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Arrhythmogenic Right Ventricular Dysplasia: a case report and review of the literature

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ABSTRACT

Arrhythmogenic right ventricular dysplasia (ARVD) is a right ventricular cardiomyopathy characterized by partial or total replacement of normal myocardium by fatty or fibrofatty tissue. This disease affects the right ventricle. The left ventricle is rarely involved. The exact prevalence is unknown. It represents, after hypertrophic cardiomyopathy, the leading cause of sudden cardiac death in young patients, especially athletes. She has a male predominance. Sudden death by ventricular tachycardia or ventricular fibrillation can be the inaugural event. The diagnostic criteria developed in 2010 are very useful in the diagnosis of ARVD, especially in mild forms. The three imaging cited, and therefore useful diagnostic modalities are echocardiography, magnetic resonance imaging (MRI) and conventional angiography RV. The therapeutic management of patients with ARVD is based on antiarrhythmics especially beta-blockers and catheter ablation. Implantable cardioverter defibrillator ICD may be indicated for primary or secondary prevention of sudden death and improves the survival of patients with ARVD at high risk.

INTRODUCTION

Recent advances in molecular biology have isolated a group of diseases affecting the right ventricular myocardium called cardiomyopathy of the right ventricle. Among the latter, there is ARVD. This is a rare disease involving episodes of ventricular tachycardia with a left bundle branch block pattern and fibrofatty degeneration of the right ventricle. This is an important cause of sudden cardiac death in young patients, especially athletes. Its prevalence is estimated between 1/2500 and 1/5000.

MATERIALS AND METHOD

We report the case of a 46 year old male with no history for disease or cardiovascular risk factor. He was admitted to the emergency for palpitations associated with two well tolerated episodes of ventricular tachycardia with a left bundle branch block pattern (Figure 1), reduced by amiodarone. The cardiovascular examination outside the critical episode was normal. The electrocardiogram objectified an Epsilon wave and abnormal repolarization in the right precordial leads (Figure 2). The Holter monitoring objectified a premature ventricular to left type of delay in stage Lown IV.

Echocardiography showed right ventricular dilatation. Coronary angiography was normal (Figure 3). The right ventricular angiography confirmed ARVD (Figure 4: 1-trabecular hypertrophy, 2-bulge below tricuspid). Heart MRI was not available in our training. Programmed Ventricular Stimulation has triggered ventricular tachycardia (Figure 5). An implantable cardioverter defibrillator (ICD) was implanted with a good clinical outcome.

Through this observation, we discuss the pathophysiological, clinical, diagnostic and therapeutic bases of ARVD.

