Arrythmogenic Right Ventricular Dysplasia

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SUMMARY

A 10-year-old well and asymptomatic female was referred for screening of acute right ventricular dilatation (ARVD) as she had an elder brother diagnosed with ARVD whom died of sudden cardiac death. Electrocardiography (ECG), transthoracic echocardiography (TTE) and cardiac magnetic resonance imaging (CMR) were performed. Results of these investigations were suggestive of ARVD. Despite being a rare cardiac disease and largely unrecognised in children and young adult population, ARVD is an important cause of ventricular arrhythmias in this group of patients and is one of the causes of sudden cardiac death (SCD) in this population.

KEY WORDS:
Arrythmogenic Right Ventricular Dysplasia; Right Ventricular Dilatation; Sudden Cardiac Death

INTRODUCTION

ARVD is characterised by myocardial atrophy, fibro fatty replacement, fibrosis and ultimately thinning of the wall with chamber dilatation and aneurysm.[1] It primarily affects right ventricle (RV) which later involve biventricular dilatation. Prevalence of ARVD is approximately 1:5000, affecting men more frequently than women with a ratio of 3:1.2 The mode of inheritance in ARVD is mostly autosomal dominant.[1] The pathogenesis of ARVD has been postulated either from congenital defect, genetics and acquired factors but the evidence thus far has not been conclusive.[2]

Four patterns of clinical presentation have been proposed:3 (1) The concealed phase: Patients are asymptomatic but are at sudden risk of cardiac death from an arrhythmia during episodes of extreme exertion. (2) The overt electrical phase: The most typical presentation, which usually occurs in young patients presenting with severe and symptomatic ventricular arrhythmias and SCD. (3) Diffuse Right Ventricular Dysfunction phase: Patients often present with right-sided heart failure and relatively preserved Left Ventricle function. (4) Biventricular phase: Both ventricles are affected with biventricular pump failure.[3]

In the absence of a clinical gold standard for the diagnosis of ARVD, in 1994, International Task Force Criteria (TFC) was established.[4] The original TFC focused on severe disease with lack of sensitivity for early stage of the disease. The recent modification of TFC included quantitative parameters which improve the diagnosis sensitivity.[4] To make a diagnosis of ARVD requires either two major criteria or one major and two minor criteria or four minor criteria (Table I). ARVD diagnosed during adulthood has recently been shown to have a good prognosis. In contrast, ARVD in childhood has a far more uncertain prognosis and may constitute 30-50% of all sudden death in ARVD.[4]

CASE REPORT

A 10-year-old girl was referred for screening of ARVD as she had strong family history. She was asymptomatic and had no history of previous hospitalisations. Her brother died suddenly at the age of 15 years old having been diagnosed with ARVD. The patient underwent baseline electrocardiography (ECG), cardiac ultrasound and Cardiac Magnetic Resonance (CMR). Her ECG revealed RV strain pattern, with T inversion at V1, V2 and V3 (Figure 1). The cardiac ultrasound showed grossly right ventricular (RV) dilatation and hyperechoic moderator band (Fig 2a, 2b). During the cardiac magnetic resonance (CMR) imaging study, she developed a few episodes of non-sustained ventricular tachycardia (VT) during breath holds. CMR revealed a global decreased contractility of the RV with the RV ejection fraction measuring 24.1%. There are markedly elevated Right Ventricular End Diastolic Volume (EDV) and End Systolic Volume (ESV), while the Left Ventricular End Diastolic Volume, Left Ventricular End Systolic Volume and Left Ventricular Ejection Fraction were still preserved. The RV was dilated and wall markedly thin (Fig 2e, 2f).

Review of her family history, revealed that her brother was diagnosed to have ARVD at the age of 14-year-old. He presented with recurrent symptoms of RV failure and Ventricular Tachycardia. ECHO (Fig 2c, 2d) and CMR (Fig 2g, 2h) showed dilatation and thin wall of Right Ventricle with minimal involvement of left ventricular dilatation. He was eventually started on antiarrhythmic agents and required regular administration of intravenous loading of amiodarone during presentation at the emergency department. While on these antiarrhythmics, ventricular arrhythmias persisted, and Implantable Cardiac Defibrillator (ICD) was implanted at the age of 15-year-old. Unfortunately, three weeks after ICD implantation, he collapsed in school and resuscitative efforts failed to revive him.

