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An Echocardiographic Study of a Rare Cause of Mitral Regurgitation: Hypoplastic Posterior Mitral Valve Leaflet

ABSTRACT

Background: The precise etiology of hypoplasia of the posterior mitral valve leaflet (PMVL) remains incompletely elucidated; however, it has been hypothesized to stem from genetic mutations occurring during fetal development. Herein, we present the anatomical characteristics of the mitral valve and associated cardiac pathologies in patients with hypoplastic PMVL.

Methods: This single-center retrospective study involved patients who presented between 2015 and 2021 at a tertiary healthcare facility. Among the cohort, 44 individuals had hypoplastic PMVL and were divided into 2 groups: those with severe mitral regurgitation (MR) and those with non-severe MR.

Results: Among the patients, 11 (25%) had severe MR. The median lengths for the PMVL was 5 mm (5-6). Moreover, 10 patients had concomitant muscular formation. We found that 13 patients had bicuspid aortic valve (BAV), while the second most common concomitant cardiac congenital pathology was secundum atrial septal defect (ASD) in 7 patients. The anterior mitral leaflet (AML) length ($P = .007$), AML prolapse ($P < .001$), and A2P2 distance ($P = .008$) were higher in the group with severe MR. In addition, muscular formation was more common in patients with hypoplastic PMVL with severe MR ($P < .001$).

Conclusion: Hypoplastic PMVL is a rare but significant anomaly that causes MR. While it can coexist with numerous congenital conditions, the most frequent associations include BAV and, secondly, ASD. Severe MR is particularly observed in cases accompanied by dilated mitral annulus, AML prolapse, and muscular formation.

Keywords: Hypoplastic posterior mitral valve leaflet, muscular formation, bicuspid aortic valve, atrial septal defect

INTRODUCTION

Mitral valve complex comprises leaflets, chordae tendineae, papillary muscles, and commissures. Mitral valve regurgitation (MR) arises from deficiencies in any of these anatomical components. Mitral leaflet defects encompass cleft leaflet, accessory mitral valve, aplasia, or hypoplasia of the leaflets.[1] Severe hypoplasia and posterior mitral valve leaflet (PMVL) agenesis (aplasia) are rare causes of MR in adults.

Descriptions of unileaflet mitral valves (partial or complete leaflet agenesis/hypoplasia) are exceedingly rare, mostly confined to case reports.[2-4] Nevertheless, asymptomatic cases have been observed, with a documented prevalence of 1 in 8800 in selected patient cohorts.[5] The precise etiology of hypoplasia of the PMVL remains incompletely elucidated; however, it has been hypothesized to stem from genetic mutations occurring during fetal development. Familial aggregation suggests a potential role of hereditary factors in disease etiology.[6] Herein, the anatomical characteristics of the mitral valve and associated cardiac pathologies in hypoplastic PMVL patients were investigated and presented.

ORIGINAL INVESTIGATION

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METHOD

Study Population

Patients who presented to our hospital between 2015 and 2021 were included in the study. Transthoracic echocardiography (TTE) and transesophageal echocardiography (TEE) data from patients attending the Echocardiography Laboratory during this period were retrospectively reviewed. Within this cohort, 44 individuals were identified with hypoplastic PMVL anomalies. Both symptomatic and asymptomatic patients with MR were included in the study. All the patients underwent both TTE and TEE. The echocardiograms performed during the specified years were retrospectively analyzed, and detailed echocardiographic assessments were carried out at the time of initial diagnosis for those with hypoplastic PMVL, without the need for patient recall for further evaluations.

Clinical, epidemiological, and laboratory data were collected from electronic health records at our institution. Hypertension (HT) was determined by a prior diagnosis, systolic blood pressure (BP) above 140 mm/Hg, or diastolic BP above 90 mm Hg. Diabetes mellitus (DM) was diagnosed based on patient history, fasting plasma glucose levels exceeding 126 mg/dL, random plasma glucose levels surpassing 200 mg/dL, HbA1c values greater than 6.5%, or the use of anti-diabetic medications. This study was performed in line with the principles of the Declaration of Helsinki. Approval granted by the Ethics Committee of our hospital. The research presented in this manuscript was developed without the utilization of artificial intelligence.

