Case Report

Hypoplastic posterior mitral leaflet associated with ostium secundum atrial septal defect

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ABSTRACT

Hypoplastic posterior mitral leaflet is a congenital dysplastic mitral valve abnormality rarely reported in adults. The presentation is varied, from being asymptomatic to significantly symptomatic due to progressive mitral regurgitation. In many of these cases additional cardiac lesions are noted, most commonly, bicuspid aortic valve and atrial septal defect. Hence, while performing imaging assessment for these conditions, a comprehensive evaluation of the mitral valve apparatus is essential so as not to miss the valvular pathology. The ideal therapeutic approach and timing for intervention in hypoplastic posterior mitral leaflet is still not clear due to a rarity of these cases. Mitral valve repair though preferred, may not be the ideal treatment modality and many patients require mitral valve replacement especially when presenting at an older age. We hereby report about a 56-year-old male patient who was diagnosed with ostium secundum atrial septal defect and hypoplastic posterior mitral leaflet. He underwent successful surgical closure of the atrial septal defect and concomitant bileaflet metallic prosthetic mitral valve replacement. His postoperative course was uneventful and was associated with amelioration of symptoms during follow-up.

Key words: Congenital mitral regurgitation; Hypoplastic posterior mitral leaflet; Ostium secundum atrial septal defect; Myxomatous mitral valve; Mitral valve repair; Mitral valve replacement.

Congenital mitral valve (MV) abnormalities are rarely diagnosed in routine clinical practice. Due to their frequent combination with other prominent cardiac anomalies and the asymptomatic course in isolated cases, the diagnosis is often missed, particularly in adult population. In a study reviewing echocardiographic examinations in 13,400 pediatric patients over 7.5 years, congenital MV abnormalities were diagnosed in only 0.5% cases [1]. Hypoplastic mitral leaflet is an even more uncommon condition, seen in 4% of patients with congenital mitral regurgitation (MR) [2]. Hypoplastic posterior mitral leaflet (HPML) is part of a spectrum of dysplastic MV lesions, extending from hypoplasia to agenesis of the posterior leaflet (unileaflet MV). Due to isolated valvular involvement in the MV apparatus, HPML is classified as causing type 2 (valvular) MR, according to updated congenital MR classification [3].

HPML has been reported in association with other cardiac lesions in around 40% of the cases, namely bicuspid aortic valve (BAV), atrial septal defect (ASD), ventricular septal defect (VSD), left ventricular non-compaction and anomalous pulmonary venous drainage [4,5]. Currently, available information on this clinical condition is restricted to isolated case reports and case series in medical literature [5,6,7].

Due to the frequent consideration of rheumatic or degenerative etiologies for MV disease in adults, congenital lesions such as HPML may be significantly under reported in our clinical context. An early diagnosis is important to ensure timely intervention before significant deterioration of cardiac function sets in. We present the case report of a middle-aged male patient with ostium secundum atrial septal defect (OS-ASD) who was incidentally diagnosed with concomitant HPML during diagnostic workup and subsequently underwent successful surgical treatment for both the lesions.

CASE REPORT

A 56-year-old male patient presented to the out-patient department with 1-month history of exertional dyspnea, chest pain and palpitations (New York Heart Association class III),

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which relieved with rest. There was no significant past history of medical illness or chronic substance abuse. He had consulted another health care facility a few days prior and had been diagnosed there with OS-ASD along with vitamin D deficiency. He was subsequently referred to a higher medical centre for further management. His ongoing medications included- furosemide, spironolactone, cholecalciferol and oral calcium supplements.

At presentation, patient was conscious and alert with heart rate- 73/min, blood pressure- 123/82 mmHg, respiratory rate- 22/min and oxygen saturation of 99% at room air. His systemic examination was significant for a soft first heart sound and fixed wide splitting of the second heart sound. Pulmonary component of the second heart sound was loud in intensity. Systolic murmurs were audible in pulmonary area (grade III/VI) and in mitral area (grade II/VI).

Patient's electrocardiogram was suggestive of sinus rhythm, first degree atrioventricular block, intraventricular conduction defect and features of left atrial abnormality (Figure 1). Transthoracic echocardiography (TTE) showed a single OS-ASD of size 25 mm with left to right shunt (Figure 2A,B, Videos 1-4). The aortic rim of the defect was deficient with the posterior and inferior vena cava rims borderline. The pulmonary to systemic flow ratio (Q_P/Q_S) was >1.5. Right atrium (RA), right ventricle (RV) and main pulmonary artery were dilated (Figure 2C-E). There was mild to moderate tricuspid regurgitation and pulmonary artery systolic pressure was 40 mmHg. The RV systolic function was normal. All the pulmonary veins drained normally into left atrium (LA).