RESULTS AND DISCUSSION

ARVD is a right ventricular cardiomyopathy characterized by fatty infiltration of the myocardium with persistent surviving myocardial fibers surrounded by a fine fibrosis (1).
Giovanni Maria Lancisi first described in 1736 a family disease, affecting the right ventricle and complicating sudden death. The disease has been described as such for the first time in 1978 by a Parisian team, naming its current name. The first series was published in 1982. Electrocardiographic abnormalities such epsilon wave, were described in 1984 (2).
ARVD affects mostly men and young subjects. The male / female ratio was 2.7 / 1 (3). It is estimated that the incidence of ARVD in the general population is 1 to 2 cases per 10,000 (4). This incidence is higher in some populations of the Mediterranean and the United States and can reach 44 cases per 10,000 (5). Although rare, this is an important cause of sudden cardiac death (SCD) in young patients, especially athletes. ARVD accounts for about 2.2% of MCS in adults in the United Kingdom (6), 5% of MCS in young people under 35 years in the United States (7) and 25% of deaths related to efforts in the Venetian region of Italy (8). The disease rarely occurs before adolescence and affected subjects are often men with active lifestyles (9).
It is basically an autosomal dominant disorder. Seven genes have been identified that are associated with ARVD: plakoglobin, desmoplakin, plakophilin-2, desmoglein-2, desmocollin-2, transforming growth factor beta-3 (TGFβ3) and TMEM43. Its phenotypic expression is highly variable. It also appears that environmental factors such as intensive sports or contraction of a viral infection with myocarditis may influence the clinical expression of the disease (10,11).
Histologically, the involution of the right ventricle is segmental, moving from the epicardium to the endocardium. It determines a fatty form and fibrofatty form. Fatty form will initially affect the infundibulum of the pulmonary artery and the free wall of the right ventricle, without parietal thinning of the affected areas and without left ventricular involvement. There is no inflammatory infiltration. Degeneration of myocytes is evident in half the cases. In the fibrofatty form, reaching seat preferentially at the base of the right ventricle, causing a thinning of these areas and can interest the left ventricle. In this form the histological examination of the myocardium can found inflammatory cells (12,13). The myocardial atrophy is caused by lesions and apoptosis. This leads to a thinning of the free wall of the right ventricle. This transformation interferes with the transmission of electrical impulses, explaining electrocardiogram abnormalities and arrhythmias. The disorder of inter-cell junctions may also explain the whole (14).
The clinical presentation of ARVD is very varied. It is a progressive disease with symptoms of stress, relapsing, ranging from asymptomatic to all varieties of ventricular arrhythmias. The most dramatic is the sudden death from ventricular fibrillation, which may be the first manifestation (15).
From easy diagnosis if severe, it is often difficult in mild forms and warrants to associate a set of clinical, electrical and imaging positive criteria. In 1994, a consensus of expert led to propose diagnostic criteria (16). These have been reviewed and published in 2010 (10). Pathological criteria, ECG, characteristics of ventricular arrhythmias and family history are shown in detail. The

diagnostic criteria for imaging are also defined. The three imaging modalities mentioned, and therefore useful in the diagnosis, are echocardiography, MRI and conventional RV angiography.

Electrocardiographic (ECG) changes were seen in up to 90% of patients with ARVD (17). These changes reflect abnormal depolarization and repolarization located on the right precordial electrodes. The most common ECG change is the inversion of the T wave in the right precordial electrode (electrodes V1-V3). Although this event may be present together with a right bundle branch block, congenital heart disease, in African Americans or children under the age of twelve years (two normal variants). His presence, especially when it extends beyond the electrode V3, should raise the suspicion of ARVD. Furthermore, the degree of inversion of the T wave may be correlated to the degree of dilatation of the RV (18). Epsilon waves also represent a delay in the right ventricular conduction. These small waves occur at the end of the QRS complex. They represent the potential of low amplitude late and fragmented in the terminal portion of the QRS complex also due to fibrofatty tissue islets in normal myocardium. Although highly specific, this event is only present in about 30% of patients with ARVD. High amplification ECG provides more detailed information on late potentials and characterization. T wave inversion and Epsilon wave represent two major ECG criteria in the diagnostic of ARVD. Another major criterion is represented by nonsustained or sustained ventricular tachycardia of left bundle-branch morphology with superior axis (19).

Echocardiographic abnormalities in ARVD are mainly located in a triangle (triangle of dysplasia), joining the pulmonary infundibulum, the tip of RV and the base. Echocardiography should be particularly attentive to dilation of the infundibulum and the existence of morphological and functional abnormalities of the RV wall. These data are interesting when rhythm abnormalities associated. New echocardiographic criteria for 2010 are as follows:

Major criteria: Regional RV akinesia, dyskinesia, or aneurysm and 1 of the following (end diastole): parasternal long-axis RV Outflow Tract (OT) ≥32 mm (≥19 mm/m²), ≥36 mm (≥21 mm/m²) or fractional area change ≤33%.

Minor criteria: Regional RV akinesia or dyskinesia and 1 of the following (end diastole): parasternal long-axis RVOT ≥29 to <32 mm (≥16 to <19 mm/m²), parasternal short-axis RVOT ≥32 to <36 mm (≥18 to <21 mm/m²) or fractional area change >33% to ≤40%.

A normal transthoracic echocardiography does not formally exclude the diagnosis. This justifies using other types of imaging in particular MRI.

