 1 : **ECG on admission showing monomorphic Ventricular tachycardia with HR-250/min and Left axis deviation**

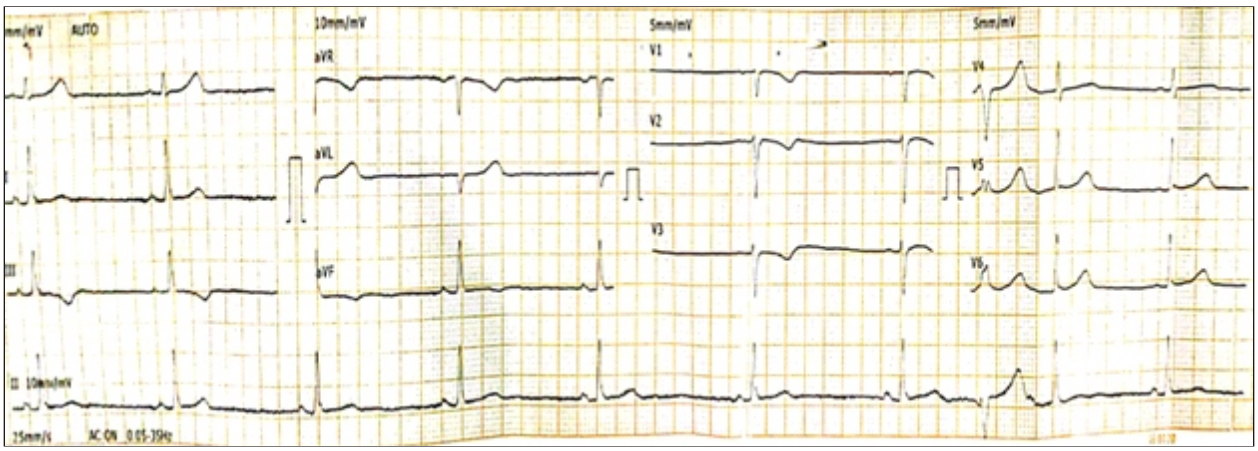

Figure 2 : **ECG after DC shock with normal axis and T wave inversion in leads V1-V3**

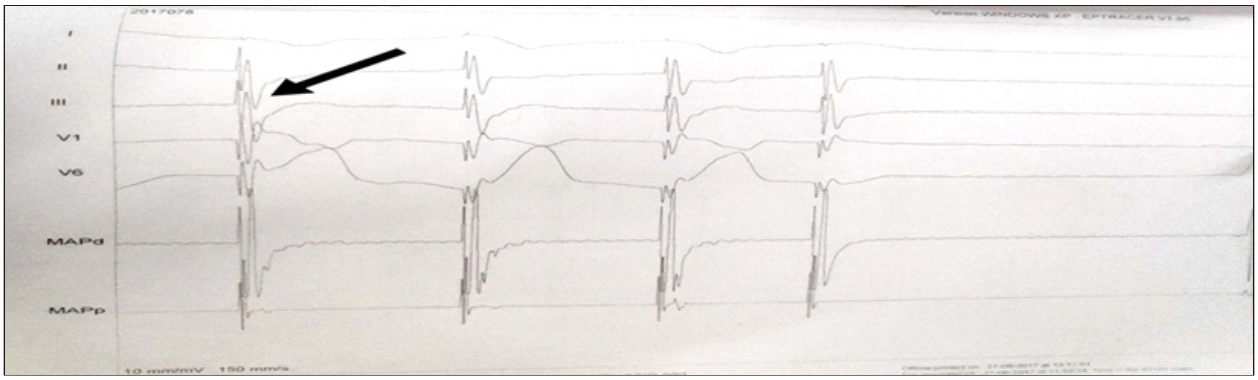

Figure 3 : **Black arrow showing epsilon waves during EPS studies**

ARVC is a cause of sudden cardiac death in children and athletes and usually manifests before the age of forty. Although ARVD is a genetic disease, genetic testing is not mandatory for the diagnosis of the same. The International Task Force Criteria has been established in 1995 and modified later with improved diagnostic accuracy[6,7].

