Case Report / Olgu Sunumu
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Yamaguchi Syndrome: A Difficult Diagnosis in the Differential Diagnosis of Acute Coronary Syndrome

Yamaguchi Sendromu: Akut Koroner Sendrom Ayırıcı Tanısında Zor Bir Tanı

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ABSTRACT

Apical hypertrophic cardiomyopathy (ApHCM) (Yamaguchi Syndrome) with hypertrophy of the ventricular apex constitutes 8% of the hypertrophic cardiomyopathies (HCMs). ApHCM can cause ventricular malignant dysrhythmias, atrial fibrillation, and ischemic chest pain. Definitive diagnosis is made by electrocardiography (ECG) and transthoracic echocardiography. A 73-year-old male patient was admitted to the emergency department with chest pain. The patient's vital signs were within the normal range. In the ECG, there were giant negative T wave in leads V4-5-6, 0.5 mm ST segment depression, and left ventricular hypertrophy in the inferior derivations. The left ventricular apex thickness was measured as 14 mm (reference range: 6-11). Although the HEART score was 4, the preliminary diagnosis of the patient was determined as ApHCM. Beta-blocker and antiplatelet therapy were started. The mortality and morbidity rates of ApHCM are higher among HCMs. Clinicians should be aware of such ECG and echocardiography findings to prevent possible morbidity and mortality.
Keywords: Apical; hypertrophic cardiomyopathy; Yamaguchi syndrome; acute coronary syndrome; echocardiography.

ÖZ

Ventriküler apeksin hipertrofisi ile seyreden apikal hipertrofik kardiyomiyopati (ApHKM) (Yamaguchi Sendromu) hipertrofik kardiyomiyopati (HKM)'lerin %8'ini oluşturmaktadır. ApHKM ventriküler malign disritmilere, atriyal fibrilasyona ve iskemik göğüs ağrısına neden olabilir. Kesin tanı elektrokardiyografi (EKG) ve transtorasik ekokardiyografi ile konur. 73 yaşında bir erkek hasta göğüs ağrısı şikayetiyle acil servise başvurdu. Hastanın vital bulguları normal sınırlardaydı. EKG'de V4-5-6 derivasyonlarında dev negatif T dalgası, 0,5 mm ST segment depresyonu ve inferior derivasyonlarda sol ventrikül hipertrofisi vardı. Sol ventrikül apeks kalınlığı 14 mm (referans aralığı: 6-11) olarak ölçüldü. HEART skoru 4 olmasına rağmen hastanın ön tanısı ApHKM olarak belirlendi. Beta-bloker ve antiplatelet tedavi başlandı. HKM'ler arasında ApHKM'nin mortalite oranları daha yüksektir. Klinisyenler olası morbidite ve mortaliteyi önlemek için bu tür EKG ve ekokardiyografi bulgularının farkında olmalıdır.
Anahtar kelimeler: Apikal; hipertrofik kardiyomiyopati; Yamaguchi sendromu; akut koroner sendrom; ekokardiyografi.

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INTRODUCTION

Hypertrophic cardiomyopathy (HCM) is an autosomal dominant disease that occurs as a result of mutation of sarcomere proteins in the myocardium (1). Unlike classical HCM, cardiomyopathies with hypertrophy of the ventricular apex are called apical HCM (ApHCM). ApHCM constitutes 8% of all HCMs (2). In the Asian population, up