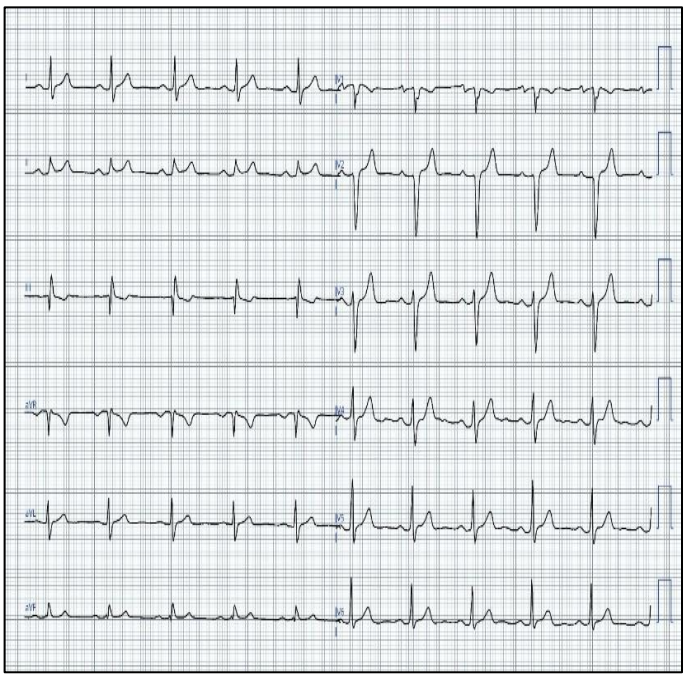

Figure 1. 12-lead electrocardiogram demonstrating sinus rhythm with first degree atrioventricular block and left atrial abnormality (lead V1).

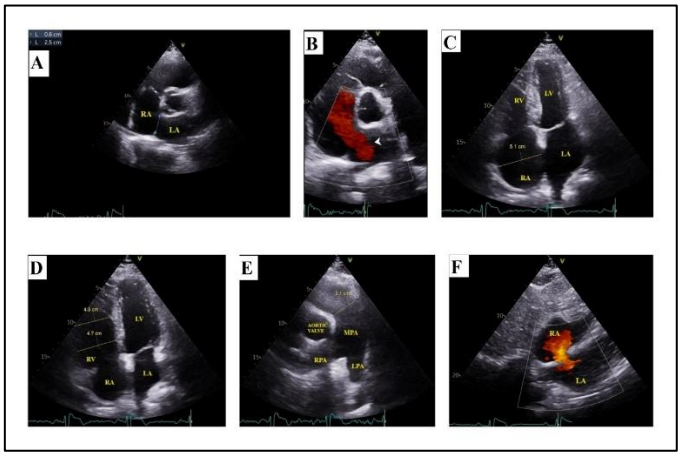

Figure 2. Transthoracic echocardiographic images (parasternal short axis view) showing 25 mm sized ostium secundum atrial septal defect with left to right flow (A,B), dilated right atrium, right ventricle and main pulmonary artery in apical 4- chamber and parasternal short axis views (C-E), subcostal view showing left to right shunt across atrial septal defect (F). LA- Left atrium, LV- Left ventricle, LPA- Left pulmonary artery, MPA-Main pulmonary artery, RA- Right atrium, RPA- Right pulmonary artery, RV- Right ventricle.

The MV showed near-total absence of posterior mitral leaflet (PML) while the anterior mitral leaflet (AML) was myxomatous and prolapsing (Figure 3A, Videos 5-7). The LA (42 mm) was dilated. This was associated with mild to moderate MR, which was posteriorly directed (Figure 3B). Papillary muscles were 2 in number with normal morphology and orientation (Figure 3C). The aortic valve was structurally and functionally normal. There were no other associated congenital cardiac anomalies. Left ventricular (LV) internal dimension was normal (40/ 26 mm). The LV ejection fraction was 55%.