MRI is well adapted imaging technique for the diagnosis of dysplasia RV. The image quality is often excellent. The cine MR imaging allows a single analysis of segmental wall motion of the RV. This criterion is the major criterion for diagnosis. MRI allows quantification of volumes and RVEF, representing another major criterion. T1 weighted imaging diagnostic aid by highlighting intramyocardial fat, and delayed enhancement imaging by the detection of fibrosis. New MRI criteria in 2010 are:

Major criteria: Regional RV akinesia or dyskinesia or dyssynchronous RV contraction and 1 of the following: Ratio of RV end-diastolic volume ≥110 mL/m² (male) or ≥100 mL/m² (female) or RV ejection fraction ≤40%.

Minor criteria: Regional RV akinesia or dyskinesia or dyssynchronous RV contraction and 1 of the following: Ratio of RV end-diastolic volume ≥100 to <110 mL/m² (male) or ≥90 to<100 mL/m² (female) or RV ejection fraction >40% to ≤45%.

RV angiography remains for many the gold standard. But it is limited to a few specialized centers. It detects akinetic or dyskinetic areas, blowing the infundibulum, tip or sub-tricuspid regions (triangle of dysplasia), and measure RVEF and volume RV (thresholds: RVEF <35%, diastolic volume RV > 110 ml / m²).

Our patient had major diagnostic criteria: clinical, electrical, échocardiographic and angiographic.

Programmed ventricular stimulation takes an interesting role in the investigation of patients at risk of arrhythmias and sudden death. Roguin showed in his series that inducibility of sustained VT was an independent predictor Factor of sudden death (20).
Improperly tolerated hemodynamically tachycardia, left ventricular impairment, sport, less than 35 years old, myocarditis, uncontrolled treatments are all risk factors that could guide the decision to placement a defibrillator.
The therapeutic management of patients with ARVD is not guided by specific recommendations. However, there are options for establishing a strategy to protect patients according to their risk of sudden death. A hybrid strategy that combines the use of antiarrhythmic drugs and radiofrequency ablation can reduce the occurrence of ventricular arrhythmias, but cannot stop the progression of the disease and the risk of sudden death (21). Programmed ventricular stimulation can guide the therapeutic management especially when the diagnosis of ARVD is done without spontaneous rhythm disorders. It serves primarily to judge the effectiveness of an anti-arrhythmic treatment in case of initial inducibility without treatment and adapt it. The importance of effective treatment and verified by an electrophysiological study was shown in the 90s by a German team (22). When the VT is no longer triggered under pharmacological treatment by the methods of programmed stimulation, there is no sudden death with a mean follow up of 3 years, despite recurrent nonfatal VT a few months to a few years later. The authors emphasize the particular interest of sotalol in high doses, 320-480 mg/d. The combination of amiodarone 400 mg/d and β blockers in small doses was effective on refractory ventricular tachycardia (23). The use of amiodarone, however, must make to consider the possibility of adverse effects in the long term, when it comes to treating young patients. Implantable cardioverter defibrillator ICD may be indicated for primary or secondary prevention of sudden death and improves the survival of patients with ARVD at high risk. The primary prevention studies are rare. The ICD in secondary prevention is very useful and should be reserved for patients at high risk (24). The recommendations of the American scientific societies for genetic heart disease, indicate the ICD in ARVD in the case of sudden death recovered (class I, level of evidence B), and if there is a sustained VT (class I, level of evidence C) (25). Our patient had an opening VT with a left bundle branch block pattern and programmed ventricular stimulation performed under treatment triggered a sustained VT. If the above treatments fail, ventriculotomy and transplantation are the management options. Right ventriculotomy is performed by disconnecting the right ventricle from the rest of the heart. Heart transplantation remains a final option for patients with bilateral ventricular involvement and uncontrollable hemodynamic compromise (26). In addition to these treatments, it is necessary to convince patients to stop the sport, and offer genetic counseling to detect the related persons.

CONCLUSION

Since the first description of ARVD in 1978, great progress was noted in the understanding of the pathogenesis, histopathology, but also in the diagnosis of this disease. This diagnosis was probably facilitated by the use of clinical, electrical and imaging criteria without favoring one over the other depending on the level of expertise of each center. Nevertheless, it remains a challenge for forms where the diagnostic criteria are incomplete.Therefore, lifelong monitoring of patients with ARVD by the primary care physician-cardiologist team is recommended.

