ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. CLINICAL CASE

SIMONA COSTEA[1], IRIS MUREŞAN[2], ADRIAN BOICEAN[3]

[1]*Clinical County Emergency Hospital of Sibiu,* [2,3]*"Lucian Blaga" University of Sibiu*

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Abstract: *Arrhythmogenic right ventricular dysplasia (ARVD) is a rare form of spontaneous or family-inherited cardiomyopathy, autosomal dominant or recessive, characterized by progressive loss of myocyte mass and its replacement with fibroadipose tissue, which leads to ventricular dysfunction, ventricular arrhythmias and sudden death. Studies have shown that ARVD is present in 0.08-9% of cases of sudden death. ARVD main feature is the tendency to ventricular arrhythmias and sudden death even in the absence of clinical ventricular dysfunction. Physical exertion produces catecholamines release and hyperextension of myocardial fibers, which can lead to stimulation of ventricular displasic areas, producing arrhythmias. Clinical case: Young man, 32 years old, smoker without no pathological personal history suffered at home a cardiac arrest; he is resuscitated, stabilized. ECG: rare ESV of normal aspect of arrhythmic episodes, crochet discrete terminal portion of QRS in V1 (epsilon wave). MRI: morphologically, there has been highlighted a small area located about 2 cm at anterior apical ventricular septal level with increased signal; paradoxical motion in right septal anterior ventricular wall with little area "buldging" in systole. There has been decided to implant a cardiac defibrillator.*

INTRODUCTION

Arrhythmogenic right ventricular dysplasia (ARVD) is a rare form of spontaneous of family inherited cardiomyopathy, autosomal dominant or recessive, characterized by progressive loss of myocyte mass and its replacement with fibroadipose tissue that determines ventricular dysfunction, ventricular arrhythmias and sudden death.(1)

Studies have shown that ARVD is present in 0.08-9% of cases of sudden death recorded annually and can achieve up to 25% of all cases of sudden death in athletes (figure no. 1). ARVD was first described in 1977 by Fontaine as a regional or global abnormality of the right ventricle, which initially may be asymptomatic, and in the advanced forms, it leads to symptoms of right heart failure.(2) It was later found that it is possible to affect the left ventricle, as well, either through ventricular interdependence, or through the extension of dysplasia process with signs of heart failure.(3)

CASE REPORT

Young man, 32 years old, smoker without any pathological personal history, suffers a cardiac arrest at home, after a party, at 4:30 o'clock in the morning; he is resuscitated stabilized and brought to the County Clinical Emergency Hospital of Sibiu, in the Emergency Room by the Mobile Emergency Service for Resuscitation and Extrication team.

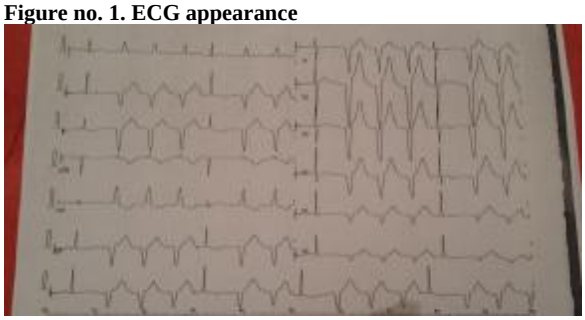
From family medical history, we learnt that his father died suddenly at the age of 64, from ischemic dilated cardiomyopathy (DCM).

Physical examination upon admission revealed: sedated, oro-tracheally intubated and mechanically ventilated patient, electrically and hemodynamically stable, blood pressure (BP) = 110/60 mm Hg without detectable murmurs without pulmonary crackles. During the following hours in hospital, the patient suffered multiple cardiorespiratory arrests by ventricular fibrillation and torsades de pointes, which were resuscitated. Antiarrhythmic therapy was administered with beta-blockers, Amiodarone, hyperpolarize solutions and electric shocks.

