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OLGU YAZISI / CASE REPORT

APİKAL TROMBÜS İLE SEYREDEN ARİTMOJENİK SAĞ VENTRİKÜL DİSPLAZİSİ KARDİYOMİYOPATİLERİN ATİPİK BULGUSU

ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA WITH APICAL THROMBUS ATYPICAL PRESENTATION OF CARDIOMYOPATIES

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ÖZ

Aritmojenik sağ ventrikül displazisi (ARVD) nadir görülen bir kardiyomiyopatidir. ARVD çoğunlukla genç yaşlarda tanı alır ve kendisini ventriküler aritmiler, çarpıntı, baş dönmesi, kalp yetmezliği ve hatta ani kardiyak ölüm ile gösterebilir. Görüntüleme yöntemleri ile sağ ventrikül (SV) dilatasyonu ve apikal anevrizma tipik bulgusudur. Fakat ARVD olgularında intraventriküler trombüs çok nadir görülmektedir. 19 yaşında erkek hasta, hastanemize çarpıntı ve bayılma şikâyetleri ile başvurdu. Elektrokardiyografisinde ön yüz derivasyonlarda T negatifliği bulunmakta idi. Ekokardiyografide sağ ventrikül dilate ve SV apeksinde anevrizmatik oluşum içinde trombüs görüldü (**Fig-1**) . Kardiyak manyetik rezonans incelemede sağ ventrikül genişlemesini, yağ infiltrasyonunu, fibrotik dokuları, SV duvar hareket bozukluğunu ve trombüslü apikal anevrizma doğrulandı. Antikoagulan tedaviyle üç ay sonra trombüsün rezole olduğu gözlendi ve ICD implante edildi. ARVD tanısında elektrokardiyografik, aritmik, histolojik ve ailesel özelliklerin yanında görüntüleme yöntemleri de büyük önem taşımaktadır. Sağ ventrikül dilatasyonu ve apikal anevrizması tanı sürecinde önemli kriterler olmakla birlikte bu gibi bulgular saptandığında trombüs varlığı da dikkatlice değerlendirilmelidir.

ANAHTAR KELİMELER: Aritmojenik Sağ Ventrikül Displazisi, Fibrofatty İnfiltrasyon, Trombüs

ABSTRACT

Arrhythmogenic right ventricular dysplasia (ARVD) is a rare form of cardiomyopathy. It commonly presents in young adults with ventricular tachycardia or sudden death. Right ventricular (RV) dilatation and apical aneurysm are the typical findings in imaging methods. However intraventricular thrombus is rarely seen in ARVD cases. A 19 year old male was admitted to hospital with palpitation and syncope. T wave inversion was detected on anterior surface electrocardiogram. Transthoracic echocardiography revealed dilated RV and apical aneurysm in which thrombus located (**Fig-1**). Cardiac magnetic rezonans imaging comfirmed RV enlargement, fatty infiltration, fibrosis, wall motion abnormalities and apical aneursym with thrombus. Anticoagulation theraphy commenced to the patient. After three months later trombus resoluted and ICD was implanted. Imaging methods have a great importance in the diagnosis of ARVD besides electrocardiographic, arrhythmic, histological and familial characteristics. While right ventricular dilatation and apical aneurysm are important criteria for the diagnosis process, the presence of thrombus should be evaluated carefully.

KEYWORDS: Arrhythmogenic Right Ventricular Dysplasia, Fibrofatty Infiltration,Thrombus

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INTRODUCTION

Arrhythmogenic right ventricular dysplasia(ARVD) is a rare form of cardiomyopathy in which the heart muscle of the right ventricle (RV) is replaced by fat and/or fibrous tissue. The right ventricle is dilated and contracts poorly. It commonly presents in young adults with ventricular tachycardia or sudden death (1). Researchers have found two patterns of inheritance for ARVD; autosomal dominant, the family members have a 50 percent chance of inheriting the condition, autosomal recessive, one form is called Naxos disease. ARVD is usually diagnosed at a young age and symptoms may include ventricular arrhythmias, palpitations, dizziness, heart failure and also sudden cardiac death. ARVD is diagnosed on medical history, physical exam, and tests (echocardiogram, Holter monitor, electrophysiologic testing, cardiac MRI, and/or cardiac CT scan). Cardiac MRI is an important test for the diagnosis as it visualizes fibrofatty infiltration of the right ventricular (RV) myocardium(2).

