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EC CARDIOLOGY
Case Report

Doc, Can I Continue to Play Sports?: Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

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Received: January 20, 2018; **Published:** February 24, 2018

Abstract

Arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D) is a rare genetic condition affecting mostly the right ventricle and sometimes also the left ventricle is involved in this cardiomyopathy. Over time it can lead to congestive heart failure, arrhythmia and sudden cardiac arrest. It can be a common cause of sudden cardiac arrest in athletes and therefore an implantable cardioverter-defibrillator is recommended.

Keywords: *Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia (ARVC/D); Right Ventricle; Arrhythmia*

Introduction

Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia (ARVC/D)

ARVC/D is characterized by progressive fibrofatty replacement of the right ventricular myocardium. It represents an underdiagnosed cardiac condition leading to recurrent ventricular tachycardia, heart failure, syncope and occasionally sudden cardiac death in younger people [1]. It is a genetic cardiomyopathy (mostly autosomal dominant) with an estimated prevalence of 1 in 1000 to 1 in 5000 [2]. Familial occurrence can be as much as 50% and it can account for 3 - 4% of deaths in sports with sudden cardiac deaths in population younger than 65 being 5% [3]. It is the second most common cause of death after hypertrophic cardiomyopathy (HOCM) causing up to 20% of sudden cardiac deaths in patients under the age of 35 years. It is more common in men than women (3:1) and people of Italian and Greek descent. Some patients might have an autosomal recessive expression with what is called Naxos Disease which is associated with woolly hair and skin changes.

Etiology

This condition is considered a part of idiopathic cardiomyopathies as per its nature of progressive disease with unclear pathogenesis and etiology. Basso., *et al*. [4] proposed four hypotheses for possible explanation of this disease status. 1st hypothesis: Apoptosis that is programmed cell death leading to progressive myocardial muscle damage and loss followed by fibrofatty replacement which increases the electrical vulnerability of the right ventricle leading to life-threatening arrhythmias [5]. 2nd hypothesis: the dysontogenic theory, this disease should be considered a congenital heart disease thereby leading to abnormal development of right ventricle and thus leading to dysplasia. The 3rd hypothesis: the degenerative theory: a metabolic disorder may affect the right ventricle and result in progressive replacement of myocardium by fat and fibrous tissue. 4th hypothesis: the inflammatory theory: the fibrofatty replacement of the right ventricle is considered as a healing process in the setting of idiopathic myocarditis [6].

Pathophysiology

In this condition, the most common location for fibrofatty replacement of the normal myocardium is between the anterior infundibulum, the right ventricular apex and inferior/diaphragmatic aspect of the right ventricle, the so-called "the triangle of dysplasia" [7] which can lead to dilation and aneurysm formation with paradoxical motion. The left ventricle and the interventricular septum can be affected at times.

There are two variants of this condition: the fatty variant mostly affects the right ventricle: almost complete replacement of myocardium without thinning of the ventricular wall and the fibrofatty variant: with significant thinning of the right ventricular wall and the left ventricle may be involved in this variant as well [8].

Clinical features and diagnosis

The classic presentation of ARVC is usually arrhythmia, in the form of ventricular tachycardia, frequently from the right ventricular outflow tract (RVOT) also known as Adenosine-sensitive ventricular tachycardia. The range of manifestations vary from asymptomatic to PVCs (premature ventricular contractions), biventricular heart failure, arrhythmia and sudden cardiac death, usually in young and the athletes [9].

The ECG changes seen in this condition may include [10]:

1. An Epsilon wave (most specific finding, seen in 30% of patients).
2. T wave inversions in V1-3 (85% of patients).
3. Prolonged S wave upstroke of 55 msec in V1-3 (95%).
4. Localised QRS widening of 110 msec in V1-3.
5. Paroxysmal episodes of ventricular tachycardia with LBBB morphology (RVOT).

The diagnostic criteria for this condition are described by the Task Force 1994 [11]: patients must have either 2 major criteria, one major and 2 minor criteria or 4 minor criteria:

1. Global and/or regional dysfunction and structural alterations:
    - Major:
        - Severe dilatation and reduction of right ventricular ejection fraction with no (or only mild) left ventricular impairment.
        - Localized right ventricular aneurysm (akinetic or dyskinetic areas with diastolic bulging).
        - Severe segmental dilatation of the right ventricle (RV).
    - Minor:
        - Mild global RV dilatation and/or reduction of ejection fraction with normal LV.
        - Mild segmental dilatation of the RV.
        - Regional right ventricular hypokinesia.

