Case study

HIGH DEGREE ATRIOVENTRICULAR BLOCK: First Presentation of Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia

ABSTRACT

Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) is an uncommon inherited cardiac disease characterized by progressive right ventricular (RV) dysfunction due to fibro-fatty replacement of the myocardium and associated with high risk of ventricular arrhythmias and sudden cardiac death. ARVC/D has a predominantly autosomal dominant inheritance, although recessive forms associated with a cutaneous phenotype, such as Naxos disease and Carvajal syndrome, are also observed. The golden standard for diagnosing ARVC/D is an endomyocardial biopsy demonstrating fibro-fatty replacement. Our case concerns a young adult patient with rare bradyarrhythmias (type high degree atrioventricular block) associated with signs and symptoms of right heart failure who was diagnosed with arrhythmogenic right ventricular cardiomyopathy ARVC with pejorative prognosis shortly died after admission.

KEYWORDS: High degree atrioventricular block, arrhythmogenic right ventricular cardiomyopathy/dysplasia, Task force criteria

INTRODUCTION

Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) is characterized by progressive replacement of myocardial cells by fibro-fatty tissue, initially only in the right ventricle, but it may extent to the left ventricle as well [1]. The diagnosis is made with the Task Force Criteria [1]. The estimated prevalence is 1:5,000 individuals [2]. Genetic alterations underlying ARVC are often mutations in one of the cardiac desmosome genes [3]. These mutations are known for their phenotype diversity within families [4]. The golden standard for diagnosing ARVC/D is an endomyocardial biopsy demonstrating fibro-fatty replacement [5]. ARVC is associated with ventricular tachyarrhythmias giving rise to syncope or sudden cardiac death. However, we present a young adult patient with bradyarrhythmias (type high degree atrioventricular block) who was diagnosed with arrhythmogenic right ventricular cardiomyopathy ARVC shortly died after admission.

CLINICAL PRESENTATION

A 51-year-old patient with no particular medical history, admitted at the cardiology emergency department for a repetitive episode of syncope associated with aggravating effort dyspnea. At admission, patient was dyspneic stage 3 of the NYHA scale, with no chest pain or palpitation, associated with episodes of syncope. Blood pressure (BP) at 110/60 mmHg, and heart rate (HR) of 40 beats per minute (bpm) associated with signs of right heart failure (oedema of the lower limbs, Turgor of the jugular veins, effort hepatlagia).

The electrocardiogram (ECG) findings showed a complete atrioventricular block with an escape rhythm of 40 bpm (Figure 1) associated with a completely right bundle block (RBB).

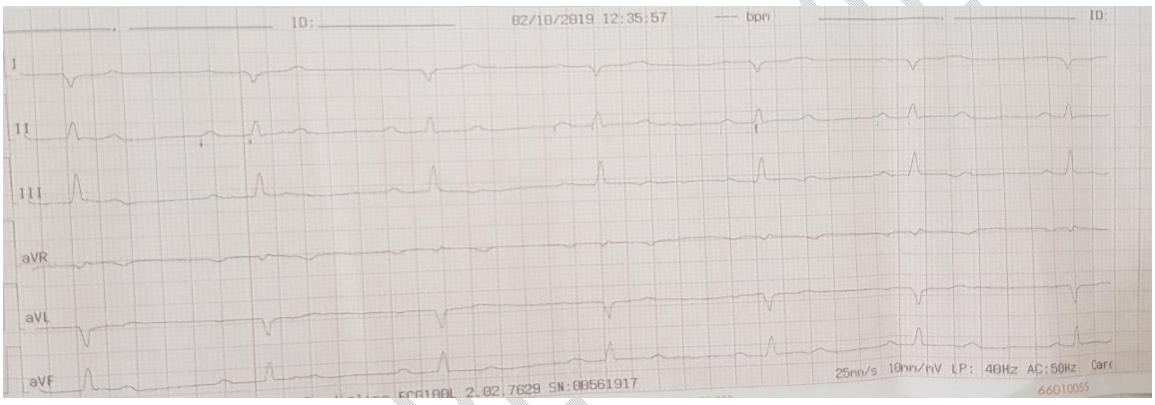

Figure 1: Electrocardiogram (ECG): 3rd degree atrioventricular bock, escape rhythm 40bpm associated with RBB.

