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Case Report on Arrhythmogenic Right Ventricular Dysplasia

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Abstract :

Arrhythmogenic cardiomyopathy (AC) is a genetic disease characterized by fibro-fatty replacement of either ventricles in isolation or in combination. It may cause tachyarrhythmias to sudden cardiac death, especially in young adult. When ventricular tachycardia (VT) is the principal manifestation then the condition is termed as arrhythmogenic right ventricular dysplasia (ARVD). Here we present a case of ARVD in a 36 years old hypertensive male who presented with sudden onset of chest tightness, palpitations, breathing difficulty, dizziness for 20 minutes. In emergency department his electrocardiogram (ECG) showed sustained VT. After 200 joule DC cardioversion he was reverted to sinus rhythm, then resting ECG showed T-wave inversion (TWI) in V1-V3 along with epsilon wave. His echocardiogram also revealed dilated right ventricle (RV) along with hypertrabeculation, RV wall motion abnormality and low RV ejection fraction (EF), his coronaries were normal on coronary angiogram (CAG). He was discharged from the hospital after Implantable Cardioverter Defibrilator (ICD) implantation with beta-blocker and advised to restrict excessive physical activity.

Keywords : Arrhythmogenic right ventricular dysplasia, Ventricular tachycardia, Epsilon wave, Implantable cardioverter defibrillator.

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Introduction :

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a genetic progressive disease characterized clinically by life threatening ventricular arrhythmia and structurally by fibro-fatty replacement of right ventricular wall. When ventricular tachycardia (VT) is principal manifestation of ARVC then the condition is termed as arrhythmogenic right ventricular dysplasia (ARVD).[1]

The prevalence of ARVC is range from 1 in 1000 to 1 in 5000 of general population which could be increase with increasing awareness of its existence.[2] The ratio of male and female is 3:1. It has an autosomal dominant inheritance pattern with variable penetration, although recessive traits with cutaneous manifestations exist.[3]

This autosomal dominant inherited cardiomyopathy is caused by mutation in genes encoding for desmosomal proteins of intercalated disk. This disease progression have three phases-concealed stage, electrical stage and structural stage.[4]

ARVC typically present between second to fifth decades of life. Symptoms varies from palpitation, dizziness, syncope, chest pain, features of right heart failure to cardiac arrest.

ECG shows evidence of ventricular premature contraction to paroxysmal episodes of ventricular tachycardia, T wave inversion V1-V3 (85% of patients) and epsilon waves (most specific finding seen in 50 %patients). ARVC is managed by anti-arrhythmic agents, ablation and by primary or secondary prevention of sudden cardiac death (SCD) with implantable cardiac defibrillator (ICD).

Case report :

A 36 years old hypertensive male was admitted to hospital with sudden onset of palpitation, chest tightness, breathing difficulty and dizziness for 20 min while he was waiting in out patient department (OPD) for cardiac consultation .

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He had H/O similar attack of pre-syncope for two times within last 6 months. He was a smoker, he had no H/O any substance abuse and he was getting bisoprolol and clinidipine for last 2 years for his hypertension. There was no family history of heart disease or sudden cardiac death.

On arrival, he was haemodynamically unstable and there was no evidence of heart failure. ECG showed sustained ventricular tachycardia of LBBB pattern (Fig-1). Ventricular tachycardia was reverted to sinus rhythm by synchronized DC cardioversion of 200 joule. His post cardioversion 12-lead ECG showed inverted T-waves in V1-V3 with epsilon wave (Fig-2).

After stabilization, on examination, the young male was lying comfortably flat on the bed, well oriented with time, place, and person. His vital signs were stable, on lung auscultation there was normal vesicular breath sounds bilaterally while on precordial auscultation the first and the second heart sounds were audible normally with equal intensity without any murmur.

His high sensitive troponin I was weakly positive (0.45ng/ml). Other laboratory tests like complete blood count, electrolytes, thyroid, renal,liver function test, and chest X-ray were normal.

