CASE REPORT

Totally endoscopic mitral valve repair in a young patient with marfan syndrome and barlow disease: a case report

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Abstract

Objective: Barlow disease is a complex form of degenerative mitral valve disease characterized by diffuse myxomatous degeneration, excess leaflet tissue, bileaflet prolapse, annular dilatation, and severe mitral regurgitation. In patients with Marfan syndrome, connective tissue abnormalities may further increase anatomical complexity. This report describes the diagnostic work-up, operative strategy, and early outcome of totally endoscopic mitral valve repair in a young adult with Marfan syndrome and Barlow disease.

Case presentation: A thirty-one-year-old male patient with Marfan syndrome, progressive exertional dyspnea, and severe degenerative mitral regurgitation underwent clinical examination, electrocardiography, chest radiography, transthoracic echocardiography, transesophageal echocardiography, and thoraco-abdomino-pelvic computed tomography angiography for preoperative planning. Surgical treatment consisted of totally endoscopic mitral valve repair using expanded polytetrafluoroethylene neochordae, cleft closure, commissural repair, and prosthetic ring annuloplasty. The computed tomography angiography did not identify pathological thoraco-abdomino-pelvic findings and supported the feasibility of peripheral cannulation and endoscopic access. The procedure was completed successfully. Post-repair echocardiography demonstrated satisfactory leaflet coaptation, no residual mitral regurgitation, absence of systolic anterior motion, normal transmitral gradients, and a coaptation zone of approximately fifteen millimeters. The postoperative course was uneventful, and the patient was discharged home on postoperative day six, with progressive resumption of daily activities.

Conclusions: This case illustrates the feasibility and favorable early outcome of totally endoscopic mitral valve repair in a selected young patient with Marfan syndrome and complex degenerative mitral valve disease treated in an experienced center. Careful multimodality imaging, individualized repair strategy, and structured follow-up are essential for durable functional results.

Keywords: Barlow disease; Marfan syndrome; mitral valve repair; mitral valve prolapse; totally endoscopic cardiac surgery

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Introduction

Mitral valve prolapse is among the most frequent valvular heart diseases, with an estimated prevalence of approximately 2-3% in the general population [1]. It is defined by systolic displacement of one or both mitral leaflets into the left atrium and may be associated with a broad spectrum of mitral regurgitation severity, ranging from mild asymptomatic disease to severe valvular dysfunction requiring surgical correction. When severe mitral regurgitation remains untreated, chronic volume overload may lead to left atrial enlargement, progressive left ventricular remodeling, heart failure, arrhythmias, and increased morbidity and mortality [5].

Degenerative mitral valve disease represents the leading cause of primary mitral regurgitation in industrialized countries and includes phenotypes with distinct anatomical and clinical characteristics. Barlow disease is considered one of the most complex degenerative phenotypes and is characterized by diffuse myxomatous degeneration, leaflet thickening and redundancy, excessive valvular tissue, elongated chordae tendineae, annular dilatation, and bileaflet prolapse [2,4,6]. These features increase the technical complexity of valve reconstruction and often require an individualized repair strategy.

Mitral valve repair is currently preferred over replacement in degenerative mitral regurgitation whenever a durable reconstruction is feasible, particularly in young patients. Repair preserves the native valve apparatus, supports postoperative ventricular function, reduces prosthesis-related complications, and avoids the need for lifelong anticoagulation associated with mechanical valve replacement [3,15,16]. In parallel, minimally invasive and totally endoscopic techniques have expanded the therapeutic armamentarium, offering reduced surgical trauma, improved cosmetic results, shorter recovery, and excellent functional outcomes in selected patients treated in experienced centers [17,18].

The objective of this case report was to present the diagnostic work-up, multimodality imaging findings, opera-

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tive technique, and early postoperative outcome of a young adult patient with Marfan syndrome and severe mitral regurgitation caused by Barlow disease who underwent totally endoscopic mitral valve repair.

Case presentation

We report the case of a 31-year-old male patient with Marfan syndrome and a known history of valvular heart disease who was admitted to our hospital for progressively worsening exertional dyspnea and reduced exercise tolerance. The patient reported that his symptoms had gradually increased over the preceding months. At admission, he was classified as New York Heart Association functional class II, with limitation during moderate physical activity but no symptoms at rest. His medical history included arterial hypertension and intermittent symptoms related to supraventricular and ventricular extrasystolic arrhythmias.