Transthoracic echocardiography (TTE) performed on the patient showed a non-dilated left ventricle (LV) with a LVEF of 30% measured by bipolar Simpson method, and a very dilated right ventricle (Basal diameter of the right ventricle = 51mm). A dyskinetic free wall of the right ventricle was observed associated with an aneurysm, and multiple trabeculations, in severe dysfunction. We also note a functional laminar tricuspid regurgitation (TR) due to dilation of the tricuspid ring and coaptation defect (Figure 2).

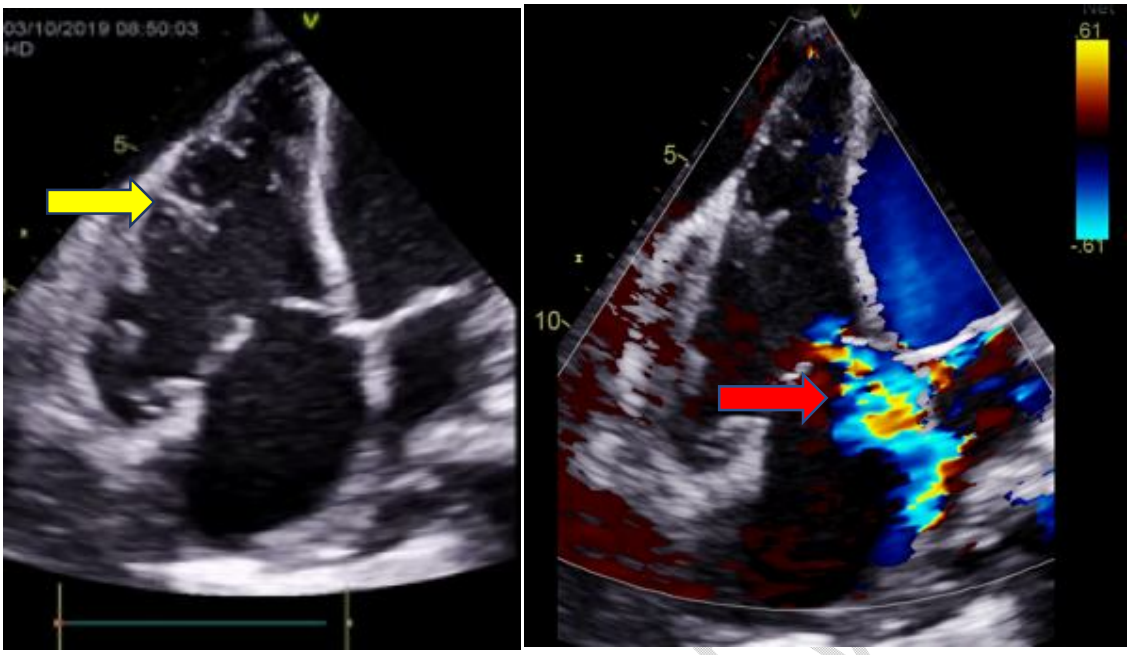

Figure 2a: Transthoracic echocardiography (TTE): 4 chambers apical view: showing very dilated right ventricle with an aneurysm, and multiple trabeculations, in severe dysfunction (yellow arrow).

Figure 2b: TTE: Doppler: 4 chambers apical view: functional laminar tricuspid regurgitation (TR) [red arrow] and medium abundance pericardial effusion.

