

# Quadricuspid Mitral Valve in Hypertrophic Cardiomyopathy



Bebiana Faria, MD, Mário Lourenço, MD, Olga Azevedo, MD, Marina Fernandes, MD, Pedro von Hafe, MD, Francisco Sousa, MD, and António Lourenço, MD, *Guimarães, Portugal*

## INTRODUCTION

The prevalence of mitral valve (MV) abnormalities in hypertrophic cardiomyopathy (HCM) is unclear, because they are often poorly appreciated and/or not documented. A quadricuspid MV is a very rare cardiac congenital malformation. We report the clinical case of a patient with HCM and a quadricuspid MV identified on echocardiography and confirmed on cardiac magnetic resonance (CMR) imaging.

## CASE PRESENTATION

A 59-year-old man with heart failure was referred to the echocardiography laboratory to clarify the mechanism and quantify the severity of MV regurgitation. In 1994, at the age of 35 years, transthoracic echocardiography had revealed HCM associated with significant left atrial (LA) dilation and mild to moderate MV regurgitation. During follow-up, atrial fibrillation was diagnosed at the age of 41 years, and transthoracic echocardiography at that time showed progressive worsening of the severity of MV regurgitation and LA dimension. In 2018, at the age of 59 years, the patient was referred for a cardiomyopathy consultation. He was in New York Heart Association functional class II, and a physical examination showed a grade III/VI systolic murmur in the mitral focus. Electrocardiography showed atrial fibrillation and left bundle branch block. The patient had no other known comorbidities besides hypertension, dyslipidemia, and smoking. Family history of sudden death was recorded in a 61-year-old brother.

Two-dimensional transthoracic echocardiography revealed aneurysmal LA dilation (LA volume index  $400 \text{ ml/m}^2$ ) and a normal-sized left ventricle with concentric severe hypertrophy (left ventricular mass  $219 \text{ g/m}^2$ ) without obstruction at rest and preserved ejection fraction. A quadricuspid MV was observed, with quadrangular mitral annular geometry and poor leaflet coaptation, leading to severe MV regurgitation. It was a central regurgitation jet that reached the LA roof, with effective regurgitant orifice area of  $0.45 \text{ cm}^2$  and regurgitant volume of 71 ml (Figures 1A, 1B, and 2, Video 1). An abnormal insertion of the papillary muscles, with at least one supernumerary papillary muscle originating in the apical lateral region of the left ventricle and apparently inserting on the MV leaflet, was also shown (Figures 1C and 1D). Three-dimensional transthoracic and transesophageal echo-

## VIDEO HIGHLIGHTS

**Video 1:** Transthoracic echocardiography, showing aneurysmal LA dilatation, concentric severe left ventricular hypertrophy, and a deformed MV with mitral regurgitation. It is also possible to observe accessory papillary muscle and quadrangular mitral annulus with four MV commissures leading a square-like mitral opening in diastole and systolic closing in a cross shape.

**Video 2:** Three-dimensional transthoracic echocardiography, from a ventricular view, showing four mobile and similarly sized leaflets, four commissures, a square-like opening in diastole, and a cross shape of the commissures during systolic closure.

**Video 3:** Cardiac magnetic resonance imaging, basal short-axis, showing MV with quadrangular annular geometry, a square-like opening pattern, and a symmetric cross shape of the commissures during systolic closure.

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cardiography confirmed the suspicion of a quadricuspid MV, showing four MV commissures, a square-like mitral opening in diastole, and four mobile and similarly sized leaflets, closing in a cross shape in systole (Figures 3A and 3B, Video 2). CMR, although technically limited by permanent atrial fibrillation, also revealed quadrangular geometry of the mitral annulus with a square-like opening pattern and symmetric cross-closure of the four MV leaflets (Figures 4A and 4B, Video 3). Results of testing with a panel of 28 HCM genes were negative. Findings on echocardiography for HCM screening of the 28-year-old son were normal.

Results of analytic study, including free light-chain assay, serum and urine electrophoresis and immunoelectrophoresis, and dried blood spot test for Fabry disease, were negative.

According to the HCM Risk-SCD score, the patient had a 5-year risk for sudden cardiac death of 9.44%, and therefore a cardioverter-defibrillator for primary prevention was implanted in 2018.

