Letter to the Editor

A rare case of mitral valve dysplasia and left ventricular noncompaction: surgical management and genetic investigation

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Introduction

Mitral valve (MV) dysplasia is a rare and clinically distinct condition. Its etiology remains unclear, and major complications include abnormal function of the MV and chordae tendineae, severe mitral regurgitation (MR) as well as progressive enlargement of the left atrium (LA) and left ventricle (LV), which can lead to various symptoms of left heart failure. Left ventricular noncompaction (LVNC) is a relatively rare cardiomyopathy characterized by the spongy formation of thick trabecular muscles and intertrabecular recesses in the LV. Clinical manifestations include progressive heart failure, refractory arrhythmias, and thromboembolism. In this report, we present the management of a patient with coexisting MV dysplasia and LVNC and explore the potential underlying causes.

Case presentation

A 26-year-old male patient was admitted to the Nanjing Drum Tower Hospital with intermittent chest tightness and shortness of breath for over 6 months, with exacerbation within the previous 2 weeks. Transthoracic echocardiography (TTE) revealed rupture of the chordae tendineae of the anterior leaflet of the MV, MV prolapse with severe regurgitation (*Figure 1A*,*1B*; Video S1), moderate tricuspid regurgitation, and severe pulmonary hypertension [pulmonary arterial systolic pressure (PASP) =70 mmHg; 1 mmHg ≈ 0.133 kPa]. Furthermore, the ratio of noncompacted myocardium to dense myocardium in the ventricle was greater than two (*Figure 1C*; Videos S2,S3), and there was an indented change in the LV consistent with LVNC (1). Following thorough preoperative examinations, the patient was diagnosed with MV prolapse and left ventricular dysfunction. After a multidisciplinary discussion, the initial plan was to perform mitral and tricuspid valve repair. However, an unexpected finding during the procedure prompted the team to choose mechanical MV replacement as the final solution. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

Surgical procedure and outcome

Intraoperative transesophageal echocardiography (TEE)

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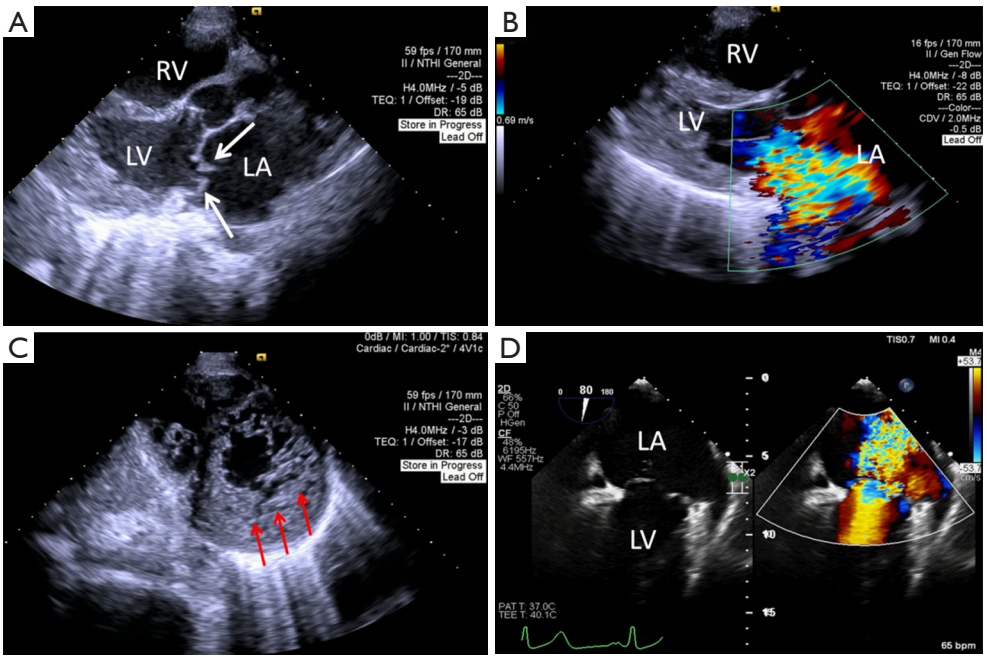

Figure 1 Echocardiographic images. (A) Long-axis view of the LV on TTE showing the entire mitral leaflet protruding into the LA during systole (white arrows indicate the prolapsed MV). (B) The long-axis view of the LV on TTE showing severe systolic MR. (C) The parasternal short-axis left ventricular apical view on TTE showing a ratio of noncompacted myocardium to dense myocardium of the ventricle greater than two (the red arrows indicate the noncompacted myocardium). (D) Intraoperative TEE showing severe systolic MR. RV, right ventricle; LV, left ventricle; LA, left atrium; TTE, transthoracic echocardiography; MV, mitral valve; MR, mitral regurgitation; TEE, transesophageal echocardiography.

confirmed the ruptured chordae tendineae of the anterior MV, which had led to significant MV prolapse with a large amount of regurgitation (*Figure 1D*; Videos S4,S5). The myocardium of the apex of the LV was loose, and a large number of intermyocardial sinuses were observed during diastole.

The patient underwent mechanical MV replacement and tricuspid annuloplasty to reduce tricuspid regurgitation. Postsurgery, the patient experienced improvement in chest tightness and shortness of breath. Follow-up TTE showed well-functioning mechanical MV and mild tricuspid regurgitation. Pulmonary artery pressure was reduced (PASP =35 mmHg), and left ventricular size decreased postsurgery (Video S6), indicating the effectiveness of the surgical treatment.