2. Tissue characterization of wall:
    - Major:
        - Fibrofatty replacement of myocardium at endomyocardial biopsy.
3. Repolarization abnormalities:
    - Minor:
        - Inverted T waves in right precordial leads (leads V2-3) (age >12; in the absence of RBBB).
4. Depolarization of conduction abnormalities:
    - Major:
        - Epsilon waves or localized prolongation (>110 msec) of the QRS complex in V1-3.
    - Minor:
        - Late potentials (signal-averaged ECG).
5. Arrhythmias
    - Minor:
        - LBBB type ventricular tachycardia (sustained and non-sustained (ECG, Holter and Treadmill exercise).
        - Frequent PVCs on Holter (> 1,000 in 24 hours).

    Family history:
    - Major:
        - Familial history confirmed at necropsy or surgery.
    - Minor:
        - Family history of premature sudden death due to suspected RV cardiomyopathy.
        - Family history (clinical diagnosis based on present criteria).

Imaging evaluation of ARVC

Imaging techniques may include conventional angiography, echocardiography, radionuclide angiography, CT and cardiac MRI.

Right ventricular angiography is regarded the standard of reference for the diagnosis of ARVC, however it is invasive. Echocardiography is non-invasive and widely available technique that can be employed for diagnosing other anatomic abnormalities as well in addition to ARVC [12]. However, MR imaging allows a 3-dimensional evaluation of ventricular anatomy and volumes and because of its excellent spatial resolution and unlimited field of view, this technique is best suited to study the right ventricle and the changes seen in ARVC [13-15].

Cardiac MRI findings

Several studies have reported on the use of MR imaging to detect the characteristic high signal intensity of fat in the RV myocardium on T1-weighted images [16] and thus more and more cases of ARVC are being diagnosed because of this recognition by MR imaging [12].

However, some studies have reported significant fatty infiltration of the RV in more than 50% of normal hearts in elderly people [17]. In addition, the reported sensitivity and specificity of this technique is 67% and 100% respectively in one study, while another study reported the sensitivity of this diagnostic test only 22% [14]. On the other hand, Basso., et al. [4] reported the sensitivity of 100% compared to the endomyocardial biopsy.

MR imaging can also be used to assess both systolic and diastolic function and the presence of early RV diastolic dysfunction could be an early marker of disease even while systolic function is preserved.

Characteristic findings on Cardiac MRI:

1. Fatty infiltration of the RV myocardium with high signal intensity on T1-weighted images.
2. Fibrofatty replacement leading to diffuse thinning of the RV myocardium.
3. Aneurysm of the RV and RVOT.
4. Dilatation of the RV and RVTO.
5. Regional contraction abnormalities.
6. Global systolic dysfunction and global diastolic dysfunction.

Management

Treatment includes:

1. Avoidance of strenuous exercise and competitive sports.
2. B-blockers, antiarrhythmic Rx and catheter ablation.
3. Heart failure management as per the guidelines.
4. Implantable cardioverter defibrillator (ICD).
5. Surgery and cardiac transplantation.

Indications for ICD:

1. Symptomatic ventricular tachycardia.
2. Cardiac arrest.
3. First-degree relative with sudden cardiac death.S
4. Failed drug therapy for ventricular tachycardia (even if asymptomatic).

Prognosis

It is a progressive disease and mostly will lead to RV failure and sudden cardiac death. The death rate in this condition has been reported to be 2.5% per year.

