CASE REPORT

A case of arrhythmogenic right ventricular cardiomyopathy with right ventricle thrombus: A case report

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SUMMARY

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a rare inherited cardiomyopathy characterised by right ventricular dysfunction, ventricular arrhythmias and increased risk of sudden cardiac death. Due to the replacement of myocardium with fibro-fatty and fibrous tissue, patients with ARVC are prone to develop ventricular tachycardia. Histologically, it is often reported as the 'triangle of dysplasia' involving the inflow tract, outflow tract and apex of the right ventricle.2 We describe a 20-years-old patient who collapsed during a futsal match and was subsequently diagnosed to have ARVC with a right ventricular thrombus from cardiac magnetic resonance imaging.

INTRODUCTION

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a rare inherited cardiomyopathy characterised by right ventricular dysfunction, ventricular arrhythmias and increased risk of sudden cardiac death.[1] Due to the replacement of myocardium with fibro-fatty and fibrous tissue, patients with ARVC are prone to develop ventricular tachycardia.[1-2]

CASE PRESENTATION

A 20-years-old gentleman with no known medical illness collapsed during a college futsal match. He was in New York Heart Association (NYHA) classification class 1 with no family history of any cardiac disease or sudden cardiac death (SCD). Cardiopulmonary resuscitation (CPR) was commenced immediately at the scene by a bystander before he was transferred to our centre. He developed multiple episodes of ventricular tachycardia at scene (Figure 1B), in the ambulance and at the emergency department. After successful defibrillation, his electrocardiogram (ECG) showed sinus tachycardia with incomplete right bundle-branch block. Epsilon wave was apparent in leads V2 and V3 (Figure 1A). He was intubated for airway protection and was extubated well after three days in the coronary care unit without any neurological deficits.

His transthoracic echocardiography showed enlargement of right ventricle and right atrium with severe tricuspid valve insufficiency (Figure 1C-F). The left ventricular ejection fraction was 55% by biplane Simpson's method. There was marked hypokinesia of his right ventricle myocardium with focal areas of dyskinesia. Cardiac magnetic resonance imaging (CMRI) (Figure 2) showed a dilated right ventricle (indexed end diastolic volume 128ml/m^2) with mildly impaired right ventricular systolic function (40%), right ventricle dyskinesia and akinesia, right ventricle fibrosis with right ventricular clots.

The left ventricle size and function were normal and there was no evidence of any myocardial infarction or infiltration from the CMRI. He was then started on oral warfarin with subcutaneous enoxaparin 1mg/kg twice daily as bridging therapy for his right ventricle thrombus. He was diagnosed with ARVC as he met the three major criteria of 2010 revised Task Force criteria (Table I) for the diagnosis of ARVC: (I) global or regional dysfunction and structural alterations; (II) depolarisation or conduction abnormalities and (III) arrhythmias. Repeated CMRI after six weeks of adequate anti-coagulation showed complete resolution of the right ventricle clot. Subsequently, he underwent implantation of an implantable cardiac defibrillator (ICD) as secondary prevention for sudden cardiac death induced by ARVC. He was counselled to avoid strenuous physical activities. He was discharge well and a family screening for ARVC was arranged.

DISCUSSION

ARVC is a progressive disease where affected individuals usually do not have any evidence of the disease at birth but starts to manifest symptoms clinically starting from 12 to 13 years of age.[3] Affected patients could remain asymptomatic for decades making this diagnosis difficult. There are reports of sporadic cases of ARVC with no familial involvement. The clinical presentation includes palpitations, syncope, chest pain, dyspnoea and all kinds of ventricular arrhythmias, especially

