Annals of Case Reports & Reviews

Case Report

doi: 10.39127/2574-5747/ACRR:1000118.
Ech-chenbouli A, et al. Annal Cas Rep Rev: ACRR-118.

A Case of Arrythmogenic Right Ventricular Cardiomyopathy Presenting as An Inferior STEMI

Ech-chenbouli Amine(MD)*, Benmallem Othmane(MD), Choukrallah Hamza(MD), ElAdaoui Abdelmajid(MD), Asklou Abdesamad(MD), Quechar Zakaria (MD), Aqli Elmehdi (MD), Arous Salim (MD), Benouna Mohamed Elghali(MD), Habbal Rachida(MD)

Department of Cardiology Ibn Rochd University Hospital Casablanca Morocco

***Corresponding author:** Ech-chenbouli Amine (MD), Rue des hopitaux, Casablanca 20380, Morocco. Tel: +212663006950; E-mail address: ech.amine@gmail.com

Citation: Ech-chenbouli A, Benmallem O, Choukrallah H, Eladaoui A, Asklou A, et al. (2020) A Case of Arrythmogenic Right Ventricular Cardiomyopathy Presenting as An Inferior STEMI. Annal Cas Rep Rev: ACRR-118.

Received Date: 11 May 2020; **Accepted Date:** 15 May 2020; **Published Date:** 21 May 2020

Abstract

Arrythmogenic right ventricular cardiomyopathy is considered to be the most frequent arrhythmogenic cardiomyopathy. It is a rare condition in the general population with an estimated prevalence of 1:5000.
We report the case of a 65 y.o man who was admitted to our emergency department for syncope and chest pain with a typical EKG of inferior STEMI who turned out to be an arrythmogenic right ventricular cardiomyopathy. Surprisingly the coronary angiogram was in the limit of normal and right ventriculorography showed a typical aspect of stack of plates suggestive f arrythmogenic right ventricular cardiomyopathy. On echocardiography we objectified a dilated right ventricle with an anterior wall hypokinesia, cardiac MRI showed in addition of right ventricular dilatation and segmental akinesia a fatty infiltration on late gadolinium enhancement.

Keywords: *Arrythmogenic right ventricular cardiomyopathy, STEMI, Cardiac MRI*

Learning objective

Arrythmogenic right ventricular cardiomyopathy is a rare condition that can be responsible for severe ventricular arrhythmias, physicians should be aware of atypical presentations. The diagnostic is made of multiple criteria and bundle of arguments based on echocardiography, cardiac magnetic resonance and genetic testing.

Introduction

ARVC is considered to be the most frequent arrhythmogenic cardiomyopathy. It is a rare condition in the general population with an estimated prevalence of 1:5000 [1]. When first described it was termed Arrhythmogenic Right Ventricular Dysplasia as it was considered as a congenital dysgenesis of myocardium of the right ventricle [2]. However, it has been proved to be an inherited cardiomyopathy caused predominantly by mutations in genes encoding desmosomal proteins (i.e. plakophillin2, plakoglobin, desmoplakin, desmoglein2, and desmocollin2) [3]. The defining pathological process in ARVC consists of myocyte atrophy and degeneration with subsequent fibrous and fatty replacement of predominantly RV myocardium [4]. ARVC usually presents during the second decade of life mostly with signs or symptoms related to ventricular arrhythmias that typically originate from the right ventricle. The disease is often progressive and associated with hot phases characterized by increased RV arrhythmias and new ECG changes [5, 6]. End disease stage is characterized by development of RV dilatation and failure. Through the case of a man presenting a STEMI which turned out to be an ARVC, we will discuss the diagnostic and therapeutic peculiarities of this entity in our context which is beginning to be better understood.

Case report

A 65 y.o man was admitted to our emergency department for syncope and chest pain. the symptomatology occurred 6 hours before admission. The patient had no personal medical history. Clinical examination showed normal blood pressure of 100/80mmgh, heart rate was 80 bpm; there was no heart or lung murmurs on auscultation, EKG showed a typical ST segment elevation in the inferior and right leads transthoracic echocardiogram revealed RV dilatation (RVOT=34mm, RVIT=40mm) with anterior wall hypokinesia and bulging of RVOT and increased trabeculation (Figure 1).