The patient was referred for cardiac surgery for MV replacement and LA reconstruction. While waiting for elective surgery, he had pneumonia with severe acute respiratory distress requiring intensive care unit admission. During intensive care unit stay, he had a rupture of a previously unknown abdominal aortic aneurysm and died. An autopsy was not performed.

## DISCUSSION

To the best of our knowledge, this is the second case report of HCM associated with quadricuspid MV.

From Hospital Senhora da Oliveira, Guimarães, Portugal.

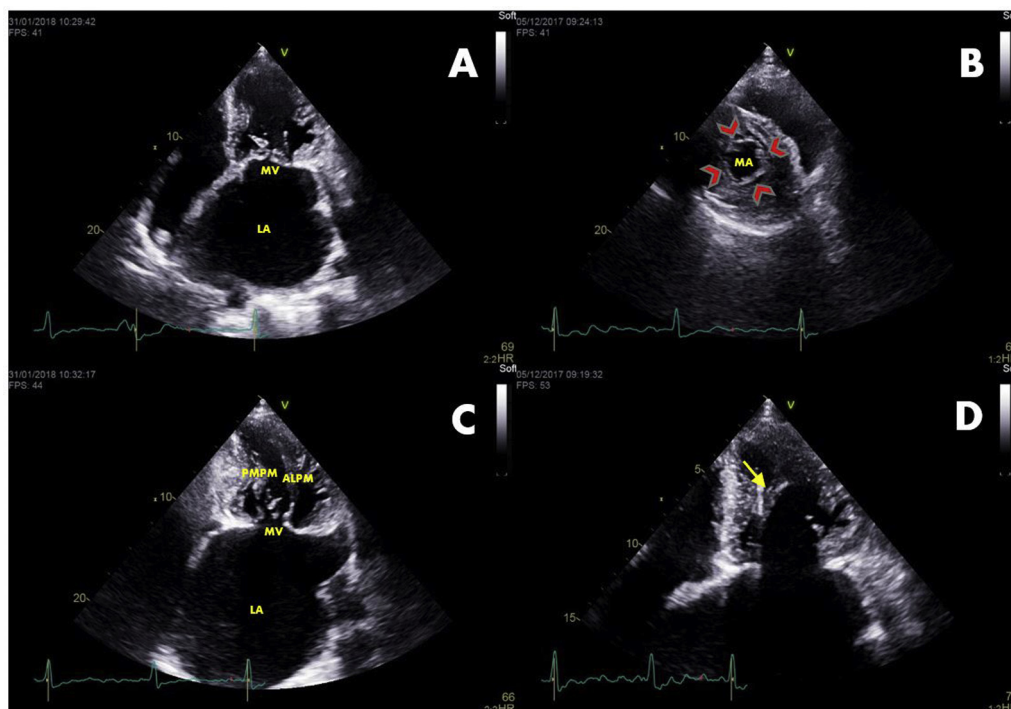
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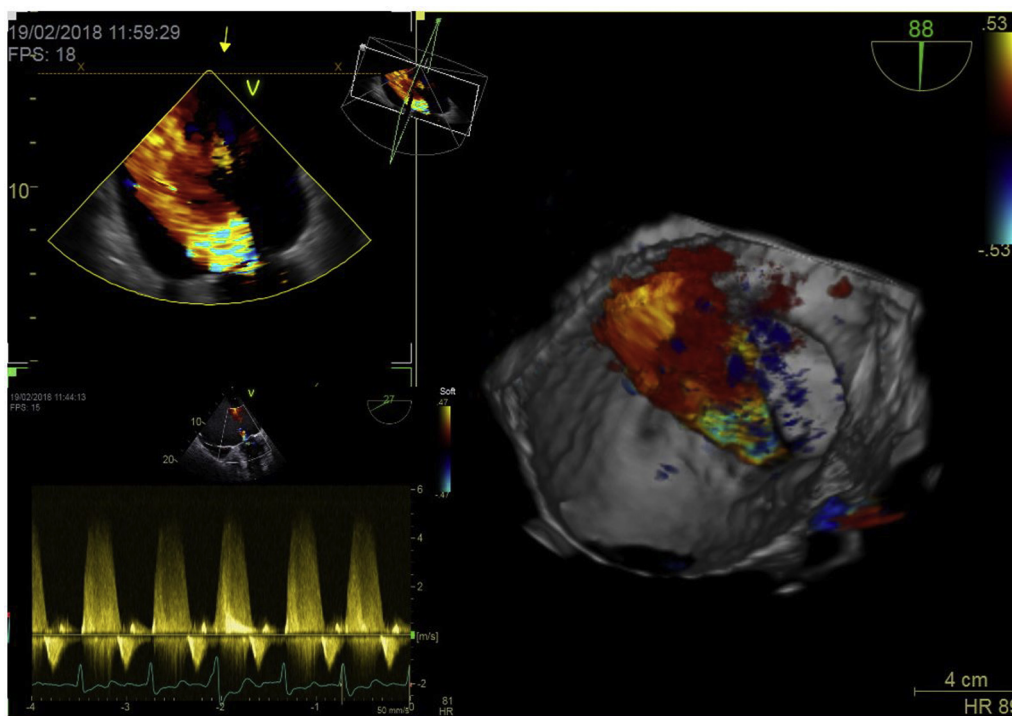
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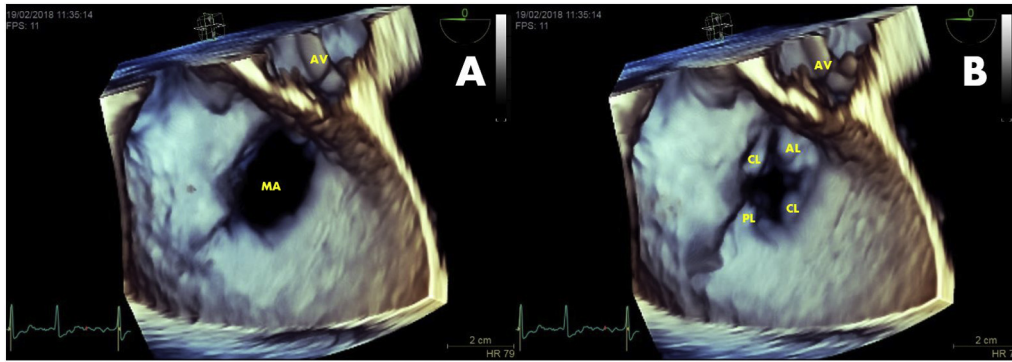
