Turk Gogus Kalp Dama 2015;23(3):549-551
doi: 10.5606/tgkdc.dergisi.2015.10929

Apical hypertrophic cardiomyopathy in childhood: a very rare variant

Çocuklukta apikal hipertrofik kardiyomiyopati: çok nadir bir değişken

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ABSTRACT

Apical hypertrophic cardiomyopathy is a rare form of hypertrophic cardiomyopathy which classically involves the apex of the left ventricle. Apical hypertrophic cardiomyopathy can be an incidental finding, or patients may present with chest pain, palpitations, dyspnea, and syncope. In this article, we describe the clinical presentation of a seven-year-old girl who was asymptomatic, presented for cardiac evaluation and was detected to have an apical variant of hypertrophic cardiomyopathy.

Keywords: Apical; cardiomyopathy; child; hypertrophic.

ÖZ

Apikal hipertrofik kardiyomiyopati klasik olarak sol ventrikülün apeksini tutan nadir bir hipertrofik kardiyomiyopati türüdür. Apikal hipertrofik kardiyomiyopati rastlantısal bir bulgu olabilir ya da hastalar göğüs ağrısı, çarpıntı, dispne ve senkop ile başvurabilir. Bu yazıda asemptomatik olan, kardiyak değerlendirme için başvuran ve hipertrofik kardiyomiyopatinin apikal bir değişkeni olduğu tespit edilen yedi yaşında bir kızın klinik görünümü tanımlandı.

Anahtar sözcükler: Apikal; kardiyomiyopati; çocuk; hipertrofik.

Apical hypertrophic cardiomyopathy (AHCM) is a rare form of HCM in which the myocardial hypertrophy predominantly involves the apex of the left ventricle. It is characterized by giant negative T-waves on electrocardiography (ECG) and a spade-shaped left ventricle (LV) cavity.[1] Although typically diagnosed in middle-aged individuals, a few pediatric cases have been reported in the literature.[2] Herein, we present a child with AHCM incidentally diagnosed via ECG and echocardiography during a cardiovascular examination.

CASE REPORT

A seven-year-old girl was referred to our hospital for further evaluation because of cardiomegaly on a telecardiogram. The patient had no family history of cardiac disease and no complaints.

A physical examination performed after her referral was normal, with nothing out of the ordinary being seen with regard to the complete blood count (CBC), biochemical parameters, or cardiac troponin I levels. The ECG showed negative T-waves in the V1-V6 leads and slight ST depression in the V4-V6 leads (Figure 1) while the echocardiography detected myocardial hypertrophy that was mostly confined to the apical portion of the LV, resulting in a spade-like configuration of the LV cavity at end-diastole (Figure 2). In addition, the systolic function of the LV was normal (fractional shortening 41%), and the LV dimension at the basal level had increased to 43 mm. Color echocardiography revealed mild mitral regurgitation but no intracavitary LV obstruction. The end-diastolic thickness of the LV apical-free wall and the basal-free wall were 15 mm and 8.3 mm, respectively while the end-diastolic thickness of the apical interventricular septum (IVS) and basal IVS were 16 mm and 7.2 mm, respectively. Furthermore, the maximal apical to posterior wall thickness ratio was 1.8. The patient was also evaluated via a 24-hour Holter monitor, and this yielded normal results.

Available online at
www.tgkdc.dergisi.org
doi: 10.5606/tgkdc.dergisi.2015.10929
QR (Quick Response) Code

Received: October 09, 2014 *Accepted:* January 16, 2015

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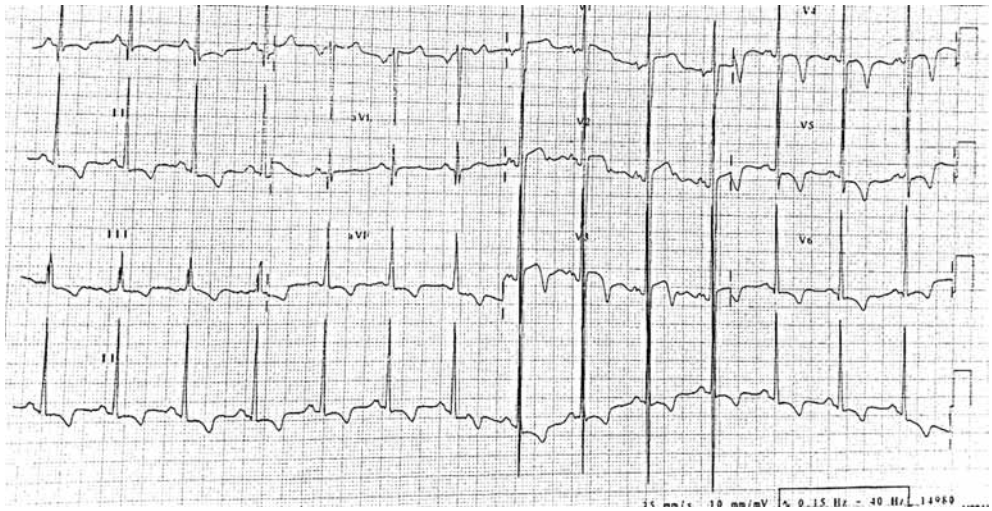

Figure 1. The patient's ECG showing the negative T-waves in the V1-V6 leads.