Following the strong family history and results of these imaging modalities, a diagnosis of ARVD was made on the young lady. An electrophysiology study was planned during

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Table I: Revised Task Force Criteria for diagnosis of ARVD4

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

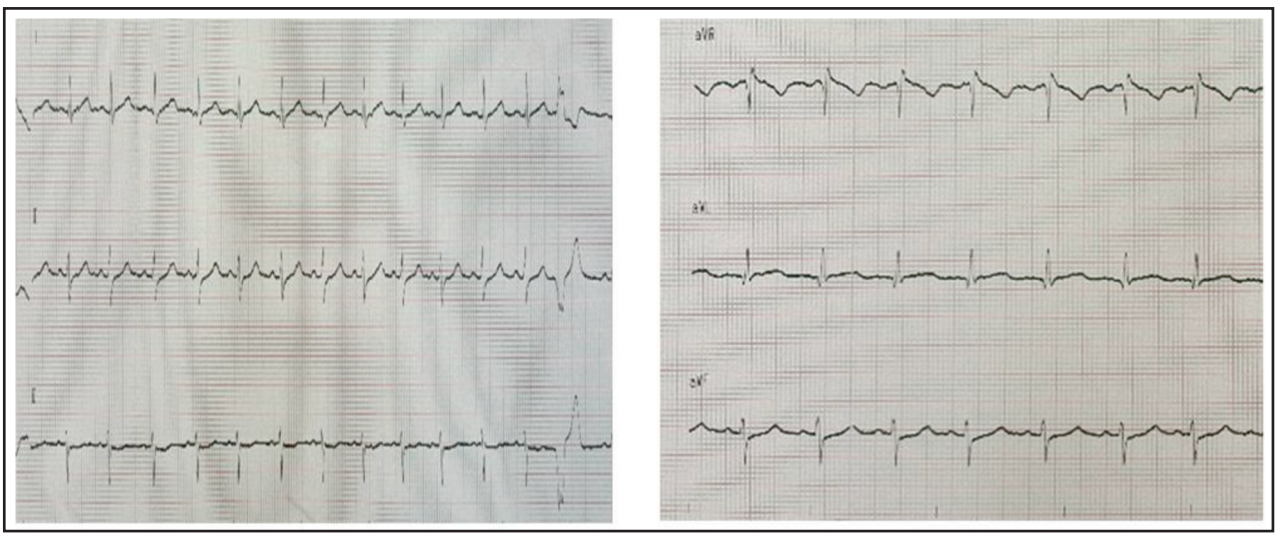

Fig. 1: 12 leads ECG show depolarisation abnormalities (T inversion) at leads V1, V2 and V3.