Clinical examination revealed regular cardiac rhythm and a grade III/IV systolic murmur best heard at the apex, with radiation toward the axilla. There were no signs of peripheral edema, hypoperfusion, cyanosis, pallor, or acute heart failure. Peripheral pulses were normal and symmetrical. Blood pressure values were within normal limits during the evaluation.

The electrocardiogram showed sinus rhythm at approximately 80 beats per minute, without conduction abnormalities, ischemic changes, or repolarization disorders. Chest radiography showed clear lung fields, open costophrenic angles, and no evidence of pulmonary congestion, pleural effusion, or acute pulmonary infiltrates. The thoracic scout image also demonstrated a longilinear habitus compatible with Marfan syndrome (Figure 1-A-B).

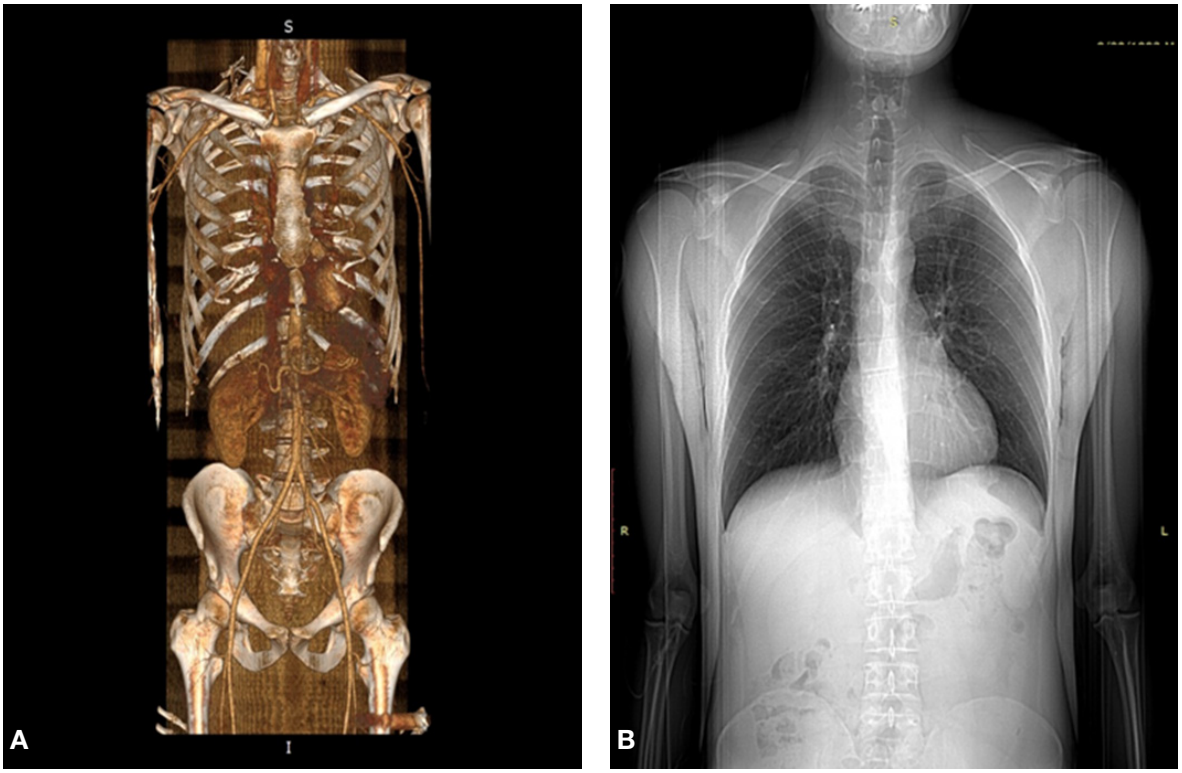


Fig. 1. A- Preoperative three-dimensional computed tomography reconstruction used for planning of the totally endoscopic approach and peripheral cannulation. No pathological thoraco-abdomino-pelvic findings were identified. B - Preoperative thoracic scout/chest radiographic image showing a longilinear habitus compatible with Marfan syndrome, with no signs of pulmonary congestion or pleural effusion.