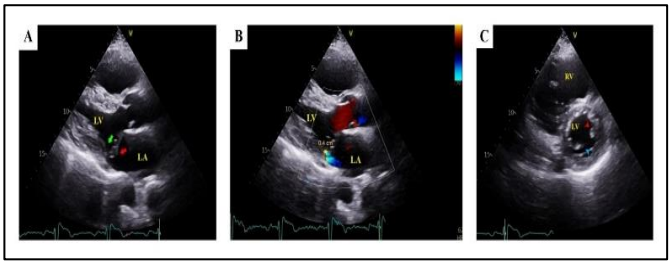

Figure 3. Transthoracic echocardiographic images in parasternal long axis view showing severely hypoplastic posterior mitral leaflet (green arrow) with a myxomatous, elongated and prolapsing anterior mitral leaflet (red arrow) (A), mild to moderate posteriorly directed jet of mitral regurgitation (vena contracta width 4mm) (B), anterolateral papillary muscle (red arrowhead) and posteromedial papillary muscle (blue arrowhead) in parasternal short axis view (C). LA- Left atrium, LV- Left ventricle, RV- Right ventricle

Transesophageal echocardiography confirmed the TTE findings of a single OS-ASD of maximum size 25 mm (Figure 4, Videos 8-11). The superior and aortic rims were deficient while the atrioventricular rim was borderline. MV morphology revealed rudimentary PML with thin rim of leaflet tissue in the usual P1 scallop location. The muscular continuation extended and protruded from the annular plane (Figure 5A, Videos 12-15). The AML was elongated, sail like 41 mm in length with the tip 10 mm in thickness (Figure 5B). MV annulus was dilated; intercommissural- 40 mm, septo-lateral- 36 mm (Figure 5C). Redundant chordae were noted in relation to AML. Moderate MR was seen with the jet in a posterior direction due to coaptation gap between AML and rudimentary PML tissue (Figure 5D).

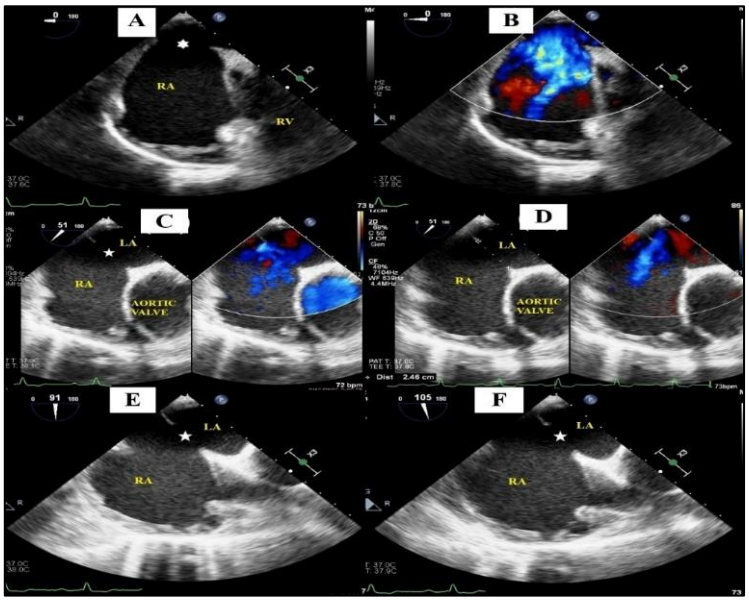

Figure 4. Transesophageal echocardiographic images at 0, 50 and 90-100 degrees showing large ostium secundum atrial septal defect (white star) with left to right shunt. Deficient superior and aortic rim with borderline atrioventricular rim noted (A-D). Superior vena caval and inferior vena caval rims are adequate (E,F). LA- Left atrium, RA- Right atrium

