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LADY WITH A BIG HEART: ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA

KEYWORDS: RBBB, epsilon waves, sotalol, sudden cardiac death, massive cardiomegaly.

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ABSTRACT *Arrhythmogenic right ventricular dysplasia (ARVD) is characterized by fibrofatty replacement of myocardium which affects mainly the right ventricle. While it is one of the rare forms of cardiomyopathies, it is the second most common cause of sudden death of young adults world-wide (after hypertrophic cardiomyopathy-obstructive type), causing up to 20% of sudden cardiac deaths in patients less than 35 years of age. Incidence of ARVD is unknown in the Indian population. A rare case of ARVD is reported. Isolated dilatation of right ventricle with significant trabeculations and pseudosacculations, ECG showing RBBB with epsilon waves and chest x- ray suggestive of gross cardiomegaly make it an interesting case.*

INTRODUCTION:

ARVD, characterized by gradual replacement of the myocardium by fat or fibrosis,usually involves the right ventricular myocardium, but may spread to the left ventricle. Clinical manifestations usually result from ventricular arrhythmias (palpitations, dizziness, syncope or sudden death). Later, right ventricular failure (or left ventricular failure) may also occur.[1] Diagnosis is made using criteria developed by a working group of the European Society of Cardiology.[2]

CASE REPORT:

A 32 year old female presented with breathlessness and anorexia since one month. She had breathlessness, palpitations and syncope since last 20 years. Breathlessness was aggravated on minimal exertion. No history orthopnea/paroxysmal nocturnal dyspnoea. Palpitations were paroxysmal and self limited, lasting 2 to 10 seconds. She had multiple syncopal attacks, once every two months, lasting upto 2 minutes; no obvious trigger. She was hospitalized frequently but never evaluated. All symptoms progressed over time. Her elder brother had similar complaints however details were not available. Both parents were asymptomatic with normal echocardiograms. She had tachycardia, HR 108/min, occasional missed beats (single or in clusters of 3 to 8) alongwith engorged neck veins. Apex beat was in left fifth intercostal space, 2.5 cm lateral to mid-clavicular line. Heart sounds were normal with grade II/VI ejection systolic murmur. Other systems were normal. Chest radiograph revealed massive cardiomegaly (cardio-thoracic ratio of >60%) with an upturned cardiac apex and relatively hyperemic lung fields, bilaterally (Figure 1).

Electrocardiogram showed sinus rhythm and normal axis, tall peaked P waves (p-pulmonale) and T wave inversion in precordial leads from V_1 to V_6. RBBBwas seen, with rsR'in $V_1$and RR' in the remaining precordial leads.Epsilon waves seen as distinct deflections of small amplitude occupied the ST segment in the right precordial leads(Figure 2). The cluster of T wave inversion in precordial leads, RBBB and epsilon waves were highly suggestive of ARVD.

Cardiac MRI with contrast (Figure 3) clinched the diagnosis, showing marked dilatation of right ventricle with significant trabeculations and dyskinesia of anterior and inferior walls, particularly at the apex, with pseudo-sacculations. Right ventricular ejection fraction was 19%. Moderate right atrial dilatation was present alongwith superior and inferior vena caval distension. Left atrium, left ventricle, septae normal; LVEF of 60%.Focal hyperenhancement suggestive of fatty infiltration was seen. Tricuspid valve and atrioventricular groove was normal in position and function, ruling out Ebstein's anomaly. Hematological and biochemical investigations were within normal limits. Patient was treated conservatively with beta blockers (sotalol), antiplatelets andantiepileptics. She was advised implantable cardioverter-defibrillator (ICD) but refused. Symptoms have abated on pharmacotherapy, reports occasional syncope though.

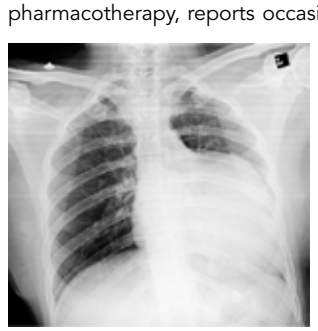

Figure 1- Chest X ray showing massive cardiomegaly

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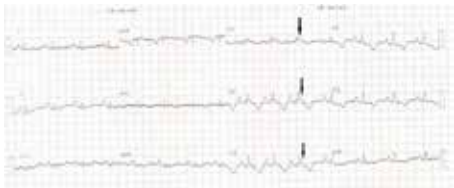