Case Report

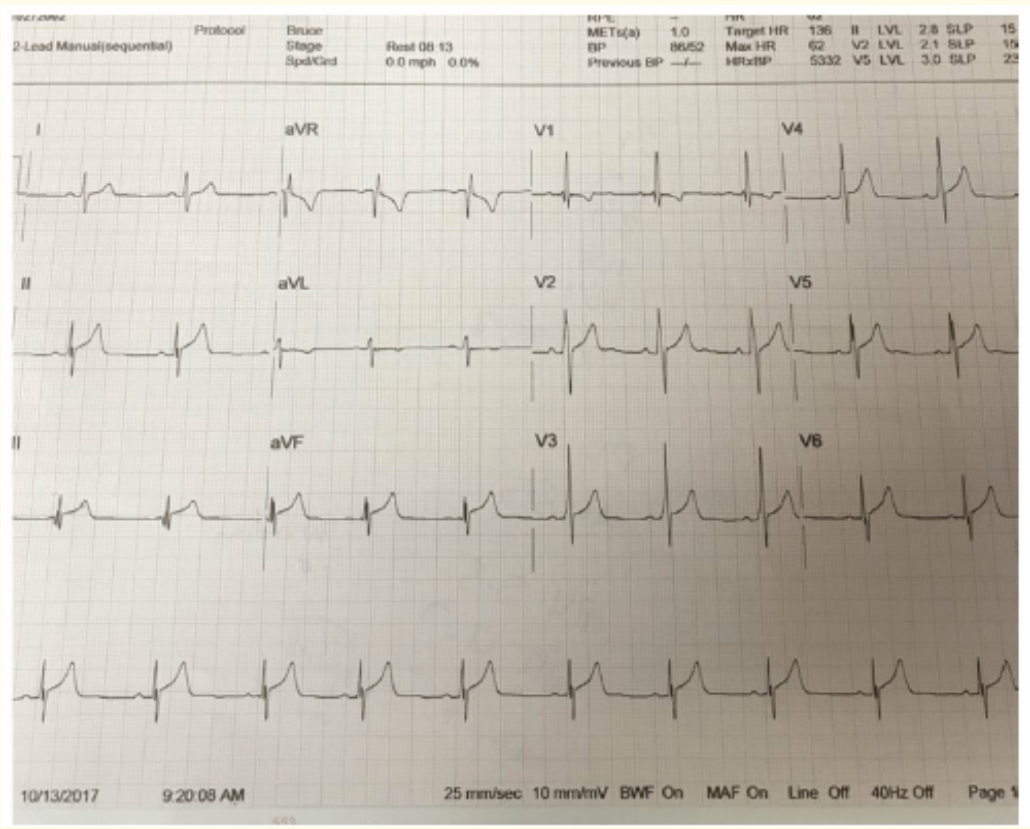
A 15-year old teenage boy was seen in my office for cardiac consultation after having chest injury during his school sports. He was initially seen in the Emergency Room locally for his evaluation and found to be stable. He was sent home requesting him to come to my office for a cardiac consult. During wrestling, he was hurt physically to his chest and needed an urgent evaluation.

By the time he came to my office, his chest pains had resolved and his vitals were stable. There is no history of any heart disease in the family and no history of congenital heart disease.

His physical examination was unremarkable. Cardiac exam: S1, S2 normal. No S3 heard. No rub, gallop or murmur noticed.

As he was clinically quite stable without any symptoms, he has not been placed on any medical regimen so far. His Holter monitor did not reveal any significant arrhythmia.

His baseline resting electrocardiogram (ECG) was abnormal: normal sinus rhythm, prominent R waves in the right precordial leads suggestive of pulmonary hypertension or right ventricular hypertrophy (RVH):



(Figure)

His 2D- echocardiographic study revealed: preserved left ventricular (LV) systolic function with LV ejection fraction (LVEF) of 60 - 65%, normal left atrial size, dilated right ventricle and mildly dilated right atrium. No evidence of any thrombus and no significant valvular dysfunction noted. No evidence of pericardial effusion.

Question: why were his ECG and 2D-echo studies abnormal? Were these studies abnormal due to his recent chest injury or these abnormal findings were already there and the history of chest injury was simply a 'distraction' in his diagnosis?

Based on these abnormal findings, a cardiac MRI was requested which reported following pertinent findings:

1. Severely increased right ventricular volume with RV apical hypokinesis.
2. Normal global LV and RV function.
3. Increased RV volume and hypokinesis suggestive of Arrhythmogenic right ventricular cardiomyopathy.
4. Genetic testing recommended.

Bigger Question: After all these tests, this teenager boy (wishes and) wants to know if he could continue to play the sport of his choice!

Update on this case

1. He has been advised strictly not to take part in any sports until further notice.
2. He has been referred for genetic testing and further work up related to familial screening.
3. His family wished for a second opinion and therefore he has been referred to The Arrhythmia Section, Pediatric Cardiology at Cleveland Clinic, Cleveland, OH.
4. After his consultation at Cleveland Clinic, he will be advised to consider an implantable cardioverter defibrillator (ICD) as part of his prophylactic therapy.
5. Being physically quite stable and asymptomatic, he is not on any medical treatment at this time.

Can he continue to play sports?

There are limited data on this issue. Any competitive sport with strong physical interaction should not be allowed. After having an ICD in place, he should be able to play, however not the competitive sports at this point.

Conclusion

Even though a rare disorder it can be truly a life changing diagnosis for someone affected with this. My patient is only 15 years old and has been advised not to take part in any competitive sports. He will be seeing an electrophysiologist soon and will likely be recommended to have an ICD implanted prophylactically.

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Volume 5 Issue 3 March 2018