Laboratory investigations revealed: normal cardiac markers, leukocytes: 20200-10300-10400/mm^3, hemoglobin = 13.5 g%; Hematocrit = 40%; Platelets = 151,000/mm^3, erythrocyte sedimentation rate (ESR)= 20-23mm/h; Fibrinogen = 528-735-5020mg/dl; C reactive protein (CRP) = negative, positive, negative; glycemia = 828mg%; = 32mg% urea, creatinine = 0,74mg%, ASAT = 91-170-22ui; ALAT = 102-112-57, Cholesterol = 157% ionogram: Na = 140-133-137-135mmol/l, K = 4,8-4,9-4,4mmol/l; Amylasemia = 53ui; = total bilirubin 0.8 mg%; direct bilirubin = 0,39mg%.

Paraclinical examinations: On ECG = rare ventricular extrasystoles, normal aspect outside the arrhythmic episodes, discrete crochet in the terminal portion of QRS in V1 (epsilon wave).

Figure no. 1. ECG appearance



Thoracic radiography showed no pleural acute lung injury; Echocardiography = no change in kinetics without dilated cavities, no valvulopathy; Angiography showed epicardial coronary arteries with no damages or defects; On MRI, there has been morphologically highlighted a small area of about 2 cm, located in the apical, anterior-septal region of the

[1]Corresponding author: Simona Costea, Str. Lucian Blaga, Nr. 2A, Sibiu, Romania, E-mail:costeasimona13@yahoo.com, Phone: +40726 576141
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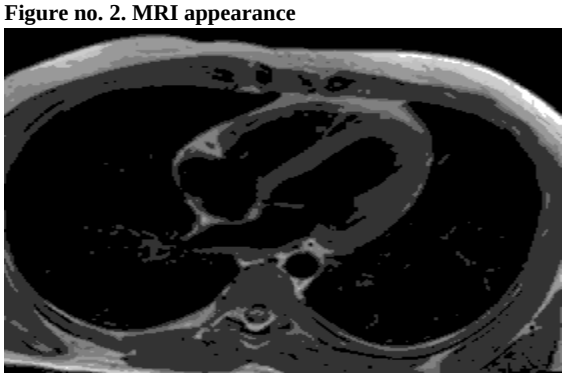
right ventricle with increased signal; paradoxical motion at the level of the right septal anterior ventricular wall with small area of "buldging" in systole.

Figure no. 2. MRI appearance



The patient is referred to the interventional cardiology ward for electrophysiology studies.

The patient was diagnosed with arrhythmogenic right ventricular dysplasia. Implanting a cardiac defibrillator is decided, the procedure taking place without any incidents.

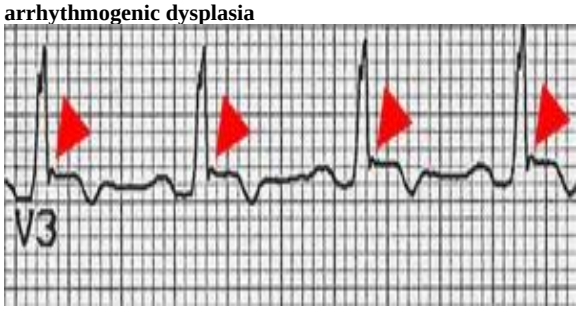
Patient evolution was good; the patient was hemodynamically stable, without subjective complaints and without arrhythmias. There has been decided to stop amiodarone treatment, continuing only the beta blocker treatment (Concor 5 mg).