Echocardiography

Transthoracic echocardiography examinations were performed utilizing a Philips S5 transducer (Philips Healthcare, Andover, MA, USA), while two-dimensional (2D) and three-dimensional (3D) multiplane TEE were conducted using a Philips IE33 xMatrix system with an X7-2t multiplane probe (Philips Healthcare). Echocardiographic data were comprehensively analyzed offline using commercially available workstation software. The cardiac measurements were evaluated by 2 experienced cardiologists specializing in imaging, and the mean value of the 2 measurements was calculated. Both the intraobserver and interobserver variability were determined to be less than 5%, based on repeated measurements in 10 randomly selected patients. The average of the 2 cardiologists' measurements was used to minimize variability, and we acknowledge that this level of variability could still impact the precision of our findings.

HIGHLIGHTS

- Hypoplastic posterior mitral valve leaflet is a rare but significant anomaly that causes mitral regurgitation.
- Although it can coexist with various congenital conditions, it is most commonly associated with bicuspid aortic valve and atrial septal defect.
- Severe mitral valve regurgitation is particularly observed in cases accompanied by dilated mitral annulus, anterior mitral leaflet prolapse, and muscular formation.

The midesophageal bicommissural view on TEE was utilized to assess mitral valve scallops using the X-plane mode. In the study by Abazid et al[7] in 2023, it was discovered that 3D TEE exhibited significant concordance with the commissural-biplane approach. Similarly, we conducted our mitral valve scallop measurements using this method. The commissural-biplane approach involved acquiring the bicommissural view and simultaneously capturing biplane images of the medial (A3), middle (A2), and lateral (A1) aspects of the anterior mitral valve. The posterior mitral valve leaflet length was measured in the midesophageal long-axis view on TEE but was not evaluated as separate scallops (Figure 1). Three-dimensional echocardiographic images were utilized during TEE (Figure 2). The lengths of the mitral valve leaflets were measured from the inner edge of the annulus to the edge of each respective scallop. A PMVL with a measurement of 8 mm or less in males and 7 mm or less in females across all of the scallops was considered indicative of a hypoplastic PMVL.[8] The muscular extension of the posterior wall of the left ventricle was also defined as a muscular formation, as it was in previous cases (Figure 3).

In all patients, pulmonary veins were evaluated using TEE. Additional examinations were not performed in patients without clinical or echocardiographic suspicion of anomalies. Such examinations were only conducted in cases where there was suspicion of partial anomalous pulmonary venous return (PAPVR).

Although 3D echocardiography was performed on many of the patients, it was not applied to all of them. Specifically, 3D echocardiography was conducted in 30 out of the 44 patients, while in the remaining 14 patients, only 2D echocardiography was used. Unfortunately, some patients have since passed away, undergone surgical valve replacement, or were lost to follow-up, making it impossible to perform additional evaluations.

Statistical Analysis

Statistical Package for Social Sciences (SPSS) version 25 was used for statistical analysis (IBM Corp., Armonk, New York, USA). The normal distribution of numerical variables was assessed using the Shapiro–Wilk normality test, histogram, skewness, kurtosis, P–P plot, and Q–Q plot tests. Descriptive statistics were presented as number of units (n), percentage (%), mean ± standard deviation, median, and interquartile range values. The homogeneity of variances was evaluated using the Levene test. Two-group comparisons for numerical variables were performed using the independent samples *t*-test when the data were normally distributed, and the Mann–Whitney *U* test when they were not. Pearson's and Fisher exact chi-square tests were used to compare groups with categorical variables. A *P*-value <.05 was considered statistically significant.

