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 sağventrikül displazisi (ARVD) nadir görülen bir kardiyomiyopatıdır. 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 sağventrikül (SV) dilatasyonu ve apikal anevrizma tipik bulgusudur. Fakat ARVD olgularında intraventriküler trombüs çok nadir görülmektedir. 19 yaşında erkek hasta, hastanemize çarpıntı ve bayılma şikayetleri ile başvurdu. Elektrokardiografisinde ön yüz derivasyonlarda T negatifliği bulunmaktaydı. Ekokardiyografide sağventrikül (SV) dilate ve SV aksinde anevrizma oluşum içinde trombüs görüldü (Fig-1). Kardiyak manyetik rezonans görüntüleme yöntemi ile sağventrikül genişlemesi, yağ infiltrasyonu, fibrofatty dokuları, SV duvar hareket bozukluğu ve trombüsün apikal anevrizma doğrulandı. Antikoagulasyon tedavisiyle üç ay sonra trombüsün rezole olduğu gözlemdi ve ICD implante edildi. ARVD tanısında elektrokardiografik, aritmik, histolojik ve ailesel özellikleri yanında görüntüleme yöntemleri de büyük önem taşımaktadır. Sağventrikül dilatasyonu ve apikal anevrizma tani sürecinde önemli kriterler olmakla birlikte bu gibi bulguları saptadığında trombus varlığı da dikkatlice değerlendirilmelidir.

ANAHTAR KELİMELER: Aritmojenik Sağ Ventrikül Displazisi, Fibrofatty Infiltrasyon, Trombus

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 rezonsans imaging confirmed RV enlargement, fatty infiltration, fibrosis, wall motion abnormalities and apical aneurysm with thrombus. Anticoagulation therapy commenced to the patient. After three months later thrombus resolved 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
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 aneurysym 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.

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).

Fig-1 Echocardiography of RV
Table 1: Comparison of Original and Revised Task Force Criteria

<table>
<thead>
<tr>
<th>Original task force criteria</th>
<th>Revised task force criteria</th>
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<tr>
<td>I. Global or regional dysfunction and structural abnormalities*</td>
<td><strong>Major</strong></td>
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<tr>
<td>Severe dilatation and reduction of RV ejection fraction with no or only mild LV impairment. Localized RV aneurysms (akinetic or dyskinetic areas with diasstolic bulging)</td>
<td><strong>Major</strong></td>
</tr>
<tr>
<td>Severe segmental dilatation of the RV</td>
<td><strong>Minor</strong></td>
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*Restrictive criteria:
- PLAK A53 V to 32 mm (corrected for body size [PLAK/BAS] 16 to 39 mm/2)
- PLAK RVOT 20 to 36 mm (corrected for body size [PLAK/BAS] 20 to 42 mm/2)
- or Fractional area change 35% to 60%

**By ECG:**
- Regional RV akinesia, dyskinesia, or aneurysm
- and 1 of the following (end diastole):
  - PLAK RVOT 33 mm (corrected for body size [PLAK/BAS] 19 mm/2)
  - PLAK RVOT 36 mm (corrected for body size [PLAK/BAS] 21 mm/2)
  - or Fractional area change 35% to 60%

**By MRI:**
- Regional RV akinesia or dyskinesia or dysynchronous RV contraction and 1 of the following:
  - Ratio of RV end-diastolic volume to BSA 110 ml/m2 (male) or 100 ml/m2 (female)
  - or RV systolic fraction 40% to 45%

**By RV angiography:**
- Regional RV akinesia, dyskinesia, or aneurysm

II. Tissue characterization of wall

**Major**
- Fibrofatty replacement of myocardium on endomyocardial biopsy
- Residual myocytes <10% by morphometric analysis (or <30% if estimated), with fibrous replacement of the RV free wall myocardium in more than 1 sample, with or without fatty replacement of tissue on endomyocardial biopsy

**Minor**
- Residual myocytes 60% to 75% by morphometric analysis (or <60% if estimated), with fibrous replacement of the RV free wall myocardium in more than 1 sample, with or without fatty replacement of tissue on endomyocardial biopsy

**III. Repolarization abnormalities**

**Major**
- Inverted T waves in right precordial leads (V1, V2, and V3) or beyond in individuals >14 years at the time of diagnosis of complete right bundle-branch block BQS 110 mm²

**Minor**
- Inverted T waves in right precordial leads (V2 and V3) (people aged 12 years, in absence of right bundle-branch block)

**IV. Depolarization/conduction abnormalities**

**Major**
- Epsilon wave with reproducible low-amplitude signals between end of QRS complex to onset of the T wave in right precordial leads (V1 to V3)

**Minor**
- Late potentials (SAECG)

**V. Arrhythmia**

**Major**
- Nonsustained or sustained ventricular tachycardia of left bundle-branch morphology with superior axis (negative or indeterminate QRS in leads II, I, and aVF) and positive in lead aV1

**Minor**
- Left bundle-branch block-type ventricular tachycardia (sustained and nonsustained) (JEG, bundle-branch block morphology with inferior axis (positive QRS in leads II, I, and aVF) and negative in lead aV1) or of unknown axis 500 ventricular extrasystoles per 24 hours (Holter)

Holler, exercise, frequent ventricular extrasystoles (1000 per 24 hours (Holter))

**Familial disease confirmed at necropsy or surgery**

**ARVC/D confirmed in a first-degree relative who meets current Task Force criteria**

**ARR/C/D confirmed pathologically at autopsy or surgery in a first-degree relative**

**Identification of a pathogenic mutation**

**Autosomal dominant inheritance**

**Arrhythmias**

Premature sudden death (<35 years of age) due to suspected ARVC/D

Familial history (clinical diagnosis based on present criteria) ARVC/D confirmed pathologically or by current Task Force criteria in second-degree relative

Short-waves: e.g., LV ejection fraction <50%, RV ejection fraction <40%, regional RV hyperkinesia, dyskinesia or aneurysm

**By 2D echo:**
- Regional RV akinesia, dyskinesia, or aneurysm
- and 1 of the following (end diastole):
  - PLAK RVOT 33 mm (corrected for body size [PLAK/BAS] 19 mm/2)
  - PLAK RVOT 36 mm (corrected for body size [PLAK/BAS] 21 mm/2)
  - or Fractional area change 35% to 60%

**By MRI:**
- Regional RV akinesia or dyskinesia or dysynchronous RV contraction and 1 of the following:
  - Ratio of RV end-diastolic volume to BSA 110 ml/m2 (male) or 100 ml/m2 (female)
  - or RV systolic fraction 40% to 45%

III. Tissue characterization of wall

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- Fibrofatty replacement of myocardium on endomyocardial biopsy
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- Epsilon wave with reproducible low-amplitude signals between end of QRS complex to onset of the T wave in right precordial leads (V1 to V3)

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- Nonsustained or sustained ventricular tachycardia of left bundle-branch morphology with superior axis (negative or indeterminate QRS in leads II, I, and aVF) and positive in lead aV1

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- Left bundle-branch block-type ventricular tachycardia (sustained and nonsustained) (JEG, bundle-branch block morphology with inferior axis (positive QRS in leads II, I, and aVF) and negative in lead aV1) or of unknown axis 500 ventricular extrasystoles per 24 hours (Holter)
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.

**REFERENCES**