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to 25% of all HCMs are ApHCM, while it is 1-10% in the non-Asian population (3). ApHCM's typical features were first described by Sakamoto and Yamaguchi in 1976 (2). ApHCM, also known as Yamaguchi Syndrome, is clinically associated with widespread giant negative T waves (≥1 mV, ≥10 mm) and high voltage QRS complex in the precordial derivations (4). In definitive diagnosis, the apical wall thickness is expected to be ≥15 mm and the ratio of maximal apical wall thickness to the posterior wall thickness is expected to be ≥1.5 (2). In recent studies, apical wall thickness ≥13 mm was accepted as a diagnostic criterion (5). Its echocardiographic findings are hypertrophy in the apical myocardium and a "spadelike" configuration of the left ventricular cavity at the end of diastole (5). Compared with classical HCM, ApHCM causes atrial fibrillation more frequently and the sudden cardiac death rate is higher, 0.5-4% vs 1.3% (3,6). For this reason, early diagnosis of ApHCM will prevent possible mortality and morbidity. ApHCM has different clinical presentations. The most common symptom is chest pain. It causes an acute coronary syndrome-like condition in which there is T wave negativity in the precordial derivations accompanied by chest pain. In the preliminary diagnosis of ApHCM, a thorough evaluation of the patient by a cardiologist and a comprehensive echocardiography can confirm the diagnosis and prevent potential morbidity and mortality.

CASE REPORT

A 73-year-old male was admitted to the emergency department with the complaint of intermittent chest pain that started 3 hours before admission and the pain was increased with breathing. The patient did not have any accompanying disease in his past medical history and had a 60-pack-year smoking history. The vital signs of the patient were in the normal range at admission (blood pressure: 140/80 mm Hg, pulse: 75 beats/min, respiratory rate: 20/min, temperature: 36.7 ºC, saturation: 94%). There was not any pathological finding in the cardiac examination of the patient. In 12-lead electrocardiography (ECG), there were signs of giant negative T wave in leads V4-5-6, 0.5 mm ST segment depression, and left ventricular hypertrophy in the inferior derivations (Figure 1). The serum troponin I values at the beginning, 3rd and 6th hour were 11.1, 10.1, and 15.6 ng/L (reference range: 14-42.9), and the CK-MB values were 2.7, 2.8, 2.5 µg/L (reference range: 0.6-6.3), respectively. The HEART score was calculated as 4 (History:0, ECG:1, Age:2, Risk factors:1, Troponin:0). In bedside echocardiography performed by an emergency physician, left ventricular apex thickness was measured as 14 mm (reference range: 6-11) and the ejection fraction was within normal values. (Figure 2). The ECG and echocardiography findings were found to be significant in terms of "Yamaguchi Syndrome". The patient was consulted by the cardiologist. Antiplatelet and beta-blocker therapy was started as a medical treatment and medical follow-up was recommended by the cardiologist. During the outpatient follow up cardiac magnetic resonance imaging was performed, and it revealed hypertrophy at the apex of the left ventricle, and the diagnosis was confirmed (Figure 3).

The patient consented to the use of his medical data for scientific purposes.