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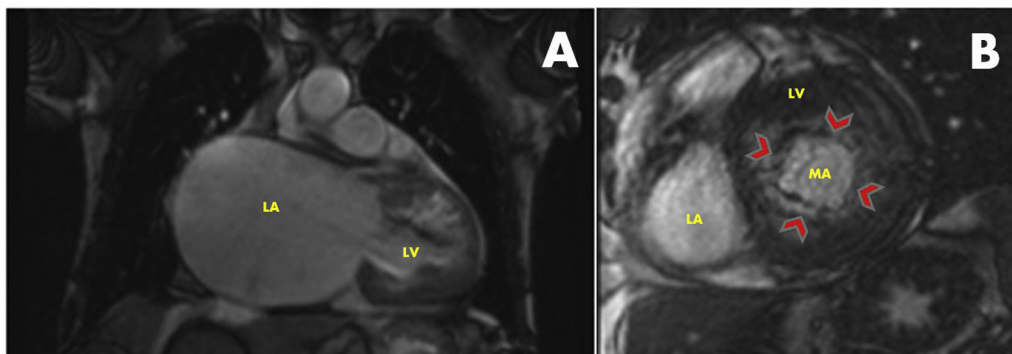
**Figure 1** Transthoracic echocardiography, apical four-chamber view **(A)**, showing aneurysmal LA dilatation, concentric severe left ventricular hypertrophy, and a deformed MV with elongated leaflets; subcostal view **(B)**, showing quadrangular mitral annular geometry (*red arrowhead*); apical two-chamber view **(C)**, showing apical insertion of papillary muscles; and apical four-chamber view **(D)**, showing accessory papillary muscle (*arrow*), originating in the apical lateral region of the left ventricle. ALPM, Anterolateral papillary muscle; LA, left atrium; MA, mitral annulus; PMPM, posteromedial papillary muscle.