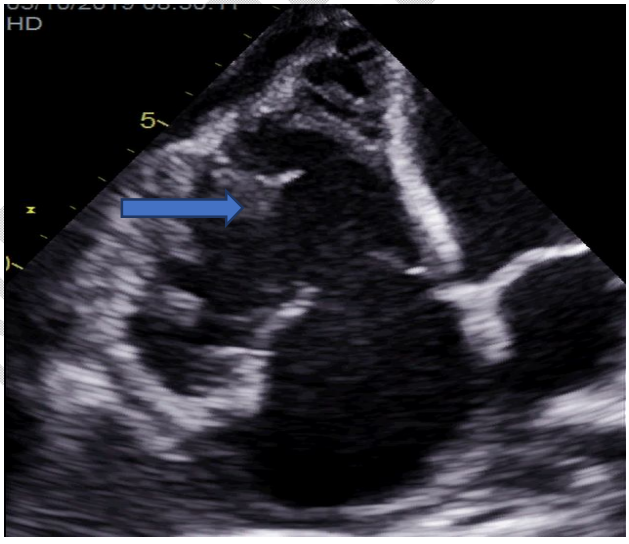

Figure 2c: TTE: 4 chambers apical view: presence of spontaneous contrast and thrombus intra trabeculae (blue arrow) with dilated left atrium.

The biological assessment showed an increased level of NT Pro BNP (NT Pro BNP:1000ng/l) in the blood with a slightly increased in the level of heart enzyme troponin (Highly sensitive troponin=70ng/l).

Faced with this clinical outcome, a cardia magnetic resonance imaging (cMRI) was performed urgently, objectifying a dilated and severe right ventricle (RV) dysfunction. A global RV hypokinesia with an indexed right ventricle tele diastolic volume (RV DTV) of 192ml/m^2 and a 21% ejection fraction of the right ventricle (EFRV) Figure 3.

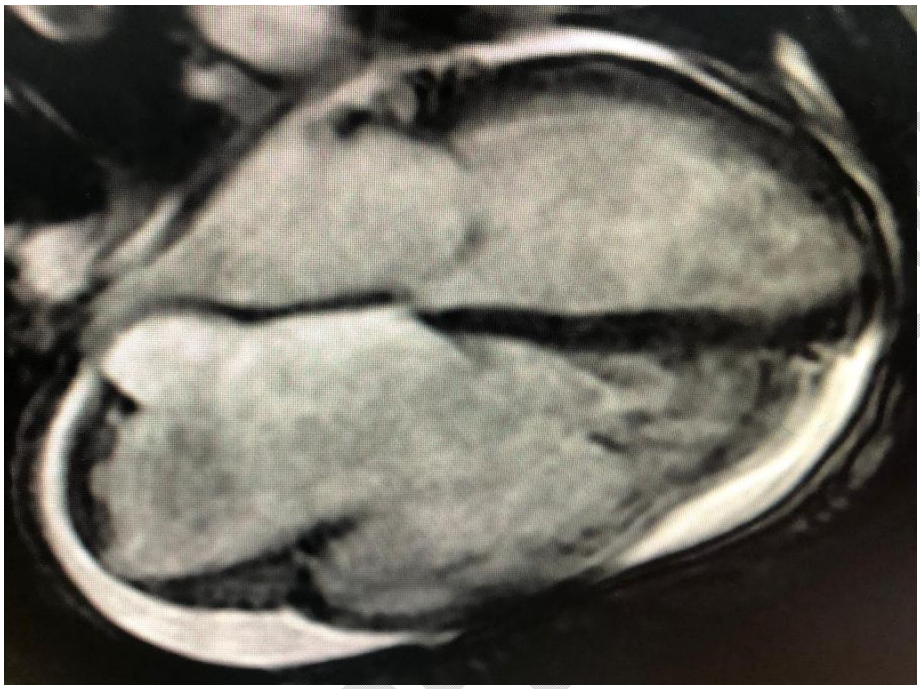

Figure 3: Cardiac MRI: cross section showing a dilated right ventricle with an impaired ejection fraction of 21%.

cMRI also identified multiple aneurysms and trabeculations located at the level of the RV which interest its free wall (Figure 4) and the infundibulum of pulmonary artery. The right atrium (RA) was also dilated (Index volume of the right atrium= 90ml/m^2), with the presence of a thrombus at the bottom level of the RA measuring 14mm x 10mm in diameter. Hypertrophy of the RV wall (figure 5).