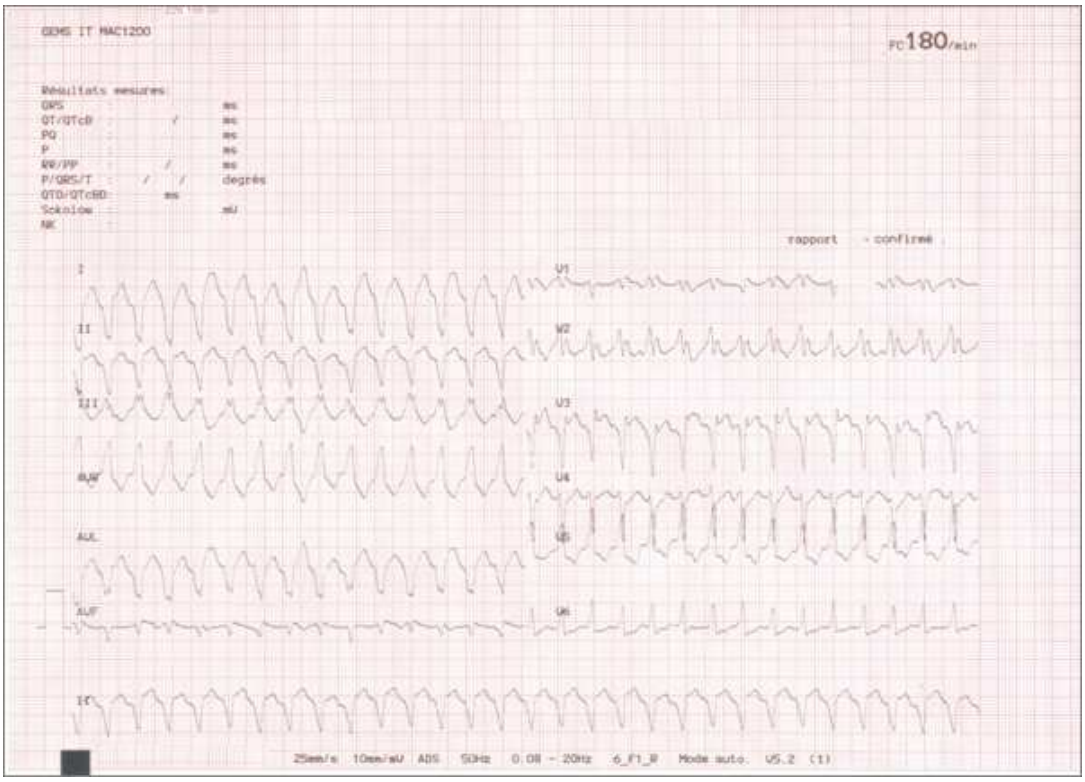

Figure 1: ECG showing ventricular tachycardia with a left bundle branch block pattern.

