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A case report of arrhythmogenic right ventricular dysplasia

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ABSTRACT

Background **Arrhythmogenic right ventricular dysplasia is an autosomal dominant disorder affecting parts of myocardium known as desmosomes, areas on the surface of heart muscle cells which link the cells together. The hallmark feature is fibro-fatty replacement of the right ventricle myocardium characterized by hypokinetic areas with associated arrhythmias originating in the right ventricle.**

Case Presentation **In this report a 42 year old man was admitted at Wuhan union Hospital with the presenting complaints of visual hallucination and difficulty in breathing on exertion, with a family history of sudden death. Clinical and imaging findings are suggestive of Arrhythmogenic right ventricular dysplasia.**

Conclusion **Despite being among the rare cardiac disease, Arrhythmogenic right ventricular dysplasia is an important cause of ventricular arrhythmias in children and young adults, it is also responsible for sudden cardiac death in the young population, making it necessary for this case report.**

Keywords: Arrhythmogenic Right Ventricular Dysplasia, Cardiac MRI

INTRODUCTION

Arrhythmogenic right ventricular dysplasia (ARVD) is an autosomal dominant disorder affecting parts of myocardium known as desmosomes, areas on the surface of heart muscle cells which link the cells together[1]. The hallmark feature is fibro-fatty replacement of the right ventricle myocardium characterized by hypokinetic areas with associated arrhythmias originating in the right ventricle[1]. Despite being among the rare cardiac disease, Arrhythmogenic right ventricular dysplasia is an important cause of ventricular arrhythmias in children and young adults, also responsible for sudden cardiac death in the young population[2].

Individuals with ARVD present with syncope or sudden cardiac death. The remainder frequently presents with palpitations or other symptoms due to right ventricular outflow tract (RVOT) tachycardia (a type of monomophormic ventricular tachycardia) [2]. The symptoms are usually exercised-related. In order to make this diagnosis the following tests are employed; electrocardiogram, Holter monitoring, echocardiography, right ventricular angiography, cardiac magnetic resonance imaging (MRI), and genetic testing.

Arrhythmogenic right ventricular dysplasia may also be related to non-genetic causes such as congenital abnormalities (affecting the right ventricle), viral or inflammatory myocarditis[3]. In the absence of a clinical gold standard for the diagnosis of ARVD, in 1994, the International Task Force Criteria (TFC) was established. The original TFC focused on the diagnosis of overt and severe disease and lacked sensitivity for early forms common among family

members. In a recent modification which included quantitative parameters, it was aimed to improve diagnostic sensitivity[3,4].

There are no pathognomonic features of ARVD. The diagnosis of ARVD is based on a combination of major and minor criteria [4]. To make a diagnosis of ARVD requires either 2 major criteria *or* 1 major and 2 minor criteria *or* 4 minor criteria (**Table 1**).

Table 1 International Task Force Criteria for the diagnosis of ARVD

Major criteria	**Right ventricular dysfunction** - Severe dilatation and reduction of RV ejection fraction with little or no LV impairment - Localized RV aneurysms - Severe segmental dilatation of the RV
	Tissue characterization - Fibrofatty replacement of myocardium on endomyocardial biopsy
	Conduction abnormalities - Epsilon waves in V_1 - V_3 - Localized prolongation (>110 ms) of QRS in V_1 - V_3
	Family history - Familial disease confirmed on autopsy or surgery
Minor Criteria	**Right ventricular dysfunction** - Mild global RV dilatation and/or reduced ejection fraction with normal LV - Mild segmental dilatation of the RV - Regional RV hypokinesis
	Tissue characterization
	Conduction abnormalities - Inverted T waves in V_2 and V_3 in an individual over 12 years old, in the absence of a right bundle branch block (RBBB) - Late potentials on signal averaged EKG - Ventricular tachycardia with a left bundle branch block (LBBB) morphology - Frequent PVCs (> 1000 PVCs / 24 hours)
	Family history - Family history of sudden cardiac death before age 35 - Family history of ARVD

CASE REPORT

We present a case of a 42 year old man who was admitted at Wuhan Union hospital with the presenting complaints of visual hallucination and difficulty in breathing on exertion. He has no history of previous admissions, nor history of drug or food allergy. Moreover, there is family history of sudden death, where his uncle died suddenly. The patient

underwent the following investigations: EKG, holter monitor, echocardiogram and cardiac MRI and their findings were as follows

Electrocardiogram revealed several premature ventricular complexes in several leads, V_3, V_4, V_5 and V_6, there were inverted T-waves observed in leads V_1, V_2 and V_3 also the characteristic Epsilon waves were observed in leads V_2 and V_3. The PVC's and inverted T-waves in lead's V_1, V_2 and V_3 are among the conduction abnormalities in minor criteria, while Epsilon waves is among the conduction abnormality in major criteria.

