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Case Report

A case of arrhythmogenic right ventricular cardiomyopathy without ventricular arrhythmias

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ABSTRACT

We submit a case report of a 21-year-old man admitted to our emergency room for acute right heart failure. Arrhythmogenic right ventricular cardiomyopathy (ARVC) was diagnosed but ventricular arrhythmias have never been detected. On the basis of the diagnosis of ARVC and for the frequent episodes of symptomatic bradycardia, the patient underwent implantation of a biventricular defibrillator (CRT-D).

In ARVC symptoms usually appear between the ages of 30–50. Especially in young patients the most common clinical presentation of ARVC are palpitations and syncope due to ventricular tachycardia with left bundle branch morphology. In cases of older patients in whom the disease has been described, the clinical presentation is mainly represented by signs and symptoms of right or biventricular heart failure.

Our case report shows how, in young patients, this disease may present at the beginning as acute right, heart failure, without ventricular arrhythmias but with episodes of symptomatic bradycardia that, require the use of cardiac resynchronization therapy to avoid the well-known long-term adverse effects of right ventricular pacing.

1. Introduction

Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC) is a genetic form of cardiomyopathy that primarily affects the right ventricle (RV). The term "cardiomyopathy" should be preferred to "dysplasia", as already suggested by WHO/International Society and Federation of Cardiology in 1996.[1] The estimated prevalence of ARVC in the general population is approximately 1:5000, affecting men more frequently than women with a ratio of 3:1.[2]

In most cases is transmitted with an autosomal dominant pattern of inheritance, with incomplete penetrance and variable expression. ARVC may also involve the left ventricle (LV) and culminate in life-threatening ventricular arrhythmias, sudden cardiac death (SCD) and/or biventricular heart failure.[3]

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2. Discussion

ARVC causes 11–22% of SCD in the young athletes patient population, resulting in approximately 22% of cases in athletes in northern Italy[4] and about 17% of SCD in young people in the United States.[5] The pathologic hallmark of disease is myocardial atrophy (myocyte loss), fibrofatty replacement, fibrosis and thinning of the wall with chamber dilation and aneurysm formation.[6] The genetics of the disease seems to support the hypothesis that it may be caused by desmosomal dysfunction (mutation of plakophilin-2, desmoglein-2, desmocollin-2, desmoplakin genes), that lead to impaired mechanical and electrical coupling between individual cells, leading to myocyte uncoupling, especially under conditions that increase myocardial strain, for example during physical effort. These changes produce electrical instability precipitating ventricular tachycardia and SCD. Currently only approximately 50% of probands are found to have a pathogenic mutation, possibly due to the high incidence of rare mutations in many patients and actually the most important utility of genetic test is in the screening of asymptomatic family members of probands who have a pathologic genetic abnormality.[7]

Diagnosis of ARVC relies on a scoring system, formulated in 2010 by the revisited Task Force, with major and minor criteria based on the demonstration of a combination of defects in RV morphology and function, characteristic depolarization/repolarization electrocardiogram abnormalities (negative T waves and/or "epsilon" waves in right precordial leads), characteristic tissue pathology, typical arrhythmias, family history, and the results of genetic testing.[8]

Assessment of ventricular structure and function is critical for the diagnosis and prognosis of ARVC. Transthoracic echocardiography provides a noninvasive method of RV imaging; suggestive findings of ARVC include global or segmental wall motion abnormalities (akinesis/dyskinesis) with cavity dilation, hypertrophic RV trabeculation, and systolic dysfunction.

Cardiac magnetic resonance (MRI) is considered the best imaging modality in evaluating the RV in ARVC and provides tissue characterization and identification of intramyocardial fat and fibrosis in addition to assessment of ventricular structure and function.

Endomyocardial biopsy appears to have low diagnostic sensitivity for several reasons as the patchy distribution of the disease and the high rate of sampling error.[9]

The main goal of therapy is to prevent life-threatening events. To achieve this it is necessary to identify high-risk patients for malignant arrhythmias and SCD. These patients should be considered for ICD implantation in primary prevention. Consequently patients with unexplained syncope, non-sustained VT on noninvasive monitoring, familial history of sudden death, extensive disease including those with LV involvement and good functional status are potential candidates for ICD implantation even in the absence of ventricular arrhythmias. ICD implantation in secondary prevention is obviously mandatory.[9]

VT ablation has an ancillary role and may be helpful in reducing symptoms and ICD firing but not able to prevent SCD. Antiarrhythmic drugs, primarily betablockers, sotalol, and amiodarone have been used for symptomatic control in patients who are not candidates for ICD or as an adjunct therapy to reduce frequent ICD firing due to recurrent VT, but the evidence available has been derived from observational studies. Finally, but not least, an important recommendation for patients with ARVD is exercise restriction because an intense physical activity appears to increase the rate of progression of the disease.[9]

3. Case report

A 21-year-old man, with no cardiovascular risk factors, was referred to our emergency room for asthenia, dyspepsia and dyspnea on exertion from about ten days. The medical history report surgery for bilateral congenital cataract in childhood.

At the admission blood pressure was 100/65 mmHg, the ECG showed junctional rhythm at 42 bpm, complete right bundle branch block, inverted asymmetrical T waves from V1 to V6 (Fig. 1).

The blood sample showed increasing of inflammatory markers and hypertransaminasemia. Abdominal ultrasound

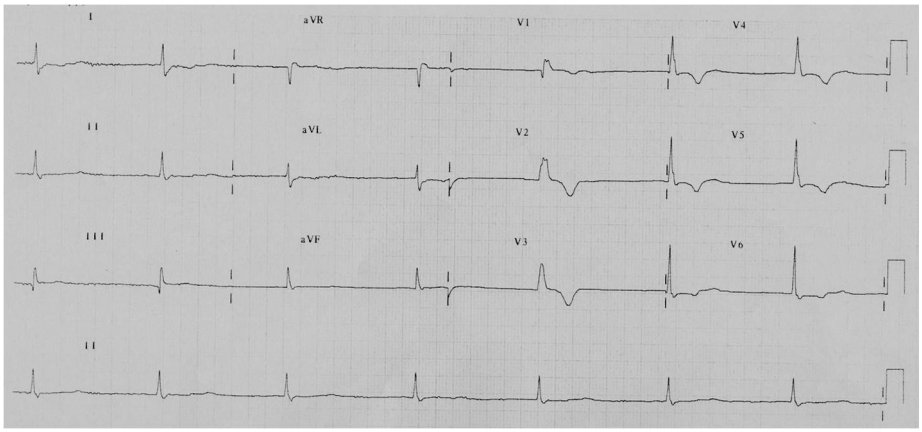

Fig. 1 – ECG at the admission.