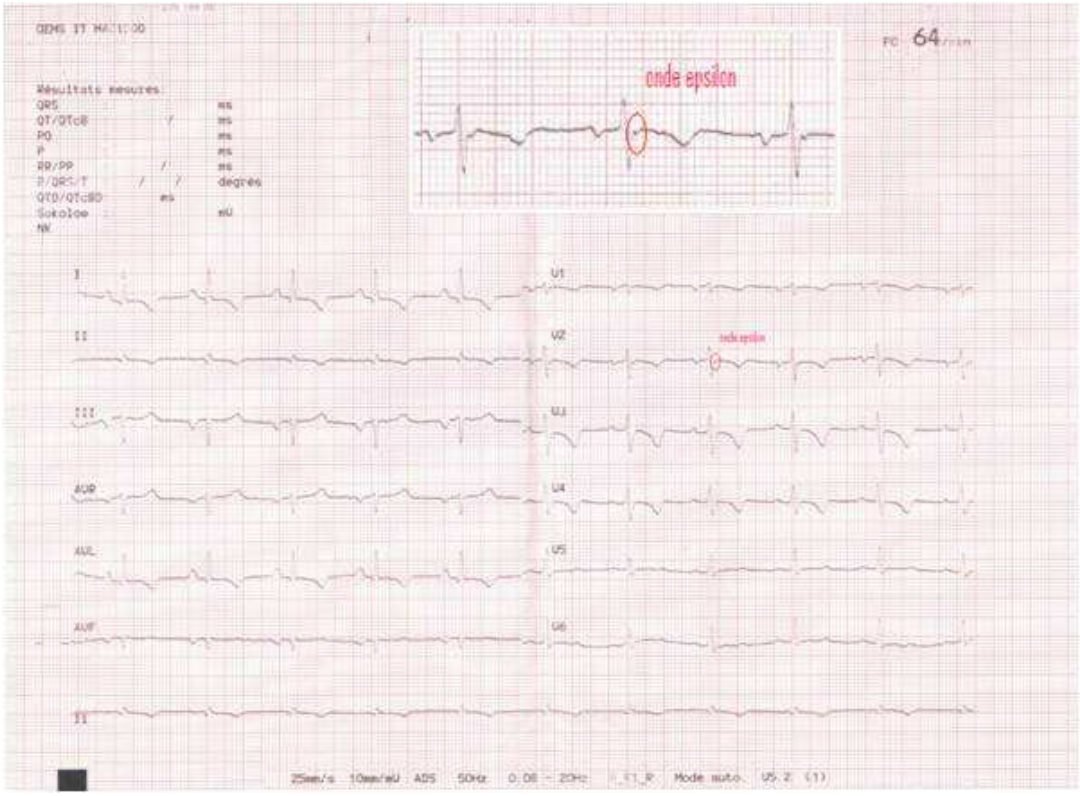

Figure 2: ECG showing Epsilon wave and inverted T waves in the right precordial leads.

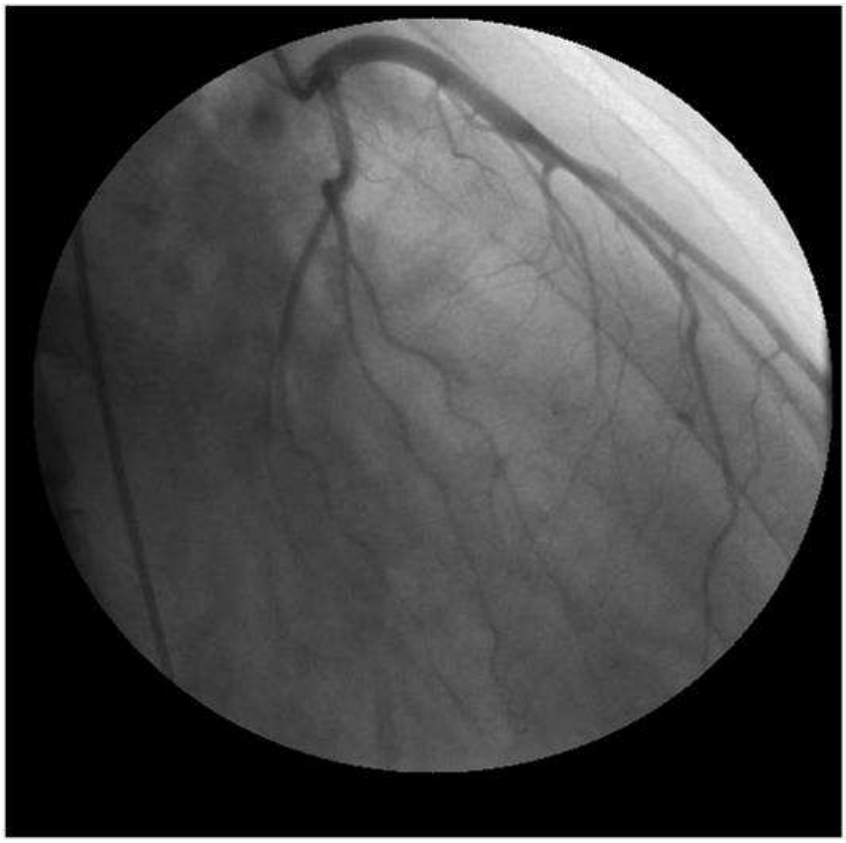

Figure 3: Coronary angiography was normal.