Preoperative transthoracic echocardiography demonstrated marked degenerative abnormalities of the mitral valve. The mitral leaflets were thickened and redundant, with excessive valvular tissue and pronounced billowing consistent with Barlow disease. Bileaflet prolapse was present, involving both the anterior and posterior leaflets. Severe mitral regurgitation was identified, predominantly central, with a maximum regurgitant velocity of 5.9 meters per second. The vena contracta width was 3 mm, and pulmonary venous flow demonstrated systolic flow reversal, consistent with severe mitral regurgitation. The left atrial volume was 219 mL, corresponding to an indexed left atrial volume of 99 mL/m2. Left ventricular systolic function was preserved, with an estimated ejection fraction of approximately 60%. The left ventricular end-diastolic diameter measured 56 millimeters, with an end-systolic diameter of 40 millimeters; the right ventricular end-diastolic diameter measured 36 millimeters, and the left atrium was mildly enlarged, measuring 43 millimeters. No regional wall motion abnormalities were noted. Given the patient's Marfan syndrome, aortic dimensions were also assessed: the aortic annulus measured 24.7mm, the aortic root (si-

nuses of Valsalva) measured 40.6 mm, and the ascending aorta measured 26.4 mm, all within acceptable limits, although the aortic root diameter was at the upper range of normal. Estimated pulmonary artery systolic pressure was 29 mmHg. The aortic valve was tricuspid, thin, mobile, and competent. The tricuspid valve had normal morphology with mild regurgitation, and no pericardial effusion was present.

Intraoperative transesophageal echocardiography provided a more detailed assessment of the mitral valve apparatus and confirmed diffuse myxomatous degeneration with excess leaflet tissue and bileaflet prolapse, predominantly involving the A2-A3 and P2-P3 segments (Figure 2-A). Mitral annular dilatation was also present, and severe regurgitation included both central and eccentric jets on color Doppler imaging (Figure 2-B). These findings confirmed the anatomical complexity of the lesion and supported a reparative strategy rather than valve replacement.

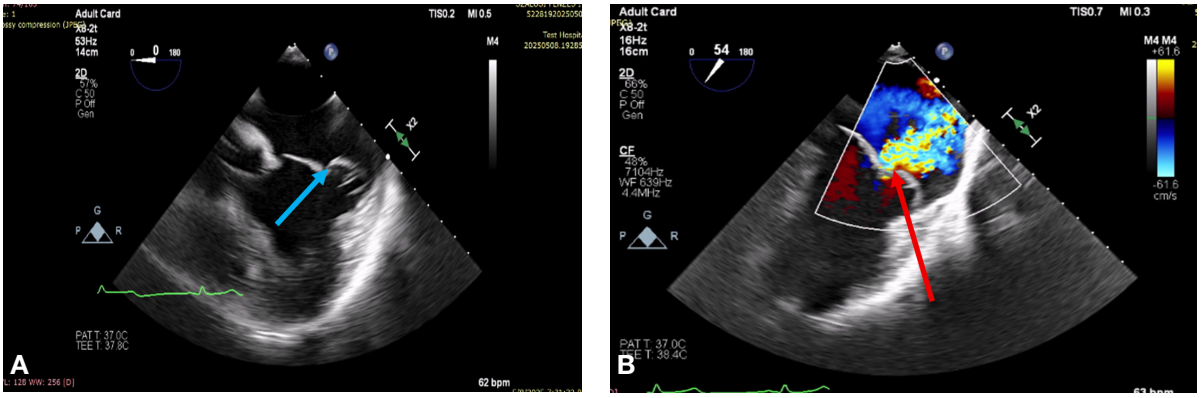


Fig. 2. A - Preoperative transesophageal echocardiography showing a myxomatous mitral valve with bileaflet prolapse and morphological features compatible with Barlow disease. B - Preoperative transesophageal echocardiography with color Doppler showing severe mitral regurgitation.

Considering the presence of symptoms, severe mitral regurgitation, preserved ventricular systolic function, favorable anatomy for reconstruction, and suitable peripheral access confirmed by computed tomography angiography, the patient was referred for surgery. Totally endoscopic mitral valve repair was performed under general anesthesia with cardiopulmonary bypass established through peripheral cannulation of the right femoral artery and combined femoral and jugular venous drainage. The procedure was performed through a right anterolateral mini-thoracotomy in the fourth intercostal space, with the camera port and the transthoracic aortic clamp (Chitwood clamp) introduced through the third intercostal space. After cardioplegic arrest, the left atrium was opened through Sondergaard's groove, and the mitral valve was exposed using a dedicated atrial retractor. Cardiopulmonary bypass time was 209 minutes, aortic cross-clamp time was 146 minutes, and total operative (skin-to-skin) time was 300 minutes. The operative setting is shown in Figure 3.