DISCUSSIONS

The main feature of the ARVD is the propensity for ventricular arrhythmias and sudden death even in the absence of clinical ventricular dysfunction. The risk of sudden death among athletes is higher, reaching up to 20-25%, which is linked to the occurrence of ventricular arrhythmias during exercise, 4.5%.(4)

Physical exertion brings about catecholamines release and hyperextension of myocardial fibers, which can lead to stimulation of ventricular dysplastic areas producing arrhythmias. Thus, in a study of 160 subjects with clinical criteria of ARDV, followed for several years, 24 patients (18.5%) died during the study period by major ventricular disorders.(5)

Figure no. 3. ECG with wave epsilon in right ventricular arrhythmogenic dysplasia



So far, there have been known six genes that are associated with autosomal dominant and recessive transmission. Genetic tests apply only to selected patients. ARDV is more common in men (ratio 3: 1) of middle age (31 ± 14).(5)

In mild forms of ARDV, electrocardiographic signs are missing or are subtle. In advanced forms of ARDV, due to myocardial tissue destruction, ventricular depolarization and repolarization changes occur predominantly in the right ventricle, which have various electrocardiographic expressions. Electrocardiogram in ARDV shows the presence of incomplete right bundle branch block, negative T waves and epsilon wave in right precordial derivations - epsilon wave appears as a small positive deflection following the QRS complex, which represents late right ventricular potentials.

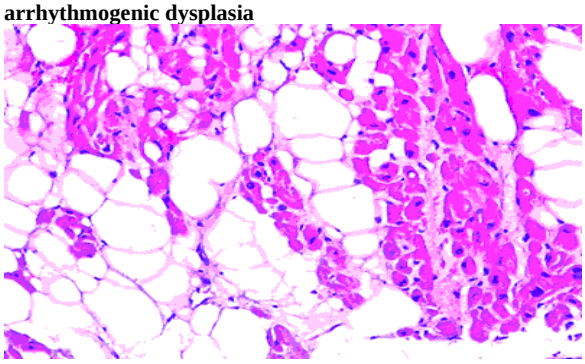
MRI is the most accurate method to assess cardiac morphology and function, due to a high spatial and contrast resolution, thereby contributing to the diagnosis of ARDV by:

1. detecting intramyocardial adipose depots, often in the free wall and anteriorly and laterally of the right ventricle ;
2. highlighting inflammation and myocardial fibrosis;
3. detecting anomalies of right ventricular wall thickening with hypertrophy of anterolateral wall > 8 mm in the early stages and its reduction <2 mm in advanced stages associated with the presence of segments dilations and aneurysms;
4. global and segmental right ventricular dysfunction, frequently localized at the level of free wall, apex and right ventricular outflow tract.(6)

Histopathological changes

Histopathological examination found inflammatory infiltrate necrosis and infiltrated fibrous fat in the free open and anteriorly-laterally to the right ventricular "triangle of dysplasia": anterior side of the infundibulum, apex and the posterior and lower sides of the filling and ejection tract of the right ventricle.

Figure no. 4. Histological appearance in right ventricular arrhythmogenic dysplasia



Replacement of myocardic tissue with fibrous adipose tissue occurs by three mechanisms:(7)

1. apoptosis;
2. myocardial inflammation;
3. myocardial dystrophy.

Differential diagnosis is made with:

- other causes of sudden death: Anomalies of the coronary arteries (aneurysms, tumours), hypertrophic or dilated severe cardiomyopathy, hyperviscosity syndrome: polycythemia vera, essential thrombocythemia;
- acute myocardial infarction; pulmonary embolism;
- brain hemorrhage;
- dyselectrolytemia;
- poisoning with unknown substances.

CONCLUSIONS

Arrhythmogenic right ventricular dysplasia is a rare condition with vital prognosis. It must systematically be evoked in case of sudden death, syncope, during exercise in young patients or in ventricular tachycardia with left bundle branch block appearance.

The presence of ventricular extrasystoles with left bundle branch block appearance requires systematic invasive exploration, especially if adrenergic character is present. Therapeutic solutions in these patients should be stratified

according to the risk of arrhythmias.(8)

Short-term prognosis depends on the severity of arrhythmias and on long term, on the hemodynamic status.

The particularity of the case lies in the fact that our patient was in respiratory arrest with no cardiac complaints in his medical history. Although arrhythmogenic right ventricular dysplasia is one of the causes of sudden death, we have repeatedly managed cardiopulmonary resuscitation after admission; the patient was diagnosed and treated properly, with full therapeutic success.

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