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CASE REPORT

# Isolated papillary muscle hypertrophy: A gap in our knowledge of hypertrophic cardiomyopathy?☆

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**Abstract** Increased thickness of left ventricular walls is the predominant characteristic and one of the diagnostic criteria of hypertrophic cardiomyopathy (HCM). This case illustrates an uncommon but important finding of isolated hypertrophy of the papillary muscles (PMs), observed in a young woman in whom an abnormal electrocardiogram was initially detected. During the investigation isolated PM hypertrophy was identified. The structural characteristics of the PMs have received scant attention in this setting and there is little information in the literature on this entity, whose real prevalence and clinical significance remain to be determined. The available information relates solitary PM hypertrophy with an early form or a different pattern of HCM. In this case PM hypertrophy was only detected due to the finding of an abnormal electrocardiogram, which prompted further diagnostic tests and a search for possible etiologies.


**PALAVRAS-CHAVE**
Hipertrofia dos músculos papilares;
Miocardiopatia hipertrófica;
Eletrocardiograma;
Ecocardiografia;

**Hipertrofia isolada dos músculos papilares: uma lacuna na miocardiopatia hipertrófica?**

**Resumo** O aumento da espessura das paredes do ventrículo esquerdo é a característica predominante e um dos critérios de diagnóstico da miocardiopatia hipertrófica (MH). Este caso relata um achado incomum, mas importante, de hipertrofia isolada dos músculos papilares (MP), observada numa mulher jovem a quem foi inicialmente detetado um eletrocardiograma anormal. Durante a investigação realizada foi identificada uma hipertrofia isolada dos MP. As características estruturais dos MP têm recebido pouca atenção neste contexto.

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Ressonância magnética cardíaca

Existe insuficiente informação na literatura sobre esta entidade, cuja prevalência e relevância clínica se encontram por determinar, mas a informação disponível relaciona a hipertrofia isolada dos MP com uma forma precoce ou um padrão diferente de MH. Neste caso, a hipertrofia dos MP só terá sido detetada, provavelmente, devido ao achado de um eletrocardiograma anormal, o que orientou para a realização de exames complementares de diagnóstico adicionais e para a procura de possíveis etiologias.



## Introduction

The clinical diagnosis of hypertrophic cardiomyopathy (HCM) is conventionally made with cardiac imaging, at present most frequently two-dimensional echocardiography, but the use of cardiac magnetic resonance (CMR) imaging is increasing. According to the 2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy, the morphologic diagnosis of HCM is based on the presence of a hypertrophied and nondilated left ventricle in the absence of another cardiac or systemic disease capable of producing the magnitude of that hypertrophy (usually $\geq$15 mm in adults or the equivalent relative to body surface area in children). There are also patients who are genotypically positive but phenotypically negative.[1] The cardiac phenotype of HCM shows great diversity in the degree and pattern of hypertrophy (asymmetric, concentric, or apical), age of onset and clinical course.[2] Although the papillary muscles (PMs) are an anatomic part of the left ventricular (LV) chamber, the significance and diverse morphology of these structures in HCM has not been systematically characterized.[3] Papillary muscle (PM) hypertrophy is a rare echocardiographic finding, with very few cases reported in the literature.[4] In an analysis of 6731 echocardiographic studies, Kobashi and colleagues[5] found 29 patients with solitary PM hypertrophy, defined as either the vertical or horizontal diameter of at least one of the two PMs more than 11 mm (measured in end-diastole). Some of these patients presented electrocardiographic findings, such as high left precordial voltage and inverted T waves, very similar to those of apical hypertrophy. There are reports in the literature suggesting that PM hypertrophy, especially of the posteromedial muscle, could play an important role in the development of negative precordial T waves.[5,6] There are also a few cases described of a diagnosis of HCM without hypertrophy at necropsy after sudden death, but the PMs were not described in the reports of these patients, so there is no information about the possible presence of hypertrophy of these structures.[7] Therefore, solitary PM hypertrophy can have clinically important implications for the screening of HCM as a newly identified subtype of or an early form of HCM.[5,8,9]

CMR provides complete tomographic imaging of the heart with high spatial resolution images, and is thus an excellent imaging method to assess the PMs with precision.[3] It is also a useful tool for further investigation as an established exam for assessment of different types of cardiomyopathies, since there are some typical findings in this exam that may suggest a particular pathology or etiology.[10]

## Case report

A 19-year-old woman presented palpitations and chest discomfort, unrelated to exertion and with no pleuritic characteristics, without reports of syncope, during the previous year. She had no relevant medical history and no known relevant family history of cardiac, renal, neurologic or genetic diseases; she was not under any medication and did not practice any sports. Her family physician performed a physical exam that was unremarkable (including blood pressure), except for an arrhythmia on cardiac auscultation. He requested an electrocardiogram (Figure 1A) which revealed sinus arrhythmia, biphasic T waves in leads DII, aVF and V3, and T-wave inversion in leads DIII and V4–V6. On the basis of this abnormal ECG clinical observation by a cardiologist was requested, in which the patient underwent more diagnostic exams. The echocardiogram revealed no significant abnormalities, except the presence of hypertrophic PMs (Figure 1B); LV mass was normal and maximum ventricular wall thickness was 11 mm at the interventricular septum in parasternal long-axis view. There were no significant valvular abnormalities. Despite the presence of prominent PMs, no significant intraventricular gradient or LV outflow tract obstruction was found on Doppler evaluation at rest. For further clarification and to assist with differential diagnosis, CMR imaging was performed. This exam showed a normal right ventricle and normal valve structures. The left ventricle had normal systolic function (60% ejection fraction), with a mass of 59 grams (35.5 g/m$^2$), end-systolic volume of 42 ml and end-diastolic volume of 104 ml. Maximum ventricular wall thickness was 11.2 mm at the interventricular septum measured in end-diastole, 4-chamber view. Both PMs were also seen to be abnormally hypertrophic (Figures 2 and 3), occupying a large part of the LV cavity during systole (Figures 2A and 3A). The anterolateral PM had a maximum diameter of 12.5 mm and the posteromedial PM measured 15 mm on the horizontal axis (end-diastole, short-axis view; Figure 3B). Delayed hyperenhancement was not observed.

Laboratory tests were performed, but the results were unremarkable (including complete blood count, liver and kidney function tests, serum levels of muscle enzymes and urine analysis). In exercise stress testing the patient