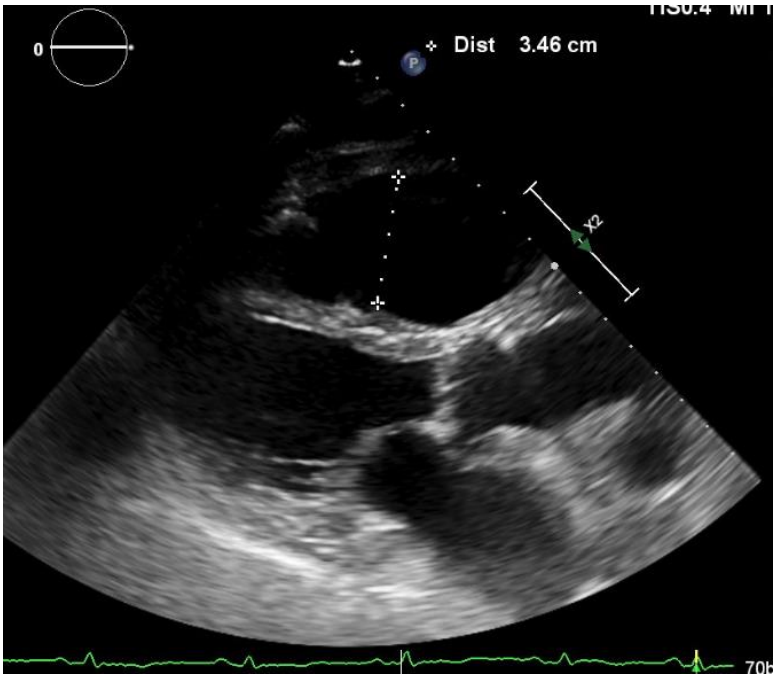

Figure 1: Echocardiography parasternal long axis viw showing Right Ventricular outflow tract dilatation at 34.6mm.

The LV appeared within normal limits. The patient had urgent Coronary angiography wish surprisingly was normal; a right ventriculography showed typical aspect of stack of plates" (figure 2) the diagnosis was corrected and a ARVC was strongly suspected. Blood test revealed a mild increase in CPK, CPKMB, Myoglobin and troponin (220 U/L, 57 U/L, 813 ng/mL and 46.1 ng/L respectively).

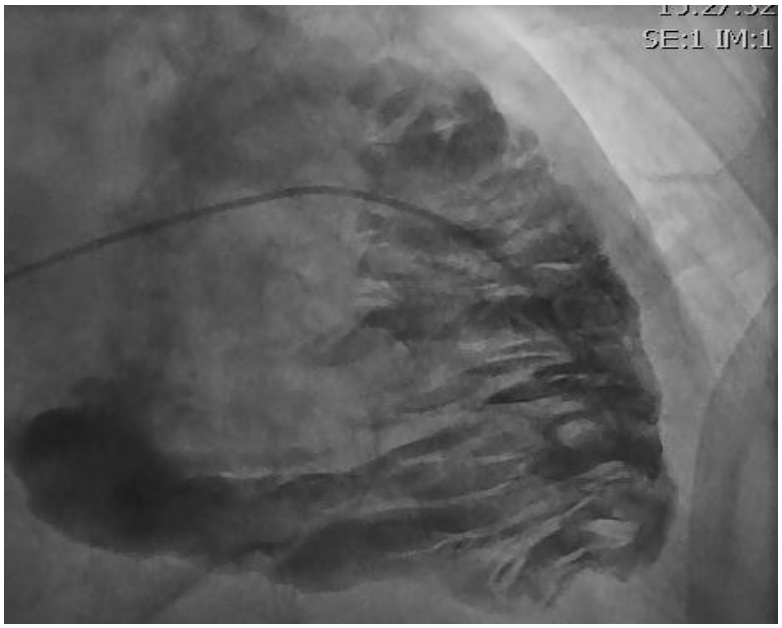

Figure 2: Right ventriculography showing a typical aspect of stack of plates suggesting Arrythmogenic right ventricular cardiomyopathy.