RESULTS

This study included 44 patients with hypoplastic PMVL. Baseline clinical and echocardiographic characteristics of the patients are shown in Table 1. The median age of the patients was 31 years. Of the patient group, 22 (50%) were

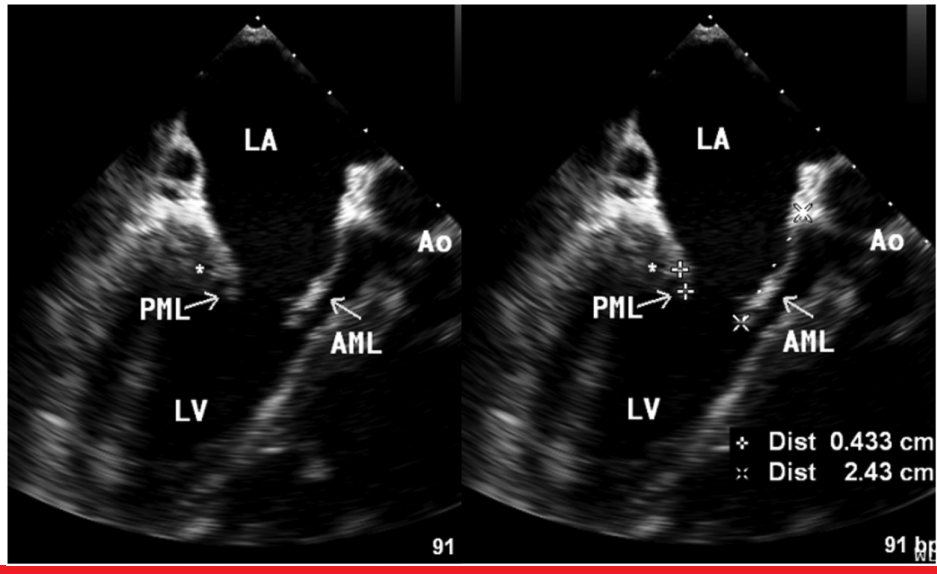

Figure 1. Transesophageal echocardiography in the midesophageal long-axis view demonstrated measurements of the anterior mitral valve leaflet (AML) and posterior mitral valve leaflet (PML).

male. Of the 44 patients included in the study, 32 were symptomatic and 12 were asymptomatic. Among the symptomatic patients, 20 were classified as New York Heart Association classification (NYHA) class I-II, 10 as class III, and 2 as class IV. Moreover, 11 patients (25%) had severe MR. Among the patients, 4 (9.1%) had HT, 3 (6.8%) had DM, and 4 (9.1%) were smokers. Coronary artery disease was present in 1 patient (2.3%), and cerebrovascular disease was also found in 1 patient (2.3%). When the patients' mitral valve scallops were evaluated, the mean lengths of the A1, A2, and A3 scallops were 28.95 ± 4.36 mm, 31.52 ± 3.31 mm, and 29.43 ± 4.09 mm, respectively. The median lengths for the PMVL was 5 mm (5-6). The anterior mitral leaflet (AML) had a mean length of 33.10 ± 3.70 mm, with 8 patients (18.2%) exhibiting AML prolapse. The A2P2 annulus diameter measured 32.38 ± 5.01 mm, and the mediolateral annulus diameter was 34.14 ± 3.55 mm. We found that 10 patients had concomitant muscular formation, and 28 (64%) patients had concomitant cardiac pathologies in addition to hypoplastic PMVL. The most common concomitant pathologies were aortic valve pathologies, wherein 13 patients had bicuspid aortic valve (BAV) and 2 patients had unicuspid unicommissural aortic valve. The second most common concomitant cardiac congenital pathology was atrial septal defect (ASD), wherein 7 patients had secundum ASD and 1 patient had primum ASD (Figure 4). Of the 11 patients with severe MR, 8 underwent surgical intervention (mitral valve replacement (MVR), while 3 received medical management due to high surgical risk or patient preference. Additionally, among the 14 patients with moderate MR, 5 underwent surgery for MVR due to concomitant cardiac pathologies. It is important to mention that mitral valve repair was not performed on any of our patients.