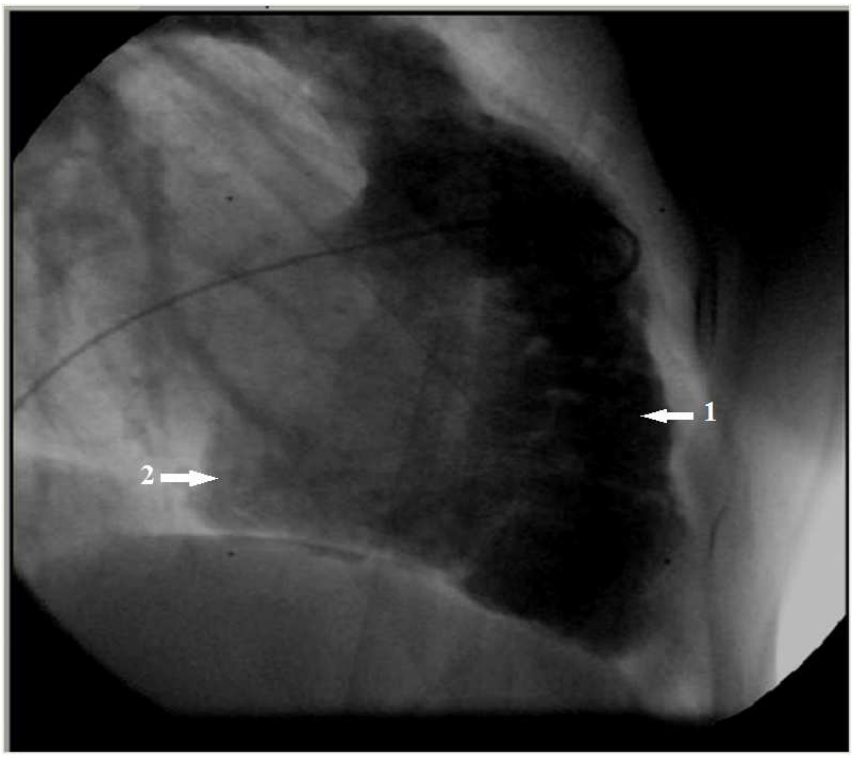

Figure 4: Right ventricular angiography confirmed ARVD
(1- Trabecular hypertrophy, 2-Bulge below tricuspid).