Echo showed: dilated right atrium, right ventricle with hypertrabeculation of right ventricular cavity. Right ventricular ejection fraction (EF) was 30 % with hypokinesia present (Fig.3). Left ventricle was normal with 60 % EF. His pulmonary artery systolic pressure was 35 mmHg. He was adviced to do cardiac magnetic resonance imagine (MRI) followed by coronary angiogram (CAG).But patient refused to do cardiac MRI. So CAG was carried out which showed normal coronaries (Fig-4).

Our patient met 2 major criteria in repolarization and depolarization abnormality categories (TWI in v1 to v3 and Epsilon waves in v1- v3). He also met major criteria of arrhythmia categories (Terminal activation duration of QRS e" 55ms and sustained VT of right ventricular outflow configuration, LBBB morphology) from the Revised Task Force Criteria of 2010 which led to definite diagnosis of ARVC in this patient.[5]

Our patient also met minor criteria of echocardiogram. As patient fulfilled the 2 major criteria of Task force by ECG and minor criteria by echocardiogram, so implantable cardioverter defibrilator (ICD) was implanted and was advised to continue beta-blocker and has been asymptomatic ever since.

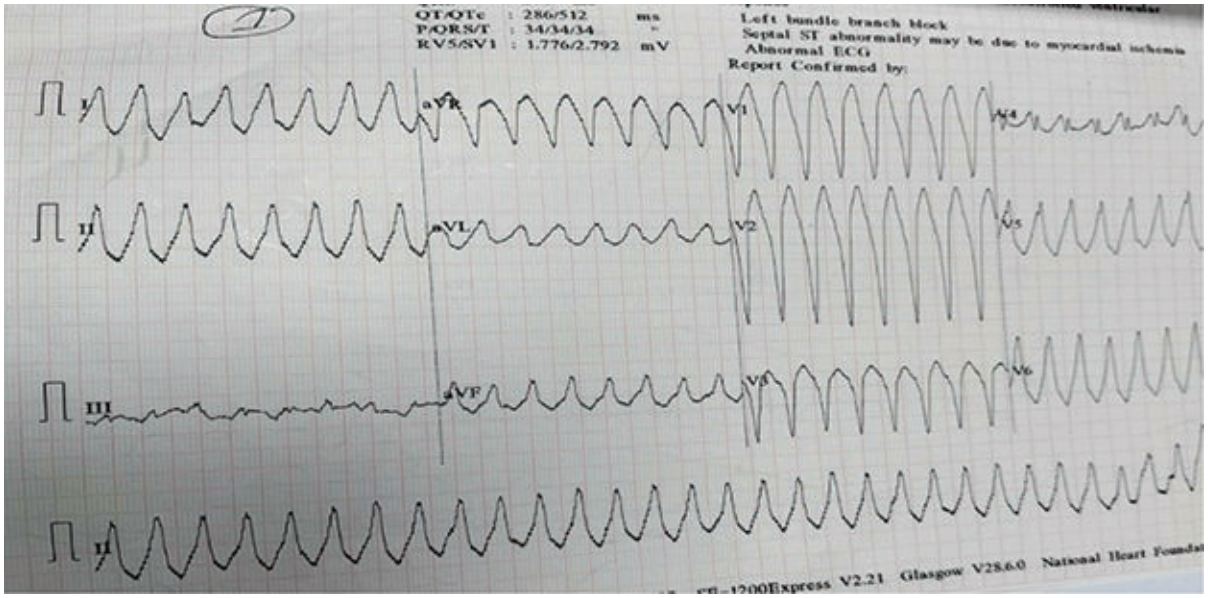

Fig.-1: *ECG shows sustained ventricular tachycardia of LBBB morphology.*

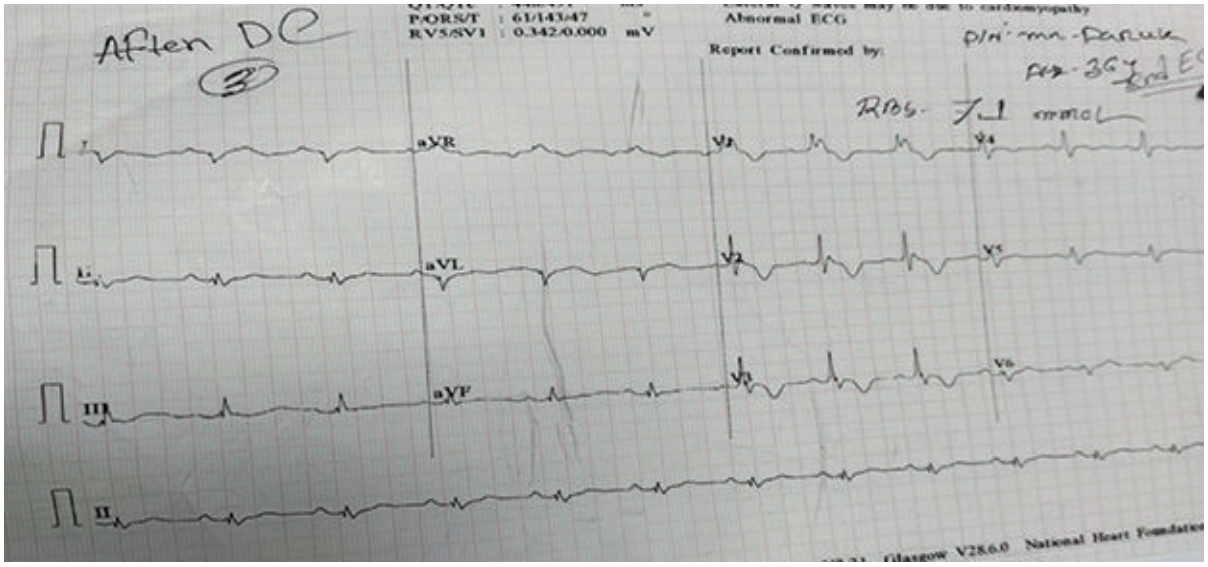

Fig.-2: *After DC cardioversion ECG shows inverted T waves in precordial leads and epsilon wave at the end of QRS complex.*