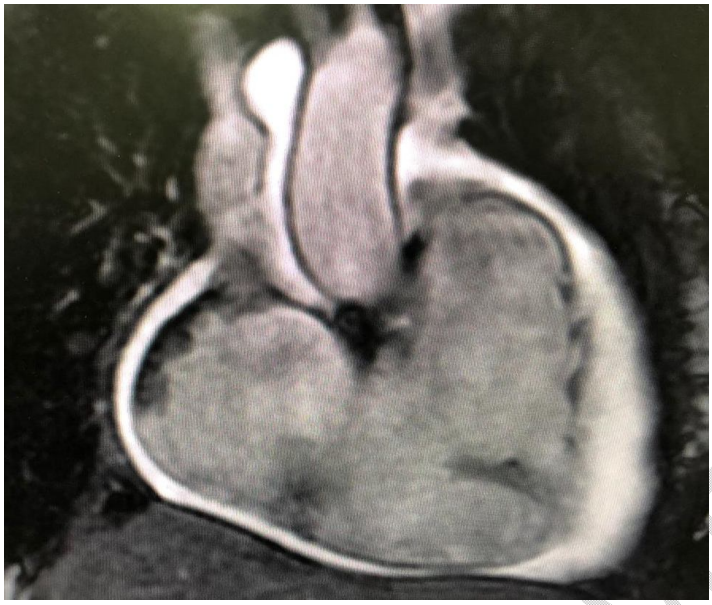

Figure 4: Cardiac MRI: T2 sequency: Hypertrophy of the right ventricle with multiple aneurysms of its free wall, presence of medium pericardial effusion.

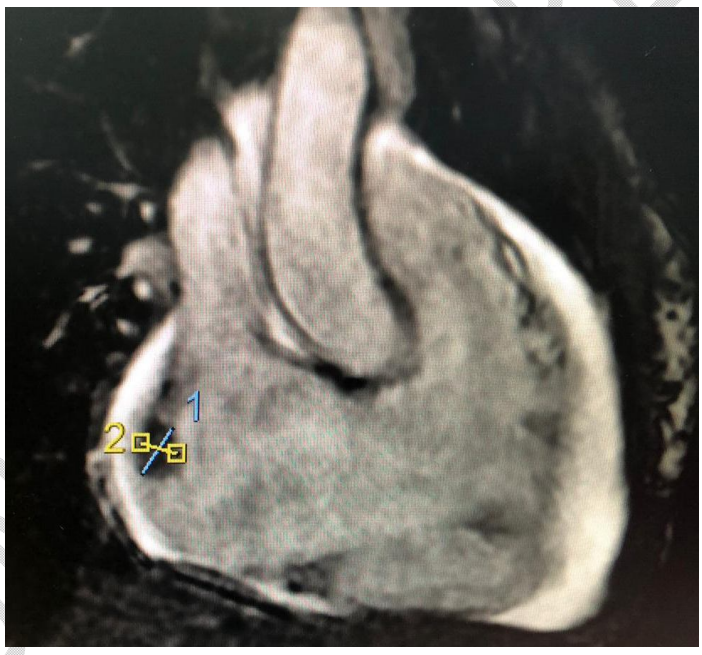

Figure 5: Cardiac MRI: Thrombus at the bottom of the right atrium associated with a dilated right ventricle without late enhancement.

The LV was non-dilated, non-hypertrophied (indexed LV tele diastolic volume=62ml/m^2 and tele systolic volume=45.3ml/m^2) with a severe impaired global function. Pericardial effusion of medium abundance in circumference associated medium bilateral pleural effusion with no remarkable fibrose tissue.