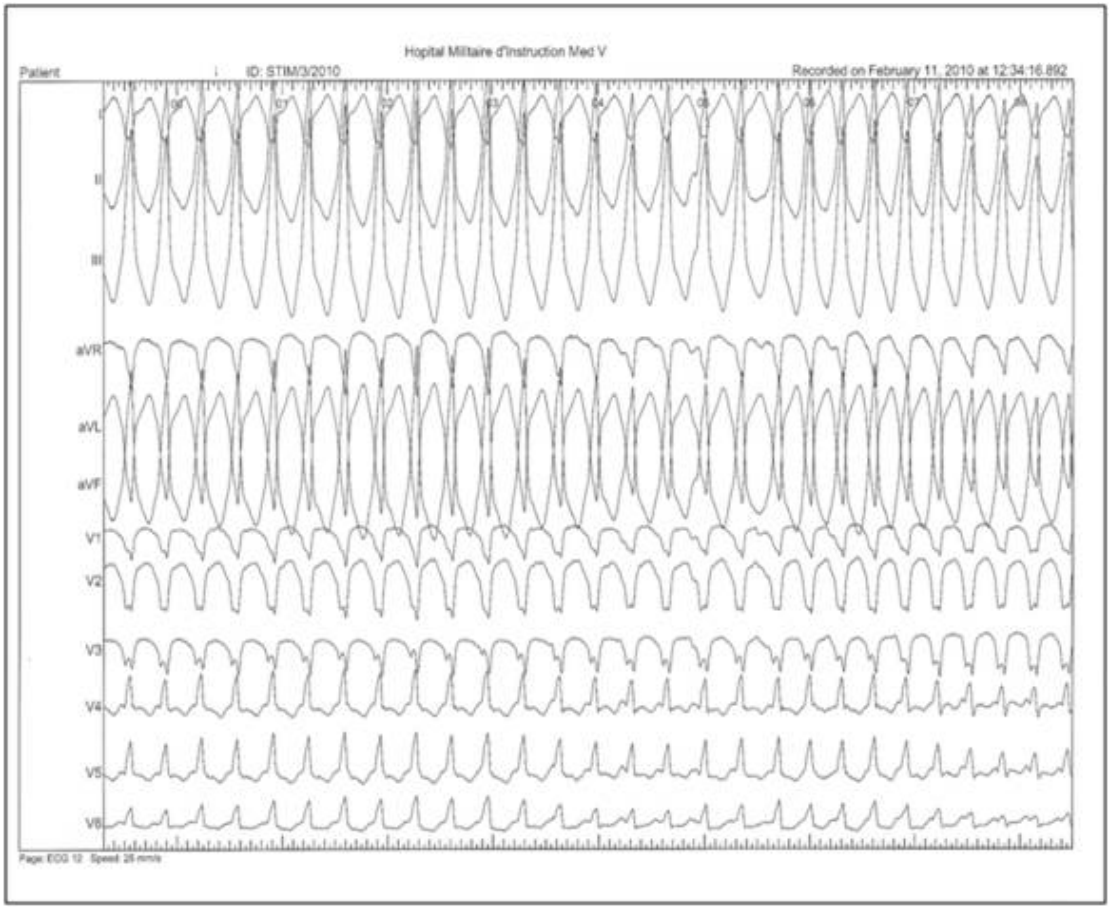

Figure 5: Ventricular tachycardia triggered under pharmacological treatment by programmed ventricular stimulation.

REFERENCES

[1]. Fontaine G, Fontaliran F, Hebert JL et al. Arrhythmogenic right ventricular dysplasia. Annu Rev Med, **1999**; 50: 17-35.
[2]. Fontaine G, Frank R, Guiraudon G et al. Signification des troubles de conduction intraventriculaire observés dans la dysplasie ventriculaire droite arhythmogène, Arc Mal Cœur, **1984**; 77:872-879.
[3]. A Jacquier, E Bressollette, JP Laissy, JY Gaubert et al. IRM et dysplasie arythmogène du ventricule droit (DAVD). J. Radiol **2004** ; 85 : 721-4.
[4]. R. Frank et al. Dysplasie ventriculaire droite arythmogène et mort subite. Annales de Cardiologie et d'Angéiologie 54 (**2005**) 21–25.
[5]. Ahmad F. The molecular genetics of arrhythmogenic right ventricular dysplasia-cardiomyopathy. Clin Invest Med **2003**; 26:167-78.
[6]. Fabre A, Sheppard MN. Sudden adult death syndrome and other non ischemic cause of sudden cardiac death: a UK experience. Heart **2005**; 91:1031-1035.
[7]. Corrado D, Theine G, Nava A, Rossi L, Pennelli N. Sudden death in young competitive athletes: clinicopathological correlation in 22 cases. Am J Med **1990**; 89:588-596.
[8]. Thiene G, Nava A, Corrado D, Rossi L, Pennelli N. Right ventricular cardiomyopathy and sudden death in young people. N Engl J Med **1988**; 318:129-133.