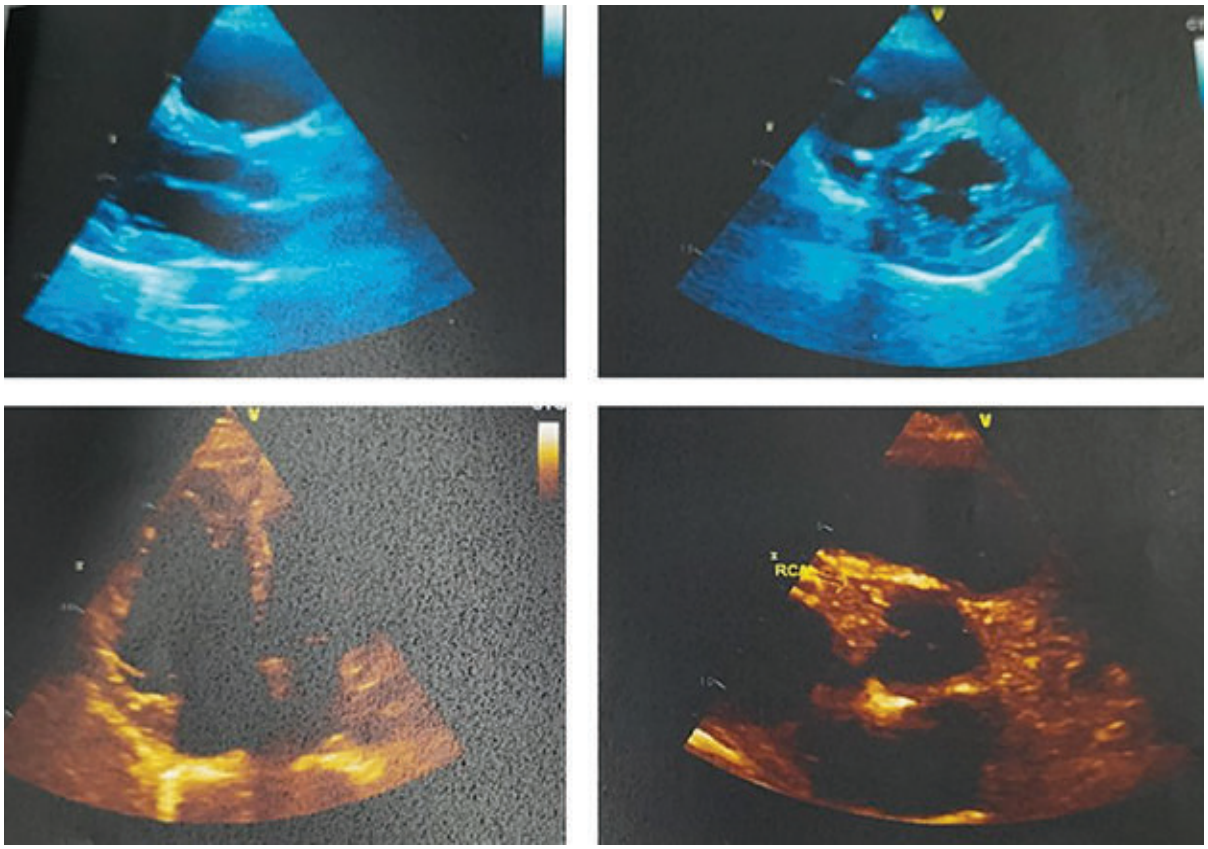

Fig.-3: *Echocardiogram shows dilated right ventricle with trabeculation and dilated right atrium.*