CASE REPORT

A 19 year old male was admitted to hospital with palpitation. There was no family history of heart disease or sudden death. On admission he was haemodynamically stable and was not in heart failure. T wave inversion was detected on surface ECG and had no other abnormalities. Laboratory tests contain complete blood count, liver-thyroid-renal parameters, serum electrolytes and cardiac markers and all of them were normal. Echo showed: dilated right ventricle with outpouching in the right ventricular cavity and apex aneurysm with thrombus in it (**Fig-1**). Due to his palpitation history Holter ECG was performed but no arrhythmogenicrythm was detected. Cardiac MRI revealed right ventricular enlargement, fatty infiltration, fibrosis, wall motion abnormalities and apical aneurysm with thrombus. Anticoagulation started with ACE (angiotensin converting enzyme) inhibitor,and beta blocker, after three months thrombus resolute and ICD was implanted.

DISCUSSION

ARVD is a leading cause of sudden death among young athletes. But it can affect people of all ages and all activity levels. The major condition which needs to be differentiated from ARVD/C is idiopathic ventricular tachycardia arising from the outflow tract (3). The electrocardiogram (ECG) provides important diagnostic information in patients suspected of having right ventricular cardiomyopathy/dysplasia. Normally, the free wall of the right ventricle is the last part of the heart to undergo depolarization. If there is selective damage to the right ventricular free wall musculature, there may be fragmentation and selective slowing and prolongation of the end of the QRS complex and this can be seen in the anterior precordial leads. The delay in depolarization may be extremely prolonged and may be visible as reproducible low frequency waves that extend beyond the QRS complex and before the T wave. These are known as postexcitation or epsilon waves (4,5). They are of low amplitude and are usually visible only on the ECG leads overlying the right ventricle.

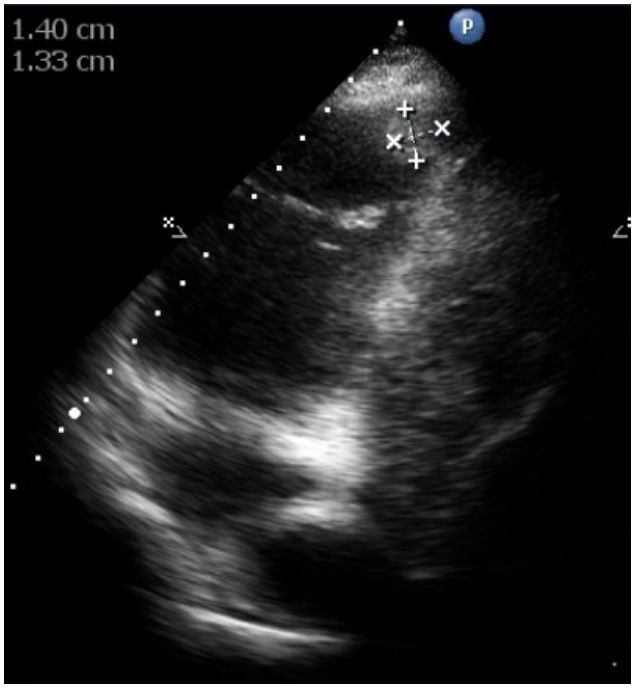
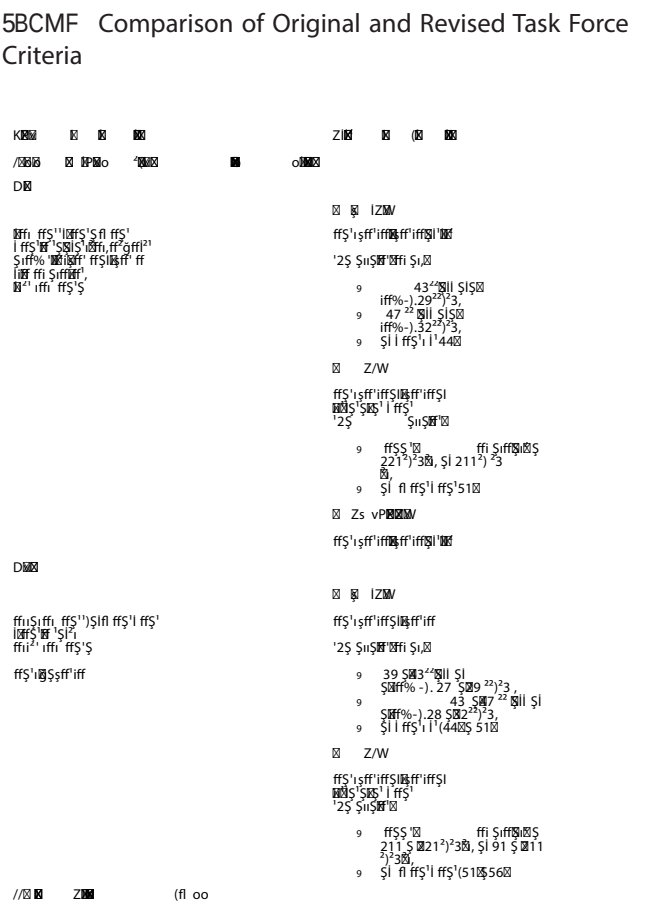