Intraoperatively, extensive myxomatous degeneration consistent with Barlow disease was observed, including marked leaflet redundancy, excess valvular tissue, bileaflet prolapse involving the anterior and posterior mitral leaflets, annular dilatation, and elongated chordae tendineae (Figure 4-A). Repair was tailored to the individual valve anatomy. Eight expanded polytetrafluoroethylene neochordae were implanted, four to the anterior leaflet and four to the posterior leaflet, to restore leaflet motion and coaptation. The neochordae were anchored to the papillary

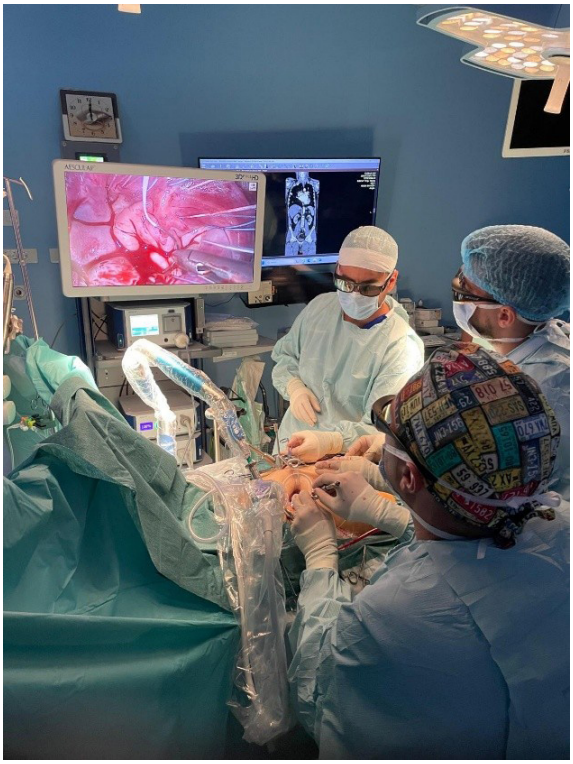


Fig. 3. Intraoperative setting of totally endoscopic mitral valve repair, demonstrating the minimally invasive operative field and the use of high-definition endoscopic visualization. The surgical team performs the procedure through limited-access ports, with intracardiac anatomy and the mitral valve apparatus displayed on the monitor. This approach enables precise valve repair while avoiding conventional median sternotomy and minimizing surgical trauma.

muscles and subsequently attached to the free edge of the prolapsing leaflets (Figures 4-B and C). Cleft closure was performed at the P2-P3 and A1-A2 levels, together with commissural repair at A1-P1. A 36-millimeter Medtronic annuloplasty ring was implanted to stabilize the mitral annulus and optimize leaflet coaptation.

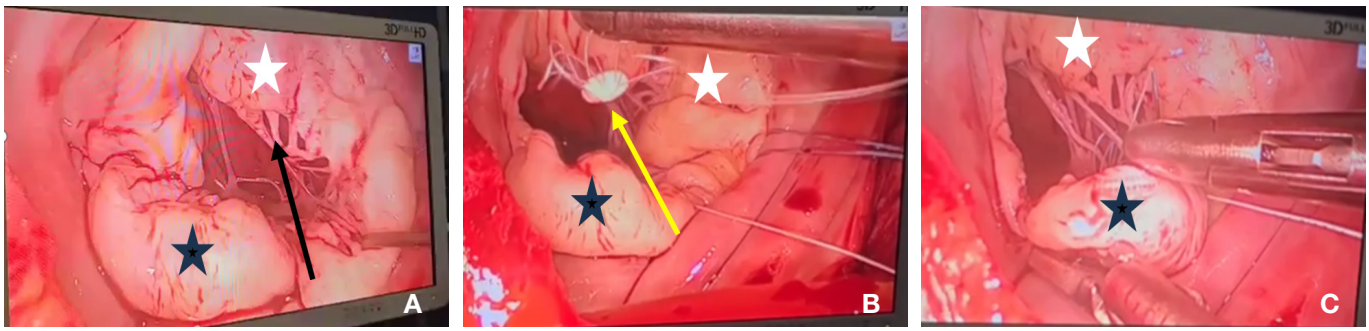


Fig. 4. A- Intraoperative endoscopic view showing a myxomatous mitral valve with excess leaflet tissue and prolapse of the anterior and posterior mitral leaflets. Black arrow indicates the native chordae. B - Intraoperative endoscopic view during implantation of expanded polytetrafluoroethylene neochordae (yellow arrow) and their anchoring to the papillary muscles. C - Intraoperative endoscopic view showing attachment of the neochordae to the free edge of the prolapsing mitral leaflet. The black star indicates the posterior mitral leaflet, while the white star indicates the anterior mitral leaflet in all three images.