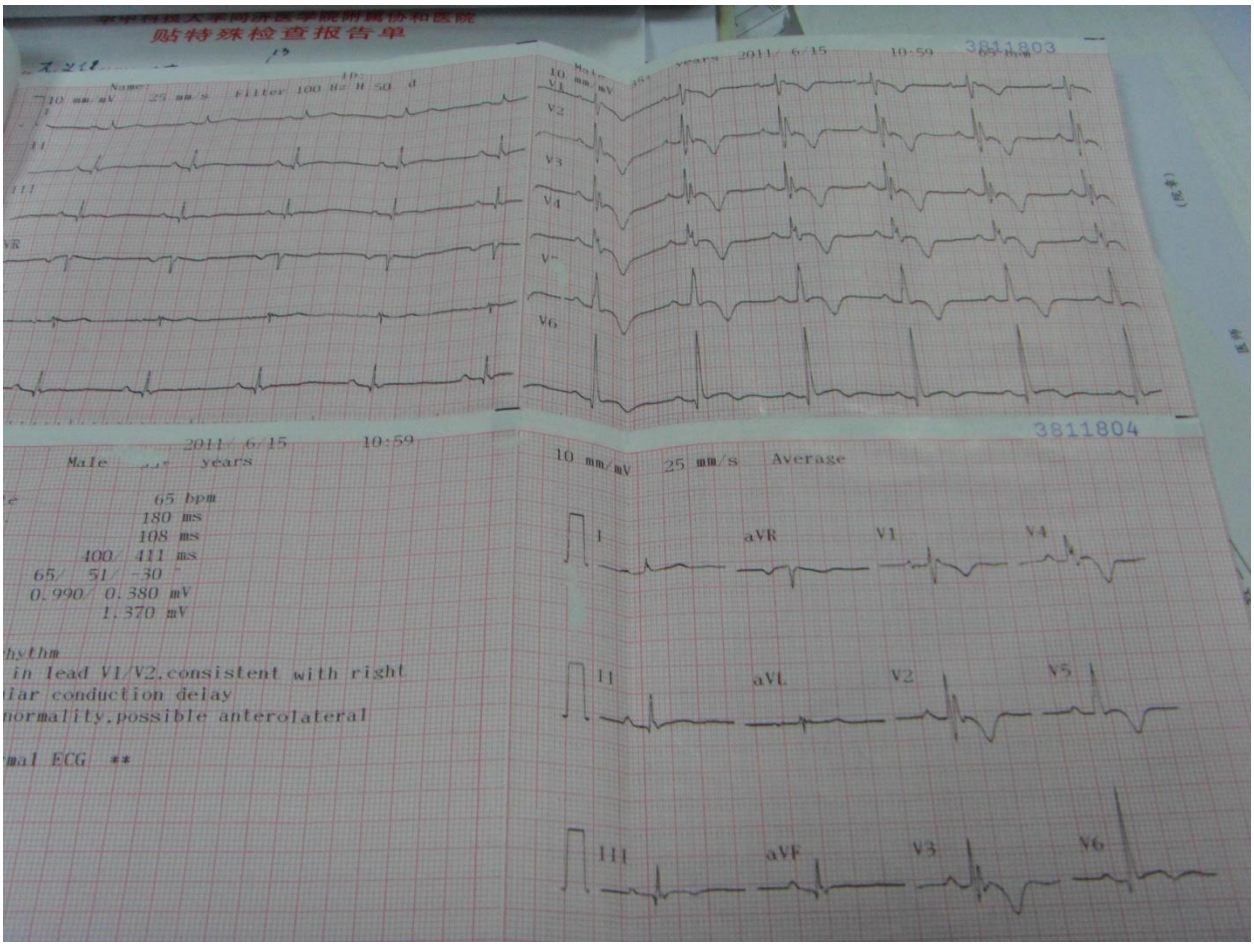

Figure 1 ECG showing Epsilon waves in leads V_2 and V_3, premature ventricular complexes and inverted T-waves in leads V_2-V_6

Holter monitor recorded ventricular tachycardia in leads II, III, aVR, V_2 and V_3 which is among the conduction abnormalities in minor criteria.