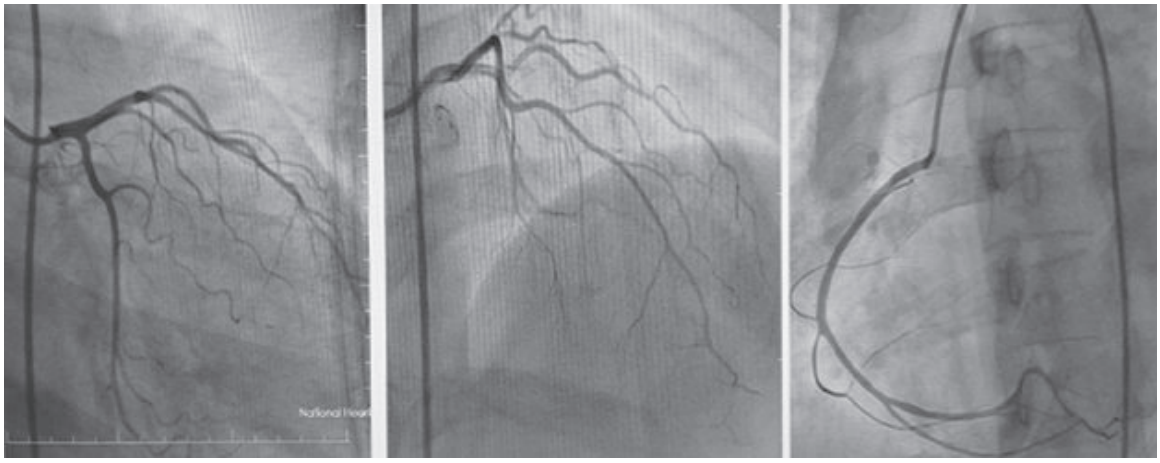

Fig.-4: *CAG shows normal coronaries.*

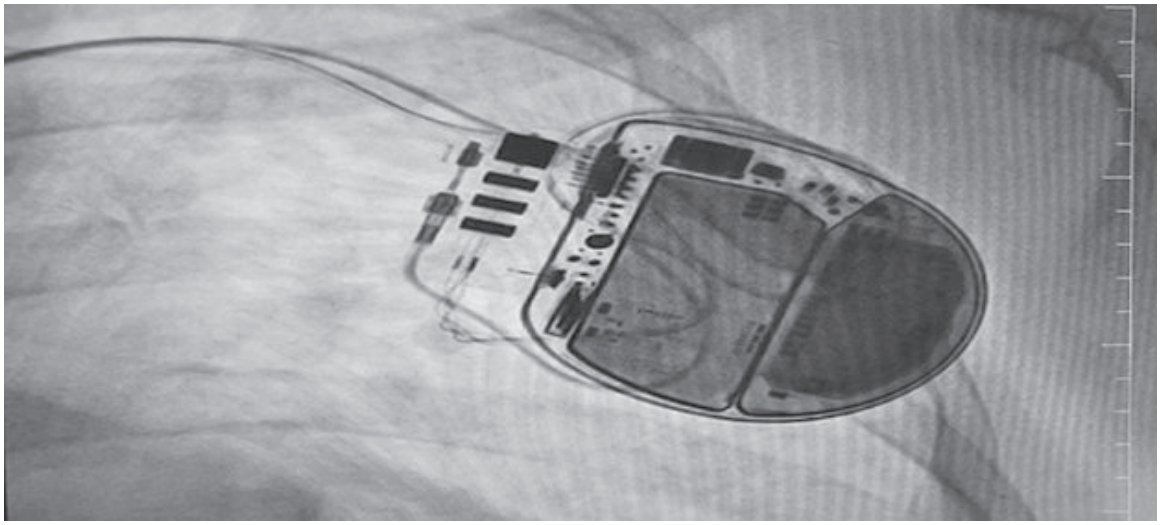

Fig.-5: *ICD in situ*

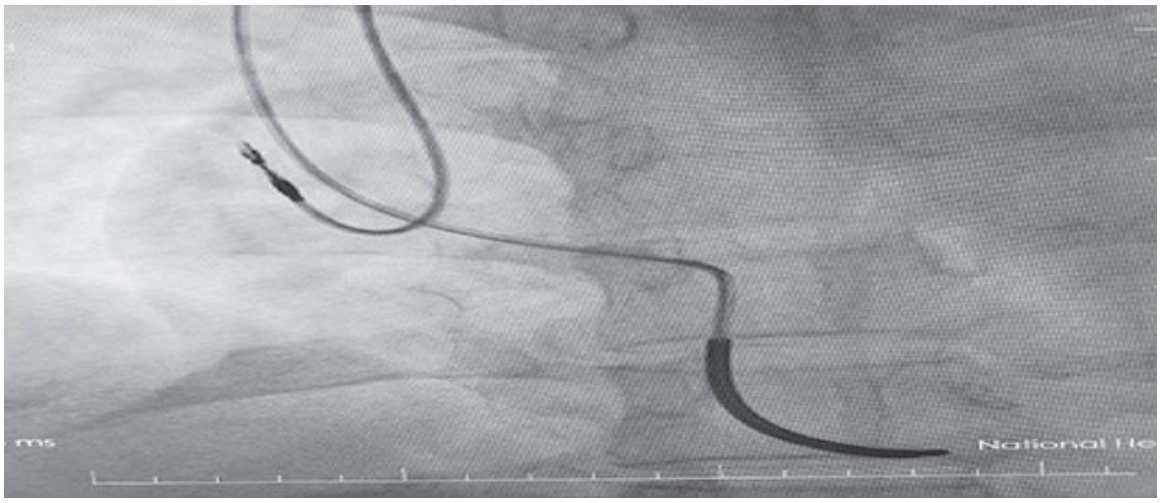

Fig.-6: *ICD lead in Right atrium and Right ventricle.*

Discussion :

Arrhythmogenic right ventricular dysplasia (ARVD) is an underdiagnosed cardiomyopathy which commonly involves progressive fibrous and fatty tissue replacement of myocardium in the inflow, apical and outflow portions of the right ventricle also known as 'The triangle of dysplasia', resulting in ventricular tachycardia with increased risk of sudden death among young adults.[6-8] It can affect any age group but the predominant is male patient in the third decade of life[9] and our male patient was from fourth decade of his life presented with the feature of VT though his troponin-I was weakly positive that can be explained by possibly the markers was released from diseased apoptotic myocardium following DC shock.