Hemoglobin, white blood cells, inflammation indexes, renal and liver function, and d-dimer were unremarkable. 2 days after his hospitalization we received the information of the death of his 32 years old son in undetermined circumstances. 24H holter ECG monitoring showed frequent isolated PVC's with LBBB morphology and inferior axis. An exercise test was performed and showed no VT or VF. MRI study showed RV dilatation with global wall motion dysfunction, RV fat infiltration and late gadolinium enhancement documenting the presence of severe RV dilatation and dysfunction (RV end diastolic volume 147 mL/m2; RVEF=30%) RV free wall thinning with diffuse hypokinesia and multiple dyskinetic areas (bulging) at RV outflow tract. Late enhancement in the diaphragmatic RV wall was presented in localized apical and lateral wall thinning and akinesia, fibrofatty infiltration of interventricular septum and lateral wall (Figures 3).

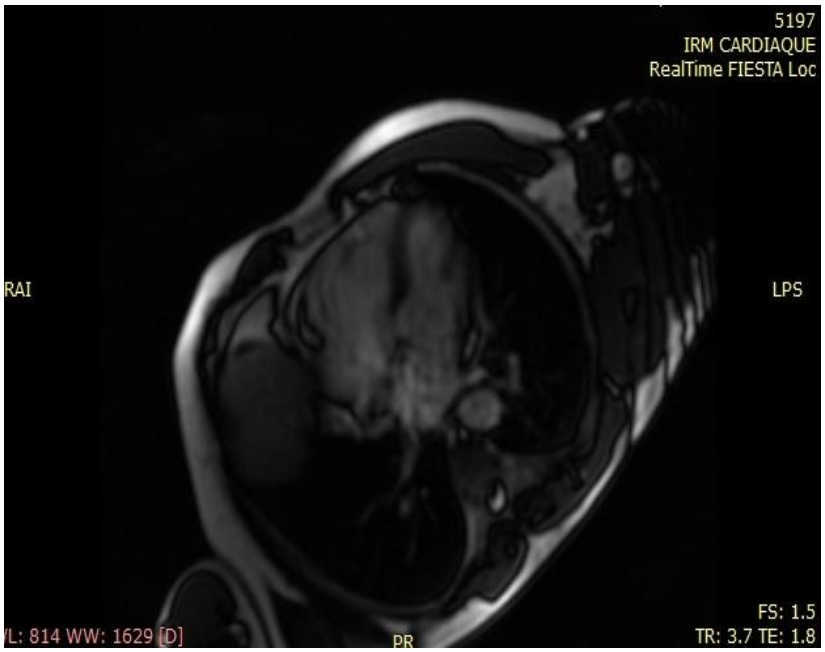

Figure 3: Cardiac 4 chamber MRI showing the RV dilatation with fatty infiltration.

These findings led us to the diagnosis of arrhythmogenic right ventricular cardiomyopathy (ARVC) without LV involvement. Bisoprolol 5 mg daily was started accordingly. And patient was programmed for defibrillator.

Discussion

ARVC is a cardiomyopathy firstly described in the 1970s characterized by a progressive replacement of the myocytes by various degrees of fibro-adipose tissue involving particularly the areas between the anterior part of the pulmonary infundibulum, the apex, and the infero-posterior wall of the RV. ARVC, also called right ventricular dysplasia (RVD) and right ventricular cardiomyopathy (RVC), was uniformly defined and classified as 1 of the 5 primary cardiomyopathies in 1995. ARVC occurs in both sexes at any age, but sudden deaths tend to occur in adults between 15 and 45 years, with a mean age of about 30 years. [7,8] The male predisposition might be associated with the disease genes and androgen hormone. Tabib et al. [7] examined 200 cases of sudden death owing to ARVC and found that the mean age was 36 years (range, 5–64 years), and 108 (54%) cases of these were males.