Echocardiogram revealed right ventricular enlargement (RVH) and mild tricuspid regurgitation, also reduced right ventricular wall motion was observed.

Cardiac MRI revealed right ventricular hypertrophy, reduced right ventricle wall motion and fatty infiltration on the right ventricle free wall.

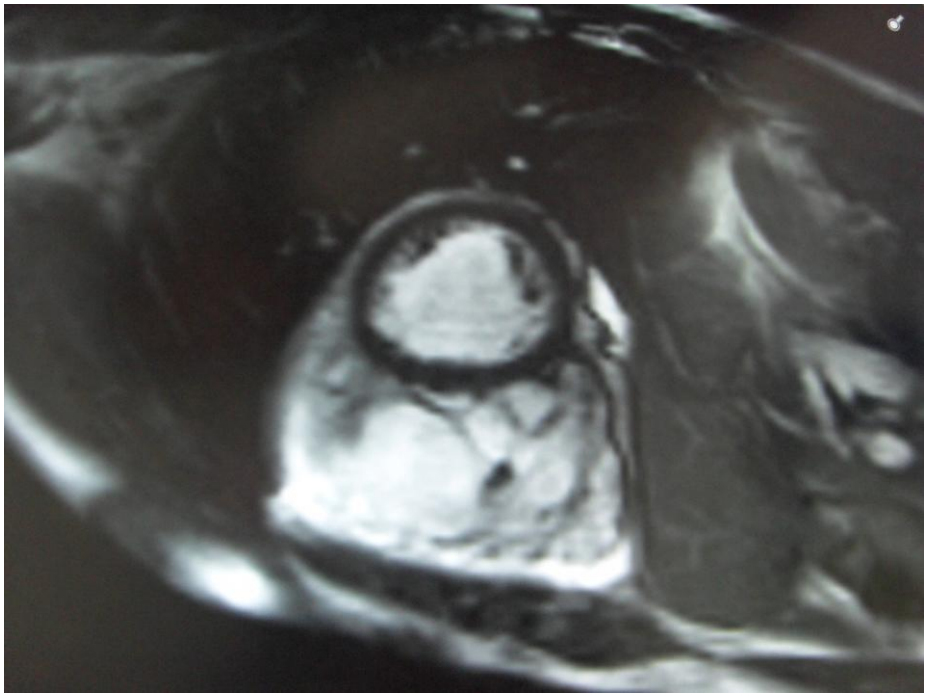

Figure 2 The normal RV has a grayish appearance on this sequence often labeled a bright-blood sequence (the blood filled cavities have no signal) and fat gives off a high signal appearing white. In this patient the RV wall is dilated