Recently, a standardized diagnostic criterion for diagnosis of ARVD was proposed by Marcus et al.[5] In 2010, experts have proposed certain diagnostic criteria for the detection of ARVC called the revised task force criteria. The diagnosis of ARVC is currently based on the presence of major and minor criteria of Task Force encompassing (i) global/regional dysfunction detected by ECHO, MRI, RV angiography, (ii) endomyocardial biopsy, (iii) repolarization abnormality,(iv)depolarization abnormality and (v) arrhythmia from ECG and lastly from (vi) Family history. 2 major criteria or 1 major and 2 minor criteria or 4 minor criteria from different categories must be fulfilled for definite diagnosis.

ARVD should be strongly suspected in a patient with ventricular tachycardia of LBBB morphology in a young adult. Other important ECG findings are inverted T waves in right precordial leads(V1-V3) and epsilon waves (V1-V3). Epsilon wave is an electric signal of depolarization observed at the end of QRS complex and beginning of T wave which is specific for ARVC.[10]

Our patient met 2 major criteria in repolarization and depolarization abnormality categories (TWI in v1 to v3 and Epsilon waves in v1- v3) and 1 major criteria of arrhythmia categories from ECG.

Echocardiographic and angiographic studies showing dilated right ventricle with outpouching in the free wall are useful in making the diagnosis which all goes in favour of our patient. Magnetic resonance imaging may be required for further confirmation showing typical fatty infiltration of right ventricular myocardium which could not be done by our patient. However, CMR is performed in those patients who are suspected to have ARVC in whom other imaging modalities have been non-diagnostic. As all the other investigations were in favour of definitive diagnosis of ARVD, so CMR not carried out in our patient.

ARVD should be differentiated from idiopathic right ventricular outflow tract ventricular tachycardia(RVOT-VT), which has a better prognosis and occurs in the absence of any significant structural heart disease. Other important differentials are athlete's heart, dilated cardiomyopathy, right ventricular infarction, myocarditis which were excluded in our patient.

Treatment options for ARVD are restriction of competitive exercise, pharmacologic therapy, risk stratification and ICD implantation, catheter ablation, cardiac transplantation. The treatment of our patient aimed at the management of ventricular tachycardia and prevention of sudden death.

According to the ESC guideline, for those at high risk of sudden death - ICD is the treatment of choice, also recommended for secondary prevention after an episode of ventricular tachycardia or ventricular fibrillation.

As our patient has recurrent H/O syncope within last 6 months and this time presented with evidence of VT, so he was treated with ICD implantation. The most recent report with a large series of patients with ARVD demonstrated that one-half of patients with ARVC who presented with a malignant or potentially malignant ventricular arrhythmia had an excellent prognosis,[11,12] once treated with an ICD. Although RV dysfunction is present, clinically significant heart failure is seen only in a minority of patients. Progression to heart failure is uncommon and occurs in fewer than 10% of patients. Our patient is still on regular follow up without any feature of heart failure and his ICD is functioning adequately.

Conclusion :

Affected patients could remain asymptomatic for decades making diagnosis of ARVD difficult. We present a young patient with ARVD, a rare clinical condition which was diagnosed by two easily available common non-invasive investigations ECG and ECHO. This case indicates that any young-adult patient who presents

with syncope or palpitations, ARVC should be a differential and further work up with any non-invasive modality should be performed provided that there is a high suspicion of ARVC. Sudden cardiac death is the most feared complication of ARVC; thus, prompt diagnosis and offering ICD placement could save the lives.

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