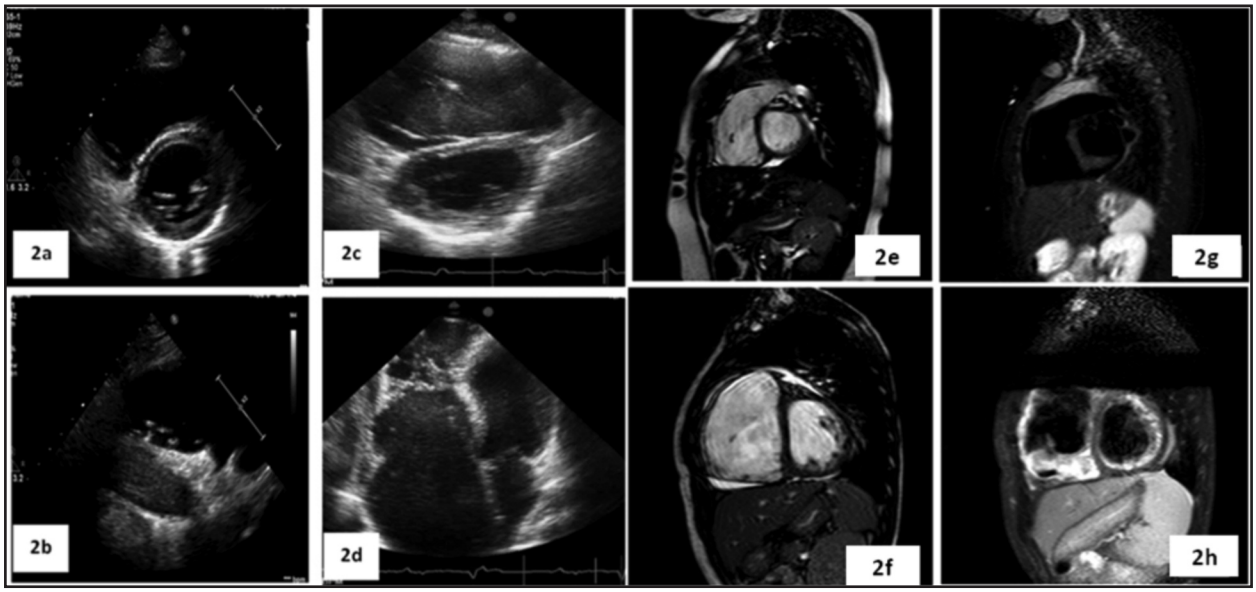

Fig. 2: ECHO (2a-2d) and CMR (2e-2h) findings of patient and her brother. Fig 2a and 2b ECHO findings of the patient showing RV dilatation (2a) and hyperechoic moderator band (2b). Fig 2c and 2d ECHO findings of the patient's brother showing RV dilatation and tethered Tricuspid valve. (2e) Sagittal T1 sequence of the patient with dilated and thinned walled right ventricle (RV). (2f) Sagittal T1 sequence of the patient's brother with dilated and thin walled RV. (2g) Sagittal T2 STIR sequence of patient showing RV wall with fatty infiltration (appears dark on T2 STIR sequence). (2h) Sagittal T2 STIR sequence showing RV wall with fatty infiltration (appears dark on T2 STIR) patient's brother shows RV dilatation and thin wall RV.

her next follow up. She was advised to avoid exercise and any strenuous physical activities and was provided regular close interval follow-up.

DISCUSSION

The clinical presentation of ARVD has been reported to vary among patients. With reference to our patient, she is likely categorised as Concealed phase of clinical manifestation as she is still young and asymptomatic. However sudden premature cardiac death is still a risk that the patient has. Her brother had developed symptoms at the age of 14-year-old and presented with biventricular pump failure. He most likely was at phase four, with biventricular or dilated cardiomyopathy.

The diagnosis of ARVD at its early stages remains a clinical challenge. No single test can be used to establish or exclude ARVD. A keen eye at the ECG for 'epsilon waves' and a detailed echocardiography looking at the right ventricle is often useful. CMR can assist substantially in the diagnosis of anatomical abnormalities and functional disturbances, as well as detecting the presence of fat or fibrous tissue once they occur. Our patient fulfilled one major and two minor criteria from the International Task Force, ARVD classification. She had severe dilatation of Right Ventricle with reduction of RV ejection fraction in cardiac MRI, repolarisation abnormalities; T inversion in V1, V2, V3 and ARVD confirmed in a first-degree relative.

ARVD is a genetic condition, and this condition can go undetected. It is important for the family to be screened so that proper precautions such as danger of exercise and proper management can be initiated early. If screening is positive, then there is a need for close monitoring of symptoms during regular close interval follow up and early intervention if required.

Management of ARVD involves pharmacological therapy and avoidance of exercise. Antiarrhythmic drugs are used but have varying outcomes. Radiofrequency ablation in recurrent or persistent ventricular tachycardia and tachyarrhythmia is also suggested but success rates have been documented to range from 25% to 70%.4 Implantable Cardiac Defibrillator therapy also plays a big role in this case.[4]

In conclusion, despite ARVD being a rare condition and may go undetected for many years, screening of family members with a confirmed and suspected diagnosis of ARVD remains important.

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