Post-repair intraoperative transesophageal echocardiography demonstrated satisfactory leaflet apposition, no residual mitral regurgitation, absence of systolic anterior motion, normal transmitral gradients, and a broad coaptation zone measuring approximately 15 millimeters (Figure 5), confirming the immediate success of the reconstruction.

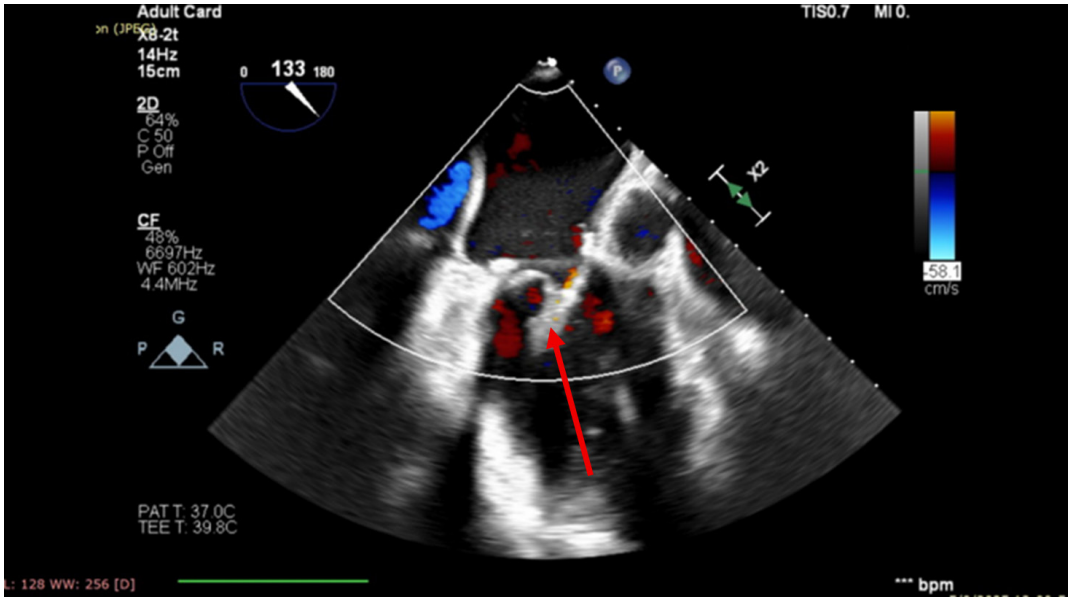


Fig. 5. Post-repair transesophageal echocardiographic control demonstrating absence of residual mitral regurgitation and a broad leaflet coaptation (red arrow) zone of approximately 15 millimeters.

The postoperative course was uneventful. The patient remained hemodynamically stable, in sinus rhythm, without significant dysrhythmias or hypotensive episodes. Follow-up transthoracic echocardiography demonstrated preserved left ventricular systolic function, with an estimated ejection fraction of approximately 55%, appropriate position of the annuloplasty ring, and no residual mitral regurgitation. Mild tricuspid regurgitation was also noted, with no pericardial effusion.

During hospitalization, the patient remained afebrile, surgical incisions healed normally, and no local or systemic signs of infection were observed. Oral anticoagulation with acenocoumarol was initiated, with international normalized ratio monitoring according to institutional protocol. The patient was discharged home on postoperative day six in stable clinical condition, asymptomatic, and able to progressively resume daily activities. Long-term cardiological follow-up was recommended, including periodic assess-

ment of ventricular function, mitral valve competence, and possible late rhythm disturbances.

Discussion

Barlow disease represents a particularly complex phenotype of degenerative mitral valve disease, as it involves diffuse structural alteration of the entire mitral apparatus rather than an isolated prolapsing segment. The combination of myxomatous degeneration, excess leaflet tissue, bileaflet prolapse, annular dilatation, and elongated chordae increases the technical difficulty of repair and may influence long-term durability [2,4,6]. The echocardiographic and intraoperative findings in this case were typical of Barlow disease and justified a comprehensive repair approach.