**Figure 2** Three-dimensional transesophageal echocardiography, from an atrial view, showing a severe MV regurgitation, with a central regurgitation jet that reaches the roof of the left atrium, secondary to poor leaflet coaptation (effective regurgitant orifice area  $0.45 \text{ cm}^2$ , regurgitant volume 71 ml).



**Figure 3** Three-dimensional transesophageal echocardiography, from an atrial view, showing a quadrangular mitral annulus (MA) and MV leaflets opening in a square shape in diastole (**A**) and closing in a cross shape in systole (**B**). AL, Anterior leaflet; AV, aortic valve; CL, commissural leaflet; PL, posterior leaflet.



**Figure 4** Cardiac magnetic resonance imaging, in steady-state free-precession, two-chamber view (**A**), showing extreme dilatation of the left atrium (LA); and short-axis view (**B**), showing the quadrangular morphology of the MV annulus (MA; red arrowhead). LA, Left atrium; LV, left ventricle.

**Table 1** Abnormalities of the mitral apparatus in HCM<sup>2-6</sup>

Annulus	Leaflets	Chordae Tendinae	Papillary muscles
<ul style="list-style-type: none"> <li>• Larger</li> <li>• Anterior displacement</li> </ul>	<ul style="list-style-type: none"> <li>• Elongated</li> <li>• Clefts</li> <li>• Prolapse</li> <li>• Coaptation more apically</li> <li>• ricuspid or quadricuspid MV</li> </ul>	<ul style="list-style-type: none"> <li>• Elongated</li> <li>• Retracted</li> <li>• Abnormal attachments to mitral leaflets or ventricular walls (false chords)</li> </ul>	<ul style="list-style-type: none"> <li>• Hypertrophied</li> <li>• Elongated</li> <li>• Shortened</li> <li>• Accessory PMs</li> <li>• Additional PM heads</li> <li>• Direct insertion to leaflet</li> </ul>

MV, mitral valve; PM, papillary muscle.

Postmortem studies and surgical findings have shown that HCM is characterized by structural abnormalities of the MV apparatus.<sup>1</sup> Abnormalities may occur in any component of the mitral apparatus (Table 1), the most common being abnormally large mitral leaflets (particularly the anterior) and anterior displacement of any part of the mitral apparatus. Elongation of MV leaflets has been described in patients with HCM in whom pathogenic mutations have been identified, suggesting that MV leaflet elongation is not acquired but is a primary phenotypic expression of HCM.<sup>2,7</sup>

Abnormalities in the size, shape, and angulation of the annulus are less appreciated.<sup>3</sup>

MV clefts have also been described in association with HCM, being an unusual cause of congenital mitral regurgitation. However, they are typically isolated and usually affect the anterior leaflet. The presence of clefts in both MV leaflets represents an extraordinary feature, although there have been reports of clefts in both leaflets with eccentric location and incomplete segmentation, not reaching the annulus.<sup>8</sup>

In this case, although deep clefts reaching the mitral annulus could explain these imaging findings, the presence in both MV leaflets and the division of the two leaflets into approximately four symmetric segments, leading to a four-leaflet MV, make this hypothesis less probable. Furthermore, the quadrangular geometry of the mitral annulus and the valvular opening and closing patterns, suggesting four independent leaflets (one anterior, one posterior, and two commissural leaflets), support the diagnosis of quadricuspid MV. Finally, the diagnosis of quadricuspid MV was made using three-dimensional transthoracic and transesophageal echocardiography and confirmed using CMR, which are well-validated tools to evaluate MV morphology and function.<sup>3,8</sup>

Abnormalities of the number of MV leaflets are rare in HCM, but single cases of tricuspid and quadricuspid MV have been described in the literature among patients with HCM.<sup>4,5</sup>

We report the second case of quadricuspid MV in HCM, while providing impressive images from echocardiography and CMR of these findings. This clinical case emphasizes the association of HCM with MV abnormalities and the need for a comprehensive evaluation of the MV apparatus in patients with HCM.

## CONCLUSION

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Quadricuspid MV is a very rare congenital malformation. To our knowledge, this is the second case report of quadricuspid MV associated with HCM.

## SUPPLEMENTARY DATA

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Supplementary data related to this article can be found at <https://doi.org/10.1016/j.case.2020.03.007>.

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