Fig-1 Echocardiography of RV

International Task Force proposed criteria for the clinical diagnosis of ARVD/C, based on structural, electrocardiographic, arrhythmic, histological and familial characteristics of ARVD/C. On the role of emerging diagnostic modalities and advances in the genetics of ARVC/D, and although 1994 criteria were highly specific, but they lacked sensitivity for early and familial disease, Marcus et al revised the task force (6). Comparison between the Original and Revised Task Force Criteria is shown in the (**Table-1)**.

5BCMF Comparison of Original and Revised Task Force Criteria



In the early stage of the disease, structural changes may be absent or subtle and confined to a localized region of the RV, typically the inflow tract, outflow tract, or apex of the RV, the “triangle of dysplasia (7).” Progression to more diffuse RV disease and left ventricular (LV) involvement, typically affecting the posterior lateral wall, is common (8). In the early “concealed phase,” individuals are often asymptomatic but may nonetheless be at risk of sudden cardiac death, notably during exertion.Later, diffuse disease may result in biventricular heart failure, whereas ventricular arrhythmias may or may not be present. The ultimate phenotype may resemble dilated cardiomyopathy. Clinical manifestations vary with age and stage of disease (9).

Although being in autosomal dominant inheritance mostly, there are recessive forms (eg, Naxos disease, Carvajal syndrome) that are associated with cutaneous phenotype. Desmosomal variations lead to impairment of cell-to-cell binding. Seven genes have beenidentified that are associated with ARVC/D: plakoglobin (JUP), desmoplakin (DSP), plakop-hilin-2(PKP2), desmoglein-2 (DSG2), desmocol-lin-2 (DSC2), transforming growth factor beta-3 (TGF_3), and TMEM43.20 (10,11,12)

NIH registry showed that the mean age of di-agnosis of ARVC/D was 38±14 years with male predominance and T wave inversion beyond V was present in 56% of newly diagnosed patients. In the first detailed clinical profile of this disease,6 T wave inversion in V1 to V4 was found in 86% (19/22 patients) in contrast to an inciEFODFPG

of T wave inversion in V1 to V3 of newly diagnosed patients in the registry stated above(13). For this purpose Jain et al evaluate one hundred patients with ARVD and detected 17 patients with RBBB, 15 patients with IRBBB. T wave inversion through V3 demonstrated optimal sensitivity and specificity in both ARVD patients without a complete RBBB or incomplete RBBB.In this way, to identify patients with ARVD, they have made a chart summarizing an algorithm that can be used of an IRBBB or CRBBB (**Figure-2**) (13).

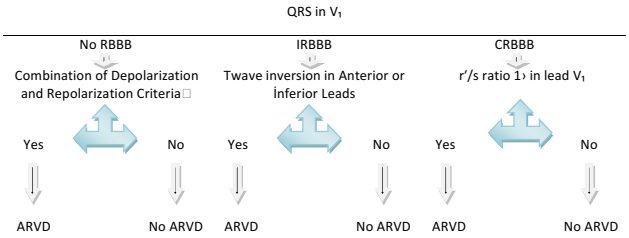


Figure-2; Electrocardiographic Evaluation for ARVD

Definite diagnosis is only possible after a comprehensive evaluation that includes evaluation of the family history, the structure and function of the RV, and screening for arrhythmias. There are two primary goals of treatment of ARVD/C; to reduce the frequency and severity of ventricular arrhythmias and to prevent or limit the worsening of ventricular function and heart failure. The proposed modifications of the original Task Force criteria represent a working framework to improve the diagnosis and management of ARVC/D. Awareness is growing that ARVC/D as such is the most well recognized form of a broad disease spectrum that includes left-dominant and biventricular subtypes. Lack of specific diagnostic guidelines contributes to under recognition of non-classic disease. Future revisions of the Task Force criteria may fill this gap.

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