Clinical presentation of ARVC typically involves palpitations, syncope, ventricular tachycardia, congestive heart failure, and SCD. in our case the clinical presentation was in the form of a STEMI which is to our knowledge never was described in the literature. The exact pathogenesis of ARVC is still unclear, but this involves a genetic factor (9). Currently, the known genetic mutations associated with ARVC include PG, PKP2, DSP, DSC2, DSG2, TGFb3, TMEM43, RYR2, TTN, and JUP [10]. Most of the patients reported had a family history and genetic tendency. In this case the patient's son had a sudden death while his father was hospitalized; we could not investigate more but we suspected a sudden cardiac death on an ARVC ground probably caused by the stress of the father's hospitalization. Currently, molecular and genetic testing of ARVC are not a routine diagnostic procedure. However, genetic testing is recommended to be a useful in dealing with the suspected ARVC cases and consequently identifies the cardiac risk of living family members.

T-wave inversions in the right precordial leads are present in up to 87% of adult patients with ARVC due to RV dilatation. It should be sought in family screening as it might be the earliest indication of disease development in asymptomatic family members (11). However, they can be found in other contexts, such as in some young athletes, in ischemic cardiomyopathy, or during acute pulmonary embolism. Incomplete or complete right bundle branch block is also described and manifested in our patient. Epsilon wave (electrical potentials of small amplitude that occur at the end of the QRS complex and at the beginning of the ST segment in the right precordial leads) represents areas of delayed activation frequently localized in the RV as a consequence of fibrous and/or fibro-fatty replacement of myocardium and it is considered a major criterion for the diagnosis of ARVC [1,3,4]. Interestingly, in our patient he had a st segment elevation on right and inferior leads epsilon wave was revealed only after defibrillation, likely as a result of a shock-induced massive increase of diastolic intracellular calcium concentration followed by activation of a transient sodium inward ion current [4,5].

The presence of segmental wall motion abnormalities (regional akinesia, dyskinesia, or dyssynchrony), combined with RV dilatation and/or dysfunction, are required for diagnosis, and were both present in our case. RV outflow tract dilatation and the reduction of RV fractional area are also part of the known criteria for the diagnosis of ARVC, but unfortunately, they are usually seen in advanced stages. RV hypertrophic trabeculation is part of the structural abnormalities of the disease and was clearly documented in our case. In addition, longitudinal strain derived from speckle tracking could be a sensitive tool for assessing regional and global myocardial function, particularly in early stages of the disease [12,13].

CMR helped to confirm the diagnostic hypothesis and to characterize the extent of the disease, showing. The diagnosis of ARVC remains challenging due to the absence of specific unique diagnostic criteria, its variable expressivity, and incomplete penetrance. In addition, although endomyocardial biopsies may help in the differential diagnosis (myocarditis, sarcoidosis and rarely Uhl's disease) they often generate nonspecific findings due to the patchy nature of the disease. Furthermore, safety concerns limit their use [14,15]. We all agreed that in our case this procedure was unnecessary and potentially harmful.

Genetic testing has reached dramatic improvement for the diagnosis of ARVC. To date, 16 genes have been associated with the ARVC phenotype, mostly those encoding desmosomal proteins [16]. We did not perform genetic screening since ECG, echo and CMR findings in our opinion were highly suggestive if not pathognomonic for the disease. However, genetic testing appears to be crucial to rule out ARVC in subjects in whom ECG, echocardiogram or CMR did not provide clear results and for screening purposes in parents or relatives of patients with diagnosed ARVC.

Conclusion

Our case shows that the presentation of ARVC can be very atypical; in front of echocardiographic signs of ARVC a particular importance should be given to Cardiac MRI since it's a very good tool to objectify right ventricular measures, function and fatty infiltration. Genetic exploration if available must be done especially for the family screening.