DISCUSSION

Historically, AHCM was thought to be confined to the Japanese population, but it can also be found in other countries as well. When all of the HCM patients in Japan were examined, the prevalence rate for AHCM was 15%, whereas in the United States, the prevalence rate was only 3%.[3] Apical hypertrophic cardiomyopathy occurs sporadically; however, a few families have been reported to have autosomal dominant inheritance.[4] Furthermore, sarcomere protein gene mutations are present in up to 30% of this specific patient population.[4] Moreover, a few sarcomere protein gene defects (ACTC, pGlu101Lys) have been reported in the apical HCM phenotype.[4] Our patient had no family history of cardiac disease, so it most likely was a sporadic case. The mean age of AHCM presentation is 41.4±14.5 years, and it is most frequently seen in males.[3] Additionally, approximately 54% of patients with AHCM are symptomatic with chest pain followed by palpitations, dyspnea, and syncope occurring the most.[3] Physical findings of an audible/palpable fourth heart sound and a new murmur are also common. Moreover, Arad et al.[4] reported that giant negative T-waves in the precordial leads were detected in 93% of these patients via ECG. Our patient was asymptomatic and had no physical findings, but the ECG showed negative T-waves in the precordial leads and slight ST depression in the V4-V6 leads.

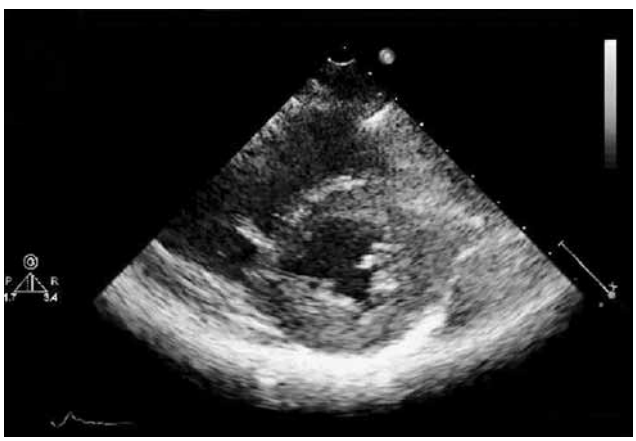

Figure 2. Transthoracic echocardiography showing the parasternal short-axis view.

The diagnosis of AHCM is normally made via either two-dimensional echocardiography or magnetic resonance imaging (MRI) and is based on the presence of an otherwise unexplained asymmetric hypertrophy of the LV that is confined predominantly to the LV apex with a maximal apical wall thickness of ≥15 mm and a maximal apical to posterior wall thickness of ≥1.5.[5] In borderline cases with a maximal wall thickness of between 13 and 15 mm, concomitant ECG changes are required to establish the diagnosis of AHCM.[6]

Apical hypertrophic cardiomyopathy can further be subdivided into two groups: the "pure apical" form with isolated apical hypertrophy which is limited to the LV apex below the papillary muscle level and the "distal dominant" form in which the apical hypertrophy extends to other segments, including the IVS and the LV anterior and posterior walls.[7] Our patient had the "pure apical" form of AHCM.

In contrast to the other variants of HCM, the prognosis of AHCM is comparatively benign. Eriksson et al.[8] reported an overall mortality rate of 10.5% for these patients while the cardiovascular mortality

rate was 1.9% after a follow-up period of 13.6±8.3 years. The "pure-apical" form of AHCM is generally associated with a benign clinical manifestation, whereas the outlook for patients with the "distal-dominant" form is not as positive and more closely resembles the clinical manifestation of typical HCM.[7] The study by Miyamoto et al.[2] featured two children with AHCM, and both remained asymptomatic for more than 15 years. Our patient had no complaints during her one-year follow-up period.

Conclusion

Apical hypertrophic cardiomyopathy may present in childhood with negative T-waves in precordial derivations and typical echocardiography findings. Although the prognosis for this disease is favorable, long-term follow-up is needed to monitor these patients for the presence of arrhythmia.

Declaration of conflicting interests

The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

Funding

The authors received no financial support for the research and/or authorship of this article.

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