[9]. Sheldon M. Singh et Gordon W. Moe. Cardiologie-Conférences scientifiques. Octobre **2006**. Vo l u m e X I, numéro 8.
[10]. Marcus FI, McKenna WJ, Sherrill D, Basso C, Bauce B, Bluemke DA, et al. Diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia: proposed modification of the Task Force Criteria. Eur Heart J **2010**; 31(7):806–14.
[11]. Sharma S, Zaidi A. Exercise-induced arrhythmogenic right ventricular cardiomyopathy: fact or fallacy? Eur Heart J **2012**; 33(8): 938–40.
[12]. Corrado D, Basso C, Thiene G. Cardiomyopathy: Arrhythmogenic right ventricular cardiomyopathy: diagnosis, presentation and treatment. Heart **2000**; 83: 588-95.
[13]. Kayser HW, van der Wall EE, Sivananthan MU, Plein S, Bloomer TN, de Roos A. Diagnosis of arrhythmogenic right ventricular dysplasia: a review. Radiographics **2002**; 22:639-50.
[14]. Saffitz JE, Dependence of electrical coupling on mechanical coupling in cardiac myocytes: insights gained from cardiomyopathies caused by defects in cell-cell connections,Ann N Y Acad Sci, **2005**; 1047: 336-344.
[15]. Dalal D, Nasir K, Bomma C, et al. Arrhythmogenic right ventricular dysplasia: a United States experience. Circulation **2005**;112: 3823-3832.
[16]. McKenna WJ, Thiene G, Nava A, Fontaliran F, Blomstrom-Lundqvist C, Fontaine G, et al. Diagnosis of arrhythmogenic right ventricular dysplasia/cardiomyopathy. Task Force of theWorking Group Myocardial and Pericardial Disease of the European Society of Cardiology and of the Scientific Council on Cardiomyopathies of the International Society and Federation of Cardiology. Br Heart J **1994**; 71(3): 215–8.
[17]. Nava A, Rossi L, Thiene G. Arrhythmogenic right ventricular cardiomyopathy/ dysplasia. Amsterdam: Elsevier; **1997**.
[18]. Nava A, Canciani B, Buja G, et al. Electrovectorcardiographic study of negative T waves on precordial leads in arrhythmogenic right ventricular dysplasia: relationship with right ventricular volumes. J Electrocardiol **1998**; 21: 239-245.
[19]. Marcus F, Mc Kenna W, Sherill D, et al. Diagnosis of arrhythmogenic right ventricular cardiomyopathy / dysplasia. Proposed modification of the task force criteria. Circulation **2010**; 121: 1533-1541.
[20]. Roguin A, Bomma CS, Nasir K, Tandri H, Tichnell C, James C, Rutberg J, Crosson J, Spevak PJ, Berger RD, Halperin HR, Calkins H. Implantable cardioverter-defibrillators in patients with arrhythmogenic right ventricular dysplasia/cardiomyopathy. J Am Coll Cardiol, **2004**; 43: 1843-52.
[21]. Nicolas BADENCO, Dysplasie ventriculaire droite arythmogène: Evaluation de l'éfficacite des anti-arythmiques dans une cohorte de 134 patients, thèse pour le Doctorat en médecine, Faculté de médecine, Université Paris V, **2009**.
[22]. Witcher T, Borgrefe M, Haverkamp W et al. Efficacy of antiarrhythmic drugs in patients with arrhythmogenic right ventricular disease. Results in patients with inducible and noninducible ventricular tachycardia. Circulation, **1992**; 86: 29-37.
[23]. Tonet J, Frank R, Fontaine G, Grosgogeat Y. Efficacy of the combination of low doses of beta-blockers and amiodarone in the treatment of refractory ventricular tachycardia. Arch Mal Coeur Vaiss. **1989** Sep; 82 (9): 1511-7.
[24]. Corrado D, Leoni L, Link MS, et al. Implantable cardioverter defibrillator therapy for prevention of sudden death in patients with arrhythmogenic right ventricular cardiomyopathy/ dysplasia. Circulation **2003**; 108: 3084-91.
[25]. Groeneveled PW, Matta MA, Suh JJ, Heidenreich PA, Shea JA. Coasts and quality of life effects of implantable cardioverter-defibrillators. Am J Cardiol **2006**; 98: 1409-1415.
[26]. Gear K, Marcus F. Arrhythmogenic right ventricular dysplasia/cardiomyopathy. Circulation **2003**; 107: e31-3.