This cardiac finding confirmed the diagnosis of arrhythmogenic right ventricular cardiomyopathy.

The patient was subsequently rushed to cath lab for coronary artery angiography and pacemaker, but the evolution was rapidly fatal, progressing to death after an extreme bradyarrhythmia and asystole.

DISCUSSION

In this case report, we describe an adult patient with a symptomatic high degree atrioventricular conduction block before ARVC became overt. He presented with a coincidentally found third degree atrioventricular block type after episodes of syncope, which the transthoracic echocardiography (TTE) suspected an ARVC confirmed by cardiac MRI.

Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) is an uncommon inherited cardiac disease characterized by progressive right ventricular (RV) dysfunction due to fibro-fatty replacement of the myocardium and associated with high risk of ventricular arrhythmias and sudden cardiac death [6]. ARVC/D has a predominantly autosomal dominant inheritance, although recessive forms associated with a cutaneous phenotype, such as Naxos disease and Carvajal syndrome, are also observed [7]. A cross-sectional study aimed to assess the prevalence and clinical significance of bradyarrhythmias in ARVC was carried out by Erpang Liang et al. from May 1995 to December 2017 [8] showed that bradyarrhythmias were commonly seen in ARVC, and intraventricular conduction block (ICB) was the most common type. In our case AV block was the first clinical presentation.

Classically, ARVC/D usually presents between the second and fourth decades of life with syncope, symptomatic arrhythmias, or sudden cardiac death (SCD) [9]. In our case, patient consulted for repetitive episodes of syncope and heart failure (HF) symptoms who later died before few hours before cardiac intervention. These symptoms were all combined in our patients who had a pejorative prognosis. A study reports that HF hospitalization has a significant relation with malignant clinical course in ARVC patients, and first-degree AVB at baseline is strongly associated with HF hospitalization [10].

There is no single diagnostic test for ARVC/D. The diagnosis is made based on major and minor clinical, electrical, and imaging criteria that have been devised by expert consensus of the Task Force Criteria (TFC) originally proposed in 1994 and further revised in 2010 [1,6]. The diagnosis was certain with major and

minor criteria confirming the diagnostic. TTE et cardiac MRI play an important role in diagnosis confirmation as well as deferential diagnosis of ARVC/D [10].

The American College of Cardiology, the American Heart Association and the European Society and Cardiology recommended ICD implantation for the prevention of SCD events [11]. Risk stratification and indication to ICD implantation in ARVC/D has been proposed by an international task force consensus statement [12]. Individuals who present with congestive heart failure are managed with diuretics and angiotensin-converting enzyme (ACE) inhibitors or aldosterone inhibitors, with heart transplantation considered in terminal stages of the disease. Anticoagulation may be used in ARVC/D patients with large, hypokinetic RV and slow blood flow because of the risk of thrombosis [13]. Therapy with beta blockers, sotalol or amiodarone may be effective in suppressing ventricular arrhythmias and possibly in preventing sudden cardiac death [14]. Management of family members of patients with ARVC/D is complex due to the incomplete penetrance and variable expressivity nature of the disease [15]. In our case screening of family members was difficult as patient was from another region.
Atrioventricular conduction abnormalities have only rarely been described in patients already diagnosed with ARVC [16]. Our patient presented both symptoms of bradyarrhythmias and heart failure is rare entity as heart failure signs a rarely neglected.

CONCLUSION

ARVC/D is an inherited disease characterized by fibro-fatty replacement of the right ventricular myocardium, which significantly increases the risk of paroxysmal ventricular arrhythmias and SCD. Diagnosis is based on the 2010 modified Task Force criteria, requiring clinical and family history, electrocardiography, and imaging. Diagnosis may be confirmed by endomyocardial biopsy. This case report confirms ARVC/D can appear as conduction disorder and heart failure increase the risk of mortality.

ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

CONSENT

As per international standard or university standard, patient(s) written consent has been collected and preserved by the author(s).

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