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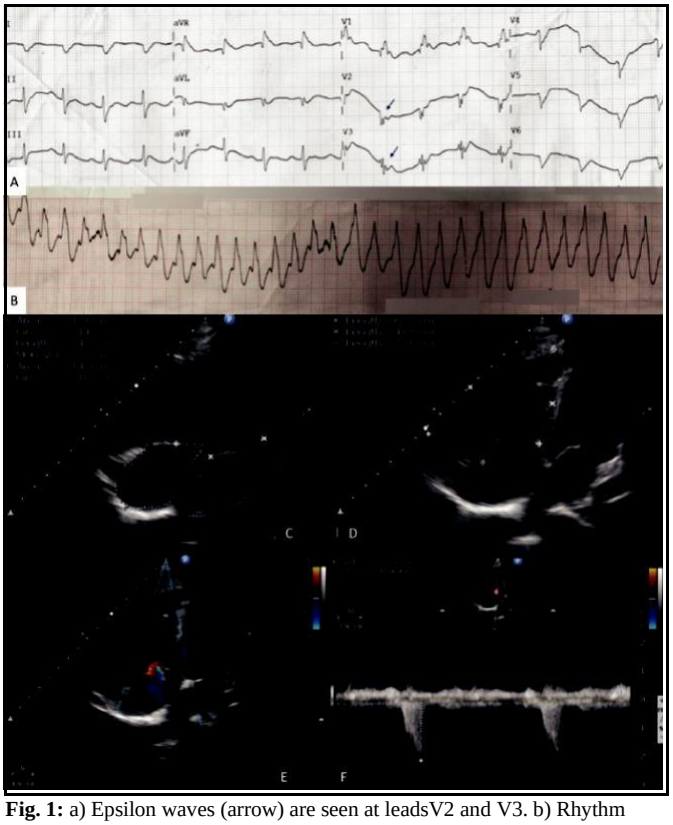

Fig. 1: a) Epsilon waves (arrow) are seen at leadsV2 and V3. b) Rhythm strip showing ventricular tachycardia. c) Dilated right atrium measuring 13 cm^2. d) Dilated right ventricle e) Tricuspid regurgitation. f) Modified view of the right heart showing tricuspid regurgitation with velocity of 2.15 m/sec.

recurrent episodes of ventricular tachycardia. ARVC is caused by mutations in a variety of genes encoding for desmosomal proteins.[3,4] The commonest inheritance described is autosomal dominant but autosomal recessive inheritance had been described as part of the cardiocutaneous syndrome with hyperkeratosis of soles and palms.[3] Multiple genes mutations tend to present earlier with more frequent ventricular arrhythmias, heart failure and left ventricle failure.[4] The goal of therapy is to prevent sudden cardiac death and slow down the progression of disease. Patients with confirmed ARVC should be advised against strenuous physical exercises as competitive sports has a strong link with development of ventricular arrhythmias and heart failure. An implantable cardiac defibrillator (ICD) is indicated in patients who have been resuscitated from sudden cardiac arrest or experienced sustained ventricular tachycardia.5 Prophylactic beta blocker is recommended in Patients with ARVC. Radiofrequency ablation is recommended to reduce arrhythmia burden and ICD shocks. Device therapy such as cardiac resynchronisation therapy (CRT) or cardiac resynchronisation therapy defibrillator (CRTD) should be considered in patients with severe heart failure. Cardiac transplant is the only therapeutic option for patients with severe advanced and refractory disease (Biventricular failure refractory to optimal medical therapy and uncontrolled arrhythmias). Screening for first degree relatives with history, physical examination, electrocardiography, and echocardiogram are recommended.