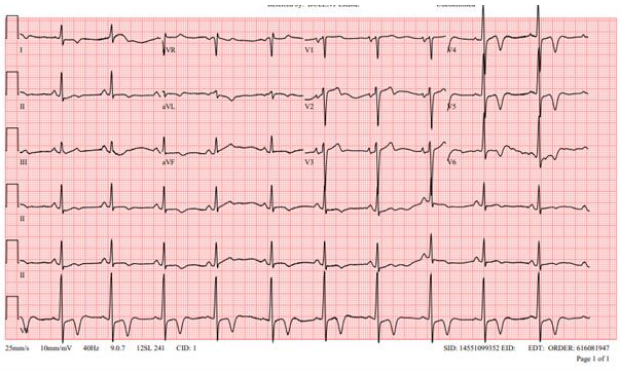

Figure 1. Giant negative T waves in Yamaguchi syndrome

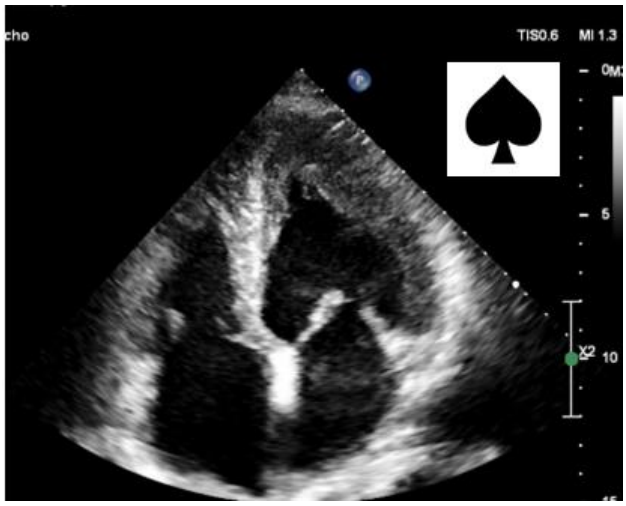

Figure 2. "spadelike" configuration of the left ventriculi in Yamaguchi syndrome

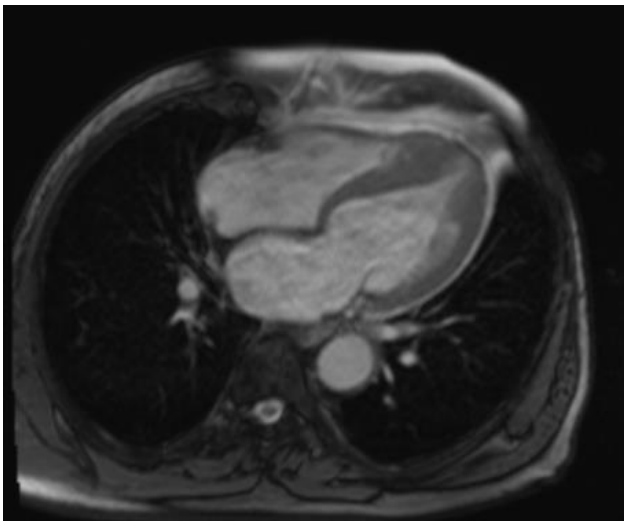

Figure 3. Magnetic resonance imaging of Yamaguchi syndrome

DISCUSSION

In contrast to the diffuse hypertrophy seen in HCM, wall thickening in ApHCM is located distal to the papillary muscle and occurs at the apex. It has been shown that the mortality and morbidity rates of ApHCM are higher than the other HCMs and it causes death more frequently,

especially in female patients (3,7,8). It may cause especially malignant ventricular dysrhythmias and apical aneurysms (8).

Continuation of contraction of the apical region in early-mid diastole in ApHCM can be seen as the cause of small vessel occlusion (9). Small vessel occlusions cause regional perfusion defects and chest pain occurs. Increased myocardial tissue, small vessel disease, and impaired vasodilator reserve have been suggested as possible causes of myocardial ischemia. T wave inversion (93%), giant T wave negativity (47%) and LVH (65%) accompanying ApHCM clinical findings cause cases to be confused with acute coronary syndrome (4). However, in most cases, coronary arteries are found to be normal on coronary angiography. Patients who had early repolarization in the anterolateral derivations in previous ECGs should be carefully evaluated for the diagnosis of ApHCM. Transthoracic echocardiography is the first choice for the evaluation of these patients. However, bedside echocardiography is not always sufficient due to the difficult evaluation of the apical region, artifacts, and poor echogenicity. In such cases, it is necessary to resort to other diagnostic methods such as magnetic resonance imaging and nuclear scintigraphy (10,11).

CONCLUSION

Apical-type hypertrophic cardiomyopathy, which can mimic acute coronary syndrome, is often miss diagnosed or undiagnosed, as it is a rare case, especially in non-Asian populations. When a giant negative T wave in the anterior derivations and signs of left ventricular hypertrophy are seen in the ECG, especially in low-risk patients for acute coronary syndrome, it should definitely be considered as a differential diagnosis and early referral should be made. Thus, some complications such as malignant ventricular dysrhythmias, myocardial infarction, and sudden cardiac death that may occur due to ApHCM (Yamaguchi Syndrome) can be prevented.

Informed Consent: Written informed consent was obtained from the patient for publication and accompanying images.

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