Figure 2- ECG showing Epsilon waves and Right Bundle Branch Block

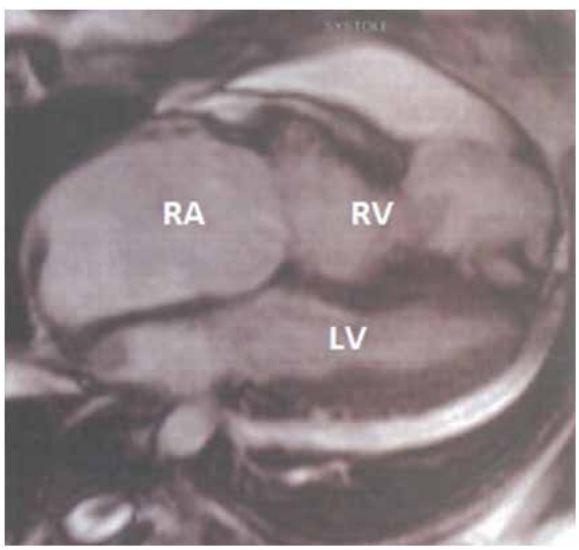

Figure 3- Cardiac MRI with contrast showing right ventricular trabeculationswith pseudosacculations and dilatation.

DISCUSSION:

ARVD is a genetic disease, transmitted from one affected parent to child as an autosomal dominant disorder.[3]It primarily affects the heart muscle in the right ventricle where damaged muscle is replaced by fat or fibro fatty tissue in a spotty or diffuse process. This interferes with the normal electrical activity leading to ventricular premature beats, ventricular tachycardia or ventricular fibrillation, of which the latter two can cause fainting or even sudden death.[1] Men are more frequently affected than women, usually discovered between second and fourth decade[4].

Table 1: Criteria for diagnosis of right ventricular dysplasia by the European Society of Cardiology[2]
I. Global and/or regional dysfunction and structural alterations
MAJOR
Severe dilatation and reduction of right ventricular ejection fraction with no (or only mild) LV 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 walls
MAJOR
Fibrofatty replacement of myocardium on endomyocardial biopsy
III.Repolarisation abnormalities
MINOR
Inverted T waves in right precordial leads (V2 and V3) (people aged more than 12 yr; in absence of right bundle branch block)
IV. Depolarization/conduction abnormalities
MAJOR
Epsilon waves or localised prolongation (> 1 10 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) (ECG, Holter, exercise testing).
Frequent ventricular extrasystoles (more than 1000/24 h) (Holter)
VI. Family history
MAJOR
Familial disease confirmed at necropsy or surgery
MINOR
Familial history of premature sudden death (<35 yr) due to suspected right ventricular dysplasia.
Familial history (clinical diagnosis based on present criteria)

*Detected by echocardiography, angiography, magnetic resonance imaging, or radionuclide scintigraphy. ECG, electrocardiogram; LV, left ventricle.

Diagnosis is made by the presence from different groups **(Table 1)** of: Two major criteria or one major plus two minor criteria or four minor criteria.[2]Right ventricular contrast angiography is the gold standard, but has been superceded by echocardiography andcardiac MRI.[5]Endomyocardialbiopsy is rarely used for diagnosis, because the disease is frequently patchy and thus can be missed. Further, the interventricular septum, which is the commonest biopsy site, may be spared in ARVD.[1]

Treatment varies according to presenting complaints. Patient is frequently asymptomatic until adolescence;alternately, he may have features of heart failure, ventricular arrhythmia, syncope,or even sudden cardiac death (SCD).[6]

In asymptomatic patients or those with non-lethal arrhythmias, beta-blockers, particularly sotalol, and antiarrhythmics like amiodarone are reportedly effective.[1,7]Radiofrequency ablation of an active focus is indicated in patients refractory to pharmacotherapy or in those with an ICD in-situ, who have recurrent ventricular arrhythmias. ICD implantation is considered in hemodynamically unstable ventricular tachycardia. However, implantation in ARVD is complicated owing to thinning of the right ventricular wall and patchy fibrosis.[1,6] Heart transplantation is considered in severely dyspneicpatients.[1]

ECG finding of epsilon waves is a classical diagnostic criterion but can be easily missed. We feelnow physicians will search for an epsilon wave on ECG and ARVD will be added to their list of causes of cardiomegaly and increase their index of suspicion for this rare condition.

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