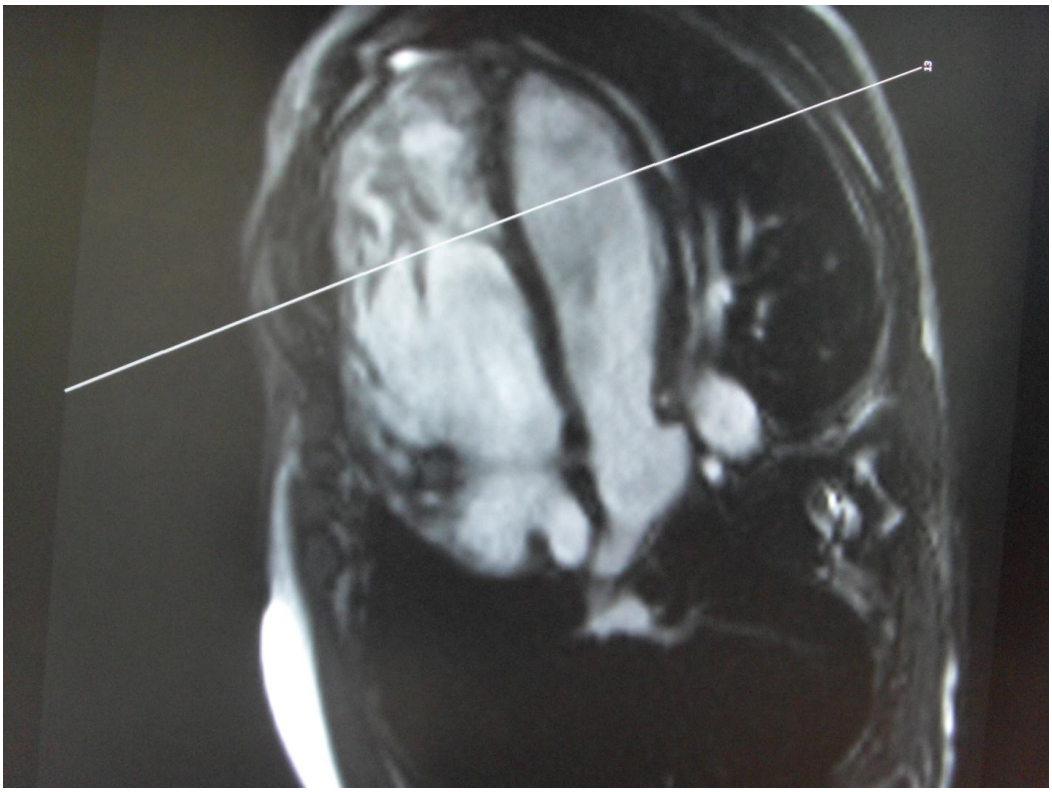

Figure 3 Showing aneurysmatic changes more obvious on the four-chamber view

Following these imaging and clinical findings, a diagnosis of ARVD was reached.

The patient was managed by Propranolol, Perindopril, and Lasix and stayed in hospital for 10 days before being discharged in good condition on the 11th day. He was advised to avoid exercise and any strenuous physical activities and asked to come back for follow-up.

This case report illustrates how ARVD was diagnosed despite how it presented and being a rare disorder.

DISCUSSION

The clinical presentation of ARVD has been reported to vary among patients, with some patients not falling under the established criteria[5]. Majority of patients affected by this disease are diagnosed at the age of 31 years, although some cases of patients being diagnosed at childhood have also been documented[6]. In their study, Dalal *et al*[7] had documented that the common clinical manifestations of ARVD are palpitations, syncopal attacks and premature death[7]. Our patient, aged 42 years, was apparently healthy except for the periodic episodes of dyspoea and visual hallucinations.

There are conflicting reports on the extent of mortality as a result of ARVD[8,9,10]. Hulol *et al*, for example, reported an overall and annual death rate of about 19% and 2%, respectively[10]. Owing to its indolent presentation, this rate is moderately high implying that the risk for a young adult dying from this disease is worthy taken care of.

Traditionally, clinical presentation has been the mainstay of diagnosis of ARVD. Of lately, changes in the function and structure of the right ventricle coupled with abnormalities in the ECG features, and biopsy of the myocardial tissues showing replacement with fibrofatty materials are also very important key to diagnosis[11]. For this particular case, ARVD was diagnosed on the basis of 2 features of major criteria (localized right ventricular aneurysms in cardiac MRI and Epsilon waves in leads V_2 and V_3 in ECG) and 3 features of minor criteria (premature ventricular complexes and inverted T-waves in leads V_2-V_6 in ECG and family history of uncle's sudden death), in order to make a diagnosis of ARVD requires either 2 major criteria or 1 major and 2 minor criteria or 4 minor criteria.

Cardiac magnetic resonance imaging has been reported to be a very sensitive imaging modality in the diagnosis of ARVD[12]. Because of this high sensitivity, MRI has sometimes been reported to cause over-diagnosis of ARVD[13]. Morphological changes seen on MRI include global reduction of myocardial thickening, dilation of RV and derangement in the systolic and diastolic functions[12]. Cardiac MRI in this case showed right ventricular hypertrophy, aneurysmatic changes and reduced right ventricular wall motion.

Of recent, genetic has been implicated as the basis of arrhythmogenic right-ventricular dysplasia. Genetic mutations particularly, of the genes coding for desmosomal junction cells have been theorized to be the major culprit in this condition. Since this condition has a familial predisposition and can go undetected, it is important for first cousins to be screened for the disorder so that proper precautions such as danger of exercise and proper management can be initiated early.

Management of ARVD involves pharmacological therapy and avoidance of exercise. Antiarrhythmics drugs have been previously used with varying outcomes. Some centers use radiofrequency ablation in cases of recurrent or persistent ventricular tachycardia and tachyarrhythmias[2]. Implantable cardioverter defibrillator therapy also plays a big role in this case[2]. However success rates of radiofrequency ablation has been documented to range from 25% to 70%[2].

Review of this presentation and clinical diagnosis of ARVD leads us to the conclusion that despite the disorder being rare and go undetected for some years, and usually detected in routine investigations, it is still important to be aware that it exists and we should be competent in diagnosing and treating it when possible.

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