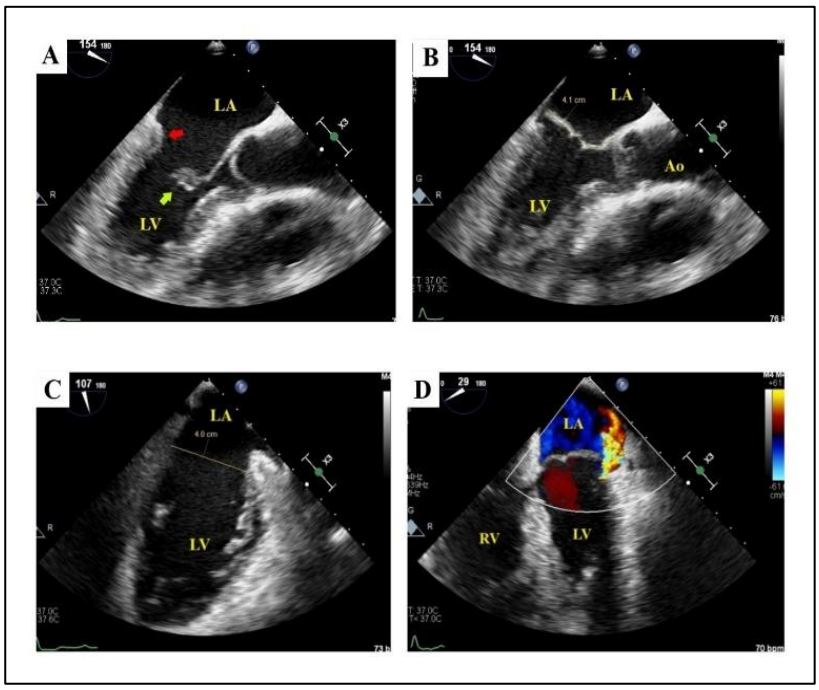

Figure 5. Transesophageal echocardiographic views in 150 and 100 degrees showing near absent posterior mitral leaflet (red arrow) with muscular continuation (A), anterior mitral leaflet is sail like, elongated (41 mm) and prolapsing (B), dilated bicommissural mitral annulus (40 mm) (C), moderate posteriorly directed jet of mitral regurgitation on long axis 29-degree view (D). Ao- Ascending aorta, LA- Left atrium, LV- Left ventricle, RV- Right ventricle

3-dimensional imaging delineated the valvular pathology in detail and demonstrated the A2 and A3 scallops prolapsing into LA (Figure 6, Videos 16-19). A diagnosis of OS-ASD with hypoplastic PML was confirmed on transesophageal echocardiography.

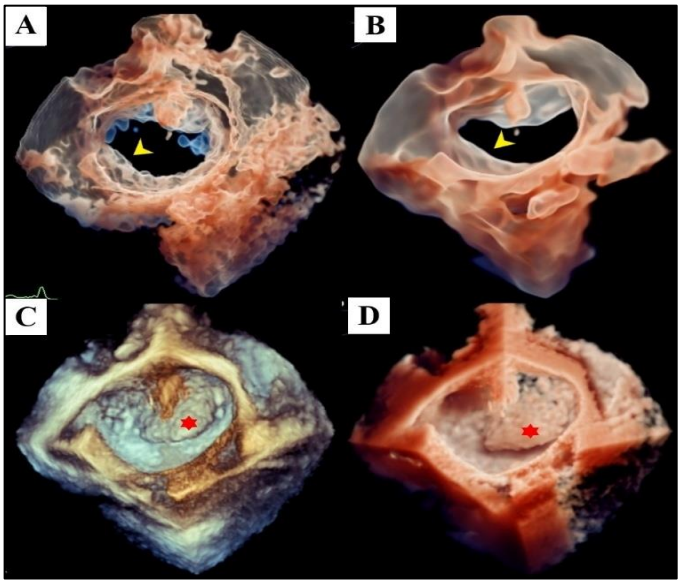

Fig 6. 3-dimensional transesophageal echocardiography images demonstrate hypoplastic posterior mitral leaflet with rudimentary tissue in the region of P1 scallop (yellow arrowhead) along with muscular continuation (A, B), anterior mitral leaflet (A2, A3 scallops- red star) seen prolapsing (C, D).

Coronary angiography revealed normal epicardial coronary arteries with no stenotic lesions noted. Patient was advised surgical OS-ASD closure with MV replacement in view of the imaging findings and symptomatic status. He underwent successful surgical closure of OS-ASD using autologous pericardial patch and MV replacement using size 31/33 mm ON-X® bileaflet mechanical prosthetic valve under cardiopulmonary bypass. Post-procedure patient had an uneventful recovery. His pre-discharge TTE showed a normal functioning MV prosthesis with a mean pressure gradient of 2 mmHg, with no paravalvular leak. Interatrial septum was adequately repaired with no residual shunt across the repair site. The RA and RV dimensions had regressed. The LV systolic function was normal. There was no pericardial collection. Subsequently, he was discharged on the 5th post-operative day on medical therapy. He has been on regular out-patient follow-up over 5 months and has noted significant improvement in symptoms postoperatively.

DISCUSSION

MV development during human embryogenesis initiates at day 18-19 of intrauterine life and is completed by week 15, although the fetal MV continues to grow in size till full maturity [8]. Endocardial cushion cells contribute to MV development in fetal heart tube [9]. Several genes are involved during this process such as NKX 2-5, GATA-4, TBX5, TGF-β, NOTCH, BMP, SOX9 [8]. Similarly, atrial septal development is mediated by genes such as TBX family, GATA-4, NKX 2-5, NOTCH, BMP, HAND, with some of them common to both the structures [10]. Alterations in genes and downstream transcription factors may be associated with abnormalities such as HPML and ASD.