In this case patient fulfilled two major criteria and also two minor criteria. Clinical features of ARVC vary from slight lightheadedness to sudden unexplained death. The attacks are mostly predisposed by exercises. The overall mortality rate of condition is 19%[8] which makes it mandatory to suspect the problem in a young adult with indolent

Table 1 : **International Task Force ARVC diagnostic criteria (2010)**

	Major criteria	Minor criteria
Global and regional dysfunction and structural alterations	Severe dilation and reduction of right ventricular ejection fraction with no/mild Left ventricular impairment Localized right ventricle aneurysms Severe segmental dilation of right ventricle	Mild global right ventricular dilation or ejection fraction reduction with normal left ventricle Mild segmental dilation of right ventricle Regional right ventricle hypokinesia
Tissue characterization of walls	Fibrofatty replacement of myocardium on endomyocardial biopsy	
Repolarization abnormality		Inverted T waves on right precordial leads (V2,V3) (Age > 12 years and absence of right bundle branch block
Depolarization/conduction abnormalities	Epsilon waves or localized prolongation of QRS complex in right precordial leads	Late potentials(signal averaged ECG)
Arrhythmias		LBBB type ventricular tachycardia (sustained or nonsustained) Frequent premature ventricular contractions (>1000/24 hrs)
Family history	Family history of ARVC confirmed on autopsy or surgery	Family history of ARVC clinically and independently diagnosed Familial history of premature sudden death (<35 years) owing to suspected ARVC

The diagnosis is established by the presence of two major criteria / one major and two minor / four minor criteria

symptoms such as lightheadedness. There are case reports of late presentation of ARVC, even in the seventies because of its occasional mild course. There are also reports of ARVC complicating pregnancy with shortness of breath and palpitations during second trimester[9]. As clinical presentations vary significantly, suspicion should be kept if there are unexplained T wave inversions in septal leads in a young adult. He should be followed up with an echocardiogram of heart and more importantly a cardiac magnetic resonance imaging. Cardiac magnetic resonance imaging in addition to fibrofatty infiltrations of myocardium can also pick up reduction of myocardial thickening, dilatation, and aneursymal changes with reduction in wall motion as well. The goal of management is to prevent sudden cardiac death. Beta-adrenergic blocking agents as well as amiodarone may be tried to prevent recurrence of arrhythmias. Implantable cardiac defibrillator is a tried out option among those patients who are at high risk. In those who are resistant to all these, radiofrequency ablation may be tried. However, it is difficult as fibrofatty infiltration is patchy and segmental. All patients should be barred from strenuous labor and exercises, and relatives should be screened with ECG and echocardiogram and preferably cardiac magnetic resonance imaging.

Conclusion :

ARVD is a cause of sudden cardiac death. As the clinical features and course of illness varies significantly, a huge amount of suspicion should be kept in mind if any young adult turns up with suggestive symptoms. We are presenting this case as a prototype of a fatal genetic disease to create

awareness among practitioners and make them competent enough to offer a treatment even if it manifests with or without family history.

References :

1. F.I. Marcus, G.H. Fontaine, G. Guiraudon, R. Frank, J.L. Laurenceau, C. Malergue, Y. Grosgogeat Right ventricular dysplasia: a report of 24 adult cases Circulation, 65 (1982), pp. 384-398
2. A. Nava, B. Bauce, C. Basso, M. Muriago, A. Rampazzo, C. Villanova, L. Daliento, G. Buja, D. Corrado, G.A. Danieli, G. ThieneClinical profile and long-term follow-up of 37 families with arrhythmogenic right ventricular cardiomyopathy J Am Coll Cardiol, 36 (2000), pp. 2226-2233.
3. S. Peters, M. Trümmel, W. MeynersPrevalence of right ventricular dysplasia-cardiomyopathy in a non-referral hospital Int J Cardiol, 97 (2004), pp. 499-501.
4. Fressart V, Duthoit G, Donal E, Probst V, Deharo JC, Chevalier P, et al. Desmosomal gene analysis in arrhythmogenic right ventricular dysplasia/cardiomyopathy: Spectrum of mutations and clinical impact in practice. Europace 2010;12:861-8.
5. Antoniades L, Tsatsopoulou A, Anastasakis A, Syrris P, Asimaki A, Panagiotakos D, et al. Arrhythmogenic right ventricular cardiomyopathy caused by deletions in plakophilin-2 and plakoglobin (Naxos disease) in families from Greece and Cyprus: Genotype-phenotype relations, diagnostic features and prognosis. Eur Heart J 2006;27:2208-16.
6. Marcus F, Basso C, Gear K, Sorrell VL. Pitfalls in the diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia. Am J Cardiol 2010;105:1036-9.
7. 4. Dalal D, Nasir K, Bomma C, Prakasa K, Tandri H, Piccini J, et al. Arrhythmogenic right ventricular dysplasia : A United States experience. Circulation 2005;112:3823-32.
8. 5. Hulot JS, Jouven X, Empana JP, Frank R, Fontaine G. Natural history and risk stratification of arrhythmogenic right ventricular dysplasia/ cardiomyopathy. Circulation 2004;110:1879-84.
9. Güdücü N, Kutay SS, Ozenç E, Ciftçi C, YigiterAB, Isçi H. Management of a rare case of arrhythmogenic right ventricular dysplasia in pregnancy: A case report. J Med Case Rep 2011;5:300.