Marfan syndrome is a connective tissue disorder with major cardiovascular implications, including aortic root disease and valvular abnormalities. Mitral valve lesions in these patients may be more complex because of diffuse extracellular matrix involvement, leaflet redundancy, chordal elongation, and progressive myxomatous degeneration [19]. The coexistence of Marfan syndrome and Barlow disease in this patient explains the pronounced degenerative phenotype and reinforces the need for detailed preoperative anatomical assessment and individualized repair planning.

Accurate preoperative and intraoperative imaging is essential for planning in complex degenerative mitral regurgitation. Transthoracic echocardiography established the severity of regurgitation and ventricular response, while transesophageal echocardiography allowed precise localization of prolapsing segments and assessment of repair feasibility. Computed tomography angiography additionally excluded relevant vascular pathology and confirmed the safety of peripheral cannulation and endoscopic access, consistent with contemporary recommendations for evaluating native valvular regurgitation [20].

The timing of surgery is a major determinant of outcome in severe primary mitral regurgitation, since early referral before irreversible ventricular remodeling may improve long-term prognosis [3,15]. In this patient, symptoms were already present, but left ventricular systolic function remained preserved, a profile that supported surgical correction and likely contributed to the favorable early outcome.

Mitral valve repair remains the preferred strategy for degenerative regurgitation when durable correction is achievable, preserving the subvalvular apparatus and avoiding prosthesis-related complications [15,16]. In Barlow disease, repair frequently requires a combination of techniques; in this case, multiple expanded polytetrafluoroethylene neochordae addressed bileaflet prolapse, cleft closure improved leaflet continuity, commissural repair corrected localized instability, and ring annuloplasty restored annular geometry, together allowing effective repair despite the anatomical complexity.

Totally endoscopic mitral surgery has evolved from a niche approach to an established option in experienced centers, potentially reducing surgical trauma, postoperative pain and recovery time while preserving repair efficacy in selected patients [17,18]. The uneventful recovery observed here supports the feasibility of this approach for complex degenerative disease when patient selection, imaging, and surgical expertise are adequate.

In young patients with connective tissue disease, bileaflet prolapse and diffuse myxomatous degeneration have also been linked to an arrhythmogenic mitral valve phenotype, particularly when mitral annular disjunction is present [7-14]. Although disjunction was not specifically assessed in this patient, his history of extrasystolic arrhythmias and bileaflet prolapse warrant continued rhythm surveillance, since correction of regurgitation does not always eliminate arrhythmic risk [10,14].

Long-term durability remains the central objective of mitral repair in young patients: recurrent regurgitation, annular changes, or rhythm disturbances may still occur despite a successful initial result. Structured follow-up, combining echocardiography and rhythm assessment, is therefore necessary, for the repaired valve and, in this patient, for ongoing aortic surveillance given the Marfan diagnosis [15]. In the present case, follow-up has so far been limited to the early postoperative period, and longer-term assessment will be required to confirm the durability of the repair.

This report has the inherent limitations of a single-patient case study and cannot support generalizable conclusions. Nonetheless, it illustrates the value of combining detailed multimodality imaging, timely referral, and individualized totally endoscopic repair in the management of complex degenerative mitral regurgitation.

Conclusion

Barlow disease may cause severe mitral regurgitation in young patients and requires careful anatomical evaluation because of the complexity of the mitral valve lesions. In the presented patient with Marfan syndrome, totally endoscopic mitral valve repair using multiple neochordae, cleft closure, commissural repair, and ring annuloplasty achieved a favorable early result, with preserved ventricular function, no residual mitral regurgitation, and an uneventful postoperative course.

This case supports the role of individualized mitral valve repair as the preferred therapeutic strategy for selected patients with complex degenerative disease. When performed in experienced centers, totally endoscopic surgery may provide effective correction with the potential benefits of a minimally invasive approach. Continued echocardiographic and rhythm follow-up remains essential to evaluate long-term repair durability and clinical outcome.

Ethical Statement

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images. This report complies with the ethical standards of the institutional research committee and the 1964 Dec-

laration of Helsinki and its subsequent amendments.

Informed Consent Statement

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Authors' contribution

A. I. P. (Conceptualization; Writing – original draft; Writing – revision & editing)
E. D. A. (Conceptualization; Supervision; Writing – revision & editing)
S. I. A. (Investigation; Data curation; Writing – review & editing)
M. M. H. (Conceptualization; Formal analysis; Methodology; Supervision; Writing – revision & editing).

Conflict of interest

No conflict of interest.

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AI Tools Disclosure

Grammarly (v1.2.182.1722) was used for grammar and clarity improvements. All AI suggestions were reviewed and approved by the authors.

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