Genetic investigation

An interesting finding during surgery was the abnormal morphology of the MV's chordae tendineae. The chordae tendineae appeared slender, long, and with an earthworm-like shape, an extremely rare pattern (*Figure 2*). Genetic testing was conducted, and although no variants highly associated with disease were found, three loci (*FLNC*, *AGL*, and *AEBP1*) showed insufficient evidence to exclude possible pathogenic variants. *FLNC*, *AGL*, and *AEBP1* have been linked to familial hypertrophic cardiomyopathy (2), glycogen storage disease type III (3), and classic Ehlers-Danlos syndrome type 2 (4) respectively, all of which may involve MV-related issues. Previous studies have indicated there to be a link between familial hypertrophic cardiomyopathy and MV complex abnormalities (5). The patient in this case did not display any echocardiographic indications of hypertrophic cardiomyopathy. It is worth noting that we did not have access to the literature to review the mutations at the *FLNC* gene locus, which is associated with MV abnormalities. However, Captur *et al.* demonstrated that carriers of the *HCM* gene display increased myocardial trabecular complexity as a preclinical abnormality, but they a lack thickened left ventricular myocardium (6). Although the pathophysiological mechanism of the disease remains uncertain, the correlation between mutations in

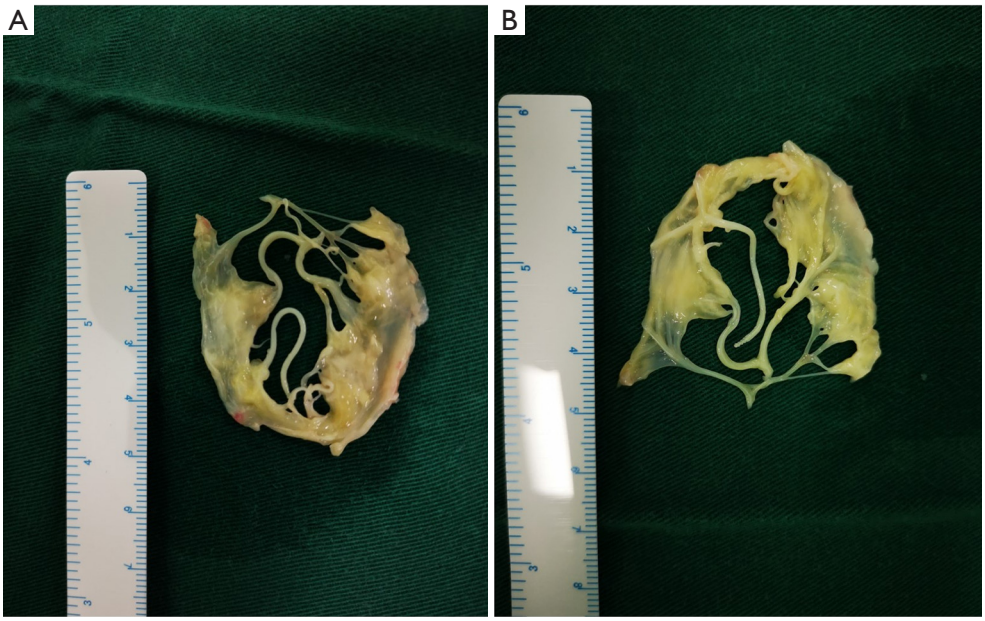

Figure 2 Dysplastic MV and chordae tendineae resected intraoperatively. Abnormal development of the MV apparatus with sparse mitral tendons can be clearly seen [(A) view from the LA; (B) view from the LV]. MV, mitral valve; LA, left atrium; LV, left ventricle.

the *FLNC* gene and left ventricular nonfilling is plausible in this instance. However, no definitive evidence was found to establish a direct association with the development of mitral chordal dysplasia and LVNC in this patient.

Conclusions

MV dysplasia is a rare congenital dysfunction of the MV. As the disease progresses, patients often develop severe MR, resulting in a variety of symptoms and signs of heart failure (7). MV dysplasia presents significant challenges in clinical management, and surgery remains the primary option for affected individuals (8,9). Pace Napoleone *et al.* reported a case of an infant with MV dysplasia, and the infant underwent Ross-Kabbani operation with satisfactory results (10). However, many patients received poor outcomes after surgery or palliative care, with some even dying (11,12). Surgical advancements, particularly MV reconstruction, show promise in patients with congenital MV dysplasia (13).

Congenital MV disease is frequently associated with various complex intracardiac malformations, and only a few occur in isolation (14). In this particular case, the patient presented not only with MV disease but also with a concurrent diagnosis of LVNC. To the best of our knowledge, this is the first documented case report of MV dysplasia coexisting with LVNC. Clinical manifestations of LVNC include refractory arrhythmias, progressive heart failure, and potentially serious complications, such as thromboembolism (15). Consequently, severe MR resulting from MV dysplasia may further exacerbate the prognosis of LVNC. Nevertheless, the surgical intervention implemented in this case proved to be beneficial, effectively alleviating the symptoms. Regular monitoring of the patient's progress is essential to assessing and evaluating the long-term outcomes effectively.

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Footnote

Conflicts of Interest: All authors have completed the ICMJE uniform disclosure form (available at https://qims.amegroups.com/article/view/10.21037/qims-23-1159/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures

performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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