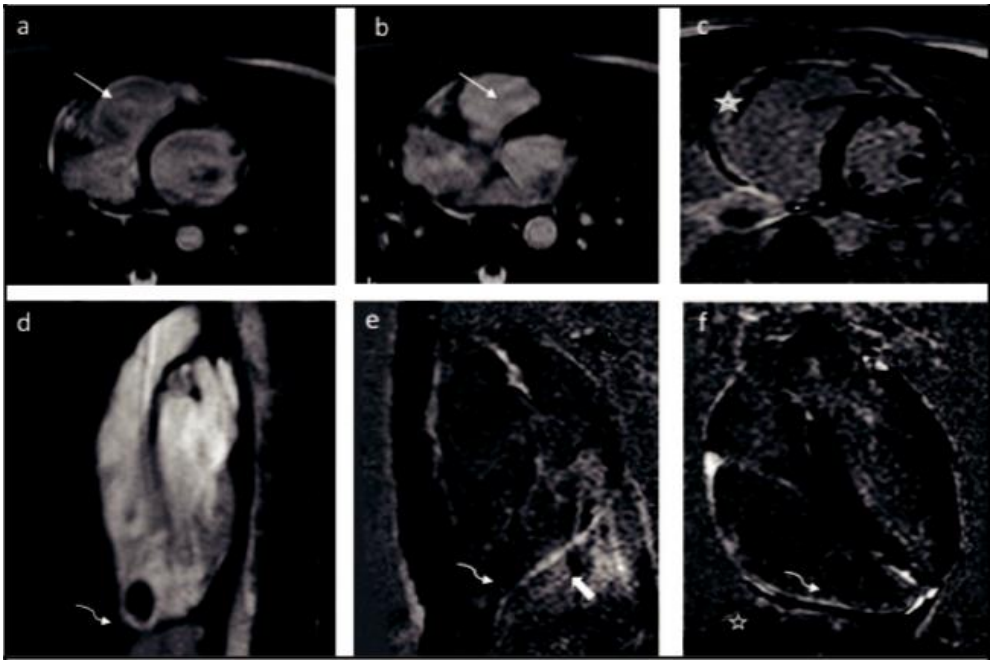

Fig. 2: (Above) (a-f): Akinesia of the right ventricular outflow tract (RVOT) area in diastole (a) and in systole (b) of transaxial images (thin white arrow). Right ventricular wall enhancement on late gadolinium enhancement (LGE) of free wall (white star) seen in short axis at mid cavity (c) and horizontal long axis (HLA) (f) as well as inferior wall (filled white arrow) of right inflow outflow (RVIO) view (e). Right ventricular thrombus at the apex (curve white arrow) seen in early gadolinium RVOT (d), RVIO view LGE (e) and HLA LGE (f).

(a) Transaxial cine diastole (b) Transaxial cine systole (c) Short axis at mid cavity late gadolinium enhancement (LGE) (d) RVOT early gadolinium (e) Right inflow outflow LGE (f) Horizontal long axis LGE.

Approximately one third of first-degree relatives of a proband diagnosed with ARVC will develop ARVC. Cardiac MRI and genetic testing can assist in the diagnosis of ARVC.

Right ventricle thrombus is rarely seen in ARVC. Wlodarska et al. reported an annual incidence of 0.5 thromboembolic events per 100 patients in 126 confirmed ARVC patients over a mean follow-up of 99 months.[5] The reported incidence of thromboembolic events include pulmonary embolism from right ventricular clots, right ventricular thrombus and right ventricular outflow tract thrombosis. Thrombus formation in ARVC can occur without any trigger. Scar formation after successful radiofrequency ablation may increase the risk of thrombus formation. Low molecular weight heparin with oral anticoagulation therapy can be used to treat right ventricle thrombus

CONCLUSION

In conclusion, ARVC is a rare condition and could be easily missed. Young adults with a history of cardiac arrest should prompt clinicians to rule out ARVC. Cardiac MRI has become a non-invasive imaging tool in the diagnosis of ARVC with high sensitivity and specificity. ARVC is a slow progressive disease. Most of the patients continue to remain asymptomatic for years. Those who develop arrhythmias, sudden cardiac death and heart failure need pharmacological therapy (beta blocker) or device therapy such as implantable cardioverter defibrillator, cardiac resynchronisation therapy or cardiac resynchronisation therapy defibrillator. Cardiac transplant is the ultimate option for those who have exhausted all other therapeutic options.

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