Our patient presented in his sixth decade with symptoms due to large ASD shunting left to right along with HPML. Due to this combination of pathologies, adverse hemodynamic effects of dysplastic MV could have been somewhat mitigated [11]. In a study documenting 68 published cases of HPML, 8 patients (12%) had concomitant OS-ASD, 1 patient (1%) had ostium primum ASD. Thus in HPML patients, ASD was the second most associated congenital heart defect following BAV (23%) [5].

As the septal defect anatomy warranted surgical closure in our patient, a crucial decision was to decide whether and how to manage the MV pathology. According to the latest guidelines, MV repair is preferred over replacement to treat primary MR [12, 13]. However, consensus guidelines are scarce regarding the timing of intervention and surgical techniques to be employed in HPML patients. Studies have shown that in ASD closure patients certain risk factors predispose to worsening MR in the postoperative phase. These include- older age, surgical closure, large LA dimension (>40 mm)/increased LA volume, severity of MR and large indexed ASD size [14, 15].

In our patient, surgical intervention was being performed for a large ASD at an older age with LA enlargement being present. A dilated MV annulus with myxomatous AML prolapse was associated with moderate MR. Hence there was a strong possibility of MR progression postoperatively. Thus a collective decision was made to replace the dysplastic valve instead of repair, considering that a reoperation at older age would entail a much higher surgical risk.

Available data regarding management of adult HPML patients indicates that MV intervention has been performed only when patients were symptomatic due to hemodynamically significant MR. Intervention for HPML was also considered during surgical correction of associated defects, as in our patient [5]. The majority of patients have undergone MV replacement due to anatomical considerations and very limited experience with MV repair [5]. In asymptomatic cases and patients with insignificant MR, medical therapy with clinical follow-up appears to be the right approach.

Long term results are not available for MV repair/ replacement in these patients [4, 16]. Despite MV repair being the desired therapeutic technique, patients must be assessed on a case-to-case basis using Heart team approach. Valvular morphology and associated lesions, age of patient, possibility of postoperative worsening MR, requirement of reoperation and surgical experience are some of the factors which must be

considered to come to a final decision. Transcatheter edge-to-edge repair is not feasible due to absence of PML tissue, but selected cases may be candidates for transcatheter mitral valve replacement; however further studies are required in this regard [5].

STUDY LIMITATION

Due to ours being a single case report study, the clinical presentation and management protocol cannot be generalized for all HPML patients, with or without associated cardiac anomalies. We have postoperative follow-up data of 5 months duration in our patient which shows a good response to therapeutic intervention. A longer duration follow-up is desirable for complete assessment of treatment benefit and cardiac function.

CONCLUSION

Hypoplastic posterior mitral leaflet is a congenital dysplastic mitral valve disease, frequently associated with other cardiac lesions, most commonly bicuspid aortic valve and atrial septal defect. Congenital etiologies must always be considered in the differential diagnosis of adult mitral regurgitation patients. The ideal surgical technique for treating these patients is still not clarified due to rarity of the condition. A Heart team approach is ideal to decide the best therapeutic management protocol.

ABBREVIATIONS

- MV- Mitral valve
- MR- Mitral regurgitation
- HPML- Hypoplastic posterior mitral leaflet
- BAV- Bicuspid aortic valve
- ASD- Atrial septal defect
- VSD- Ventricular septal defect
- OS-ASD- Ostium secundum atrial septal defect
- TTE- Transthoracic echocardiography
- RA- Right atrium
- RV- Right ventricle
- TAPSE- Tricuspid annular plane systolic excursion
- LA- Left atrium
- PML- Posterior mitral leaflet
- AML- Anterior mitral leaflet
- LV- Left ventricle

VIDEO LEGENDS

- **Videos 1-4-** Transthoracic echocardiography in parasternal short axis view, apical 4-chamber view and subcostal view demonstrating atrial septal defect with left to right shunting and dilation of right sided cardiac chambers.
- **Videos 5-7-** Transthoracic echocardiography in parasternal long axis view and parasternal short axis view demonstrating hypoplastic posterior mitral leaflet, myxomatous and elongated sail like anterior mitral leaflet with prolapse and mild to moderate eccentric mitral regurgitation.
- **Videos 8-11-** Transesophageal echocardiography (0–105-degree views) showing large ostium secundum atrial septal defect (left to right shunt) with deficient superior and aortic rims.
- **Videos 12-15-** Transesophageal echocardiography (25–150-degree views) showing hypoplastic posterior mitral leaflet and myxomatous prolapsing anterior mitral leaflet with moderate mitral regurgitation.
- **Videos 16-19-** 3-dimensional transesophageal echocardiography views demonstrating the morphology of hypoplastic posterior mitral leaflet with anterior mitral leaflet prolapse (A2, A3 scallops).

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