Ethical approval

Informed consent was obtained from the patient's family for publication of this case report.

Declaration of interests

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

References

1. C. Basso, D. Corrado, F.I.Marcus, A.Nava, G. Thiene, Arrhythmogenic right ventricular cardiomyopathy, Lancet 373 (2009) 1289–1300.
2. F.I. Marcus, G.H. Fontaine, G. Guiraudon, R. Frank, J.L. Laurenceau, C. Malergue, et al., Right ventricular dysplasia: a report of 24 adult cases, Circulation 65 (1982) 384–398.
3. K. Pilichou, G. Thiene, B. Bauce, I. Rigato, E. Lazzarini, F. Migliore, et al., Arrhythmogenic cardiomyopathy, Orphanet J. Rare Dis. 11 (33) (2016).
4. G. Thiene, A.Nava, D. Corrado, L. Rossi, N. Pennelli, Right ventricular cardiomyopathy and sudden death in young people, N. Engl. J. Med. 318 (1988) 129–133
5. A. Protonotarios, A. Anastasakis, D.B. Panagiotakos, L. Antoniades, P. Syrris, A. Vouliotis, et al., Arrhythmic risk assessment in genotyped families with arrhythmogenic right ventricular cardiomyopathy, Europace 18 (2016) 610–616.
6. A. Bhonsale, J.A. Groeneweg, C.A. James, D. Dooijes, C. Tichnell, J.D. Jongbloed, et al., Impact of genotype on clinical course in arrhythmogenic right ventricular dysplasia/cardiomyopathy-associated mutation carriers, Eur. Heart J. 36 (2015) 847–855.
7. Tabib A, Loire R, Chalabreysse L, et al. Circumstances of death and gross and microscopic observations in a series of 200 cases of sudden death associated with arrhythmogenic right ventricular cardiomyopathy and/or dysplasia. Circulation 2003;108:3000–5.
8. Ye D, Edwards WD, Rizkalla W. Sudden unexpected death in a 31-yearold man caused by arrhythmogenic right ventricular cardiomyopathy. Arch Pathol Lab Med 2005;129:1330–3.
9. Basso C, Bauce B, Corrado D, et al. Pathophysiology of arrhythmogenic cardiomyopathy. Nat Rev Cardiol 2011;9:223–33.
10. Sato T, Nishio H, Suzuki K. Identification of arrhythmogenic right ventricular cardiomyopathy-causing gene mutations in young sudden unexpected death autopsy cases. J Forensic Sci 2015;60:457–61.
11. F. Migliore, A. Zorzi, P. Michieli, M. Perazzolo Marra, M. Siciliano, I. Rigato, et al., Prevalence of cardiomyopathy in Italian asymptomatic children with electrocardiographic T-wave inversion at preparticipation screening, Circulation 125 (2012) 529–538
12. Prakasa KR, Wang J, Tandri H, et al. Utility of tissue Doppler and strain echocardiography in arrhythmogenic right ventricular dysplasia/cardiomyopathy. Am J Cardiol 2007;100:507-12.
13. Te Riele AS, Tandri H, Sanborn DM, Bluemke DA. Noninvasive multimodality imaging in ARVD/C. J Am Coll Cardiol Img 2015;8:597-611
14. Pieroni M, Dello Russo A, Marzo F, et al. High prevalence of myocarditis mimicking arrhythmogenic right ventricular cardiomyopathy differential diagnosis by electroanatomic mapping- guided endomyocardial biopsy. J Am Coll Cardiol 2009; 53:681-9.
15. Ott P, Marcus FI, Sobonya RE, et al. Cardiac sarcoidosis masquerading as right ventricular dysplasia. Pacing Clin Electrophysiol 2003;26:1498-503.
16. Awad MM, Calkins H, Judge DP. Mechanisms of disease: molecular genetics of arrhythmogenic right ventricular dysplasia/ cardiomyopathy. NatClin Pract Cardiovasc Med 2008;5: 258-67.