The characteristics of patients with and without severe MR are presented in Table 2. Among the clinical characteristics of the patients, only age was associated with severe MR. Regarding the echocardiographic characteristics, the ejection fraction ($P = .032$), end-diastolic diameter ($P = .013$),

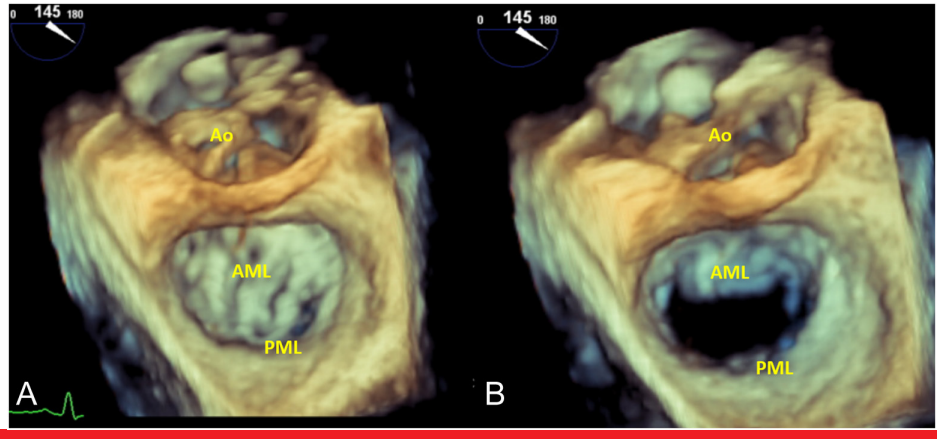

Figure 2. Three-dimensional transesophageal echocardiography short axis mid-systolic (A) and mid-diastolic view of the hypoplastic posterior mitral valve leaflet (B).

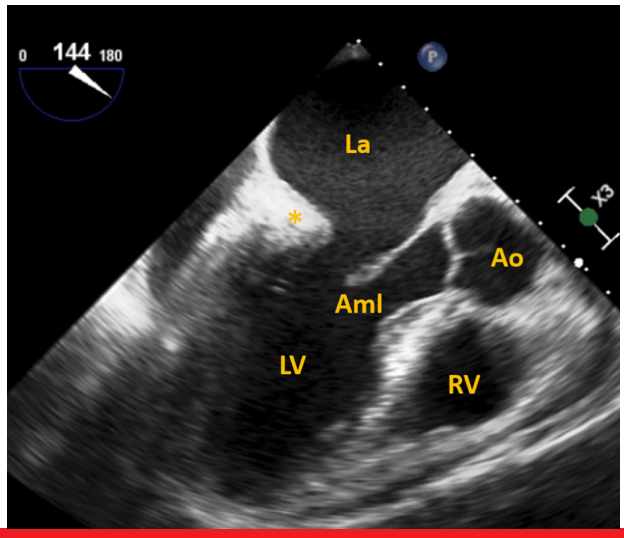

Figure 3. Transesophageal echocardiography midesophageal long-axis view showing the protrusion from the left ventricular free wall (asterisk) referred to as muscular formation.

end-systolic diameter ($P = .010$), and left atrium diameter ($P < .001$) were higher in the severe MR group. Regarding the mitral valve characteristics, the AML length ($P = .007$), AML prolapse ($P < .001$), and A2P2 distance ($P = .008$) were higher in the group with severe MR. In addition, patients with hypoplastic PMVL with severe MR more frequently had concomitant muscular formation ($P < .001$) (Figure 5).

DISCUSSION

Although hypoplastic PMVL has been described in prior case reports and case series, this study is, to the best of our knowledge, among the first to specifically focus on this group of patients. In the present study, the PMVL characteristics and concomitant congenital cardiac pathologies in patients with hypoplastic PMVL were demonstrated.

The study conducted by Rusted et al[8] on 50 cadavers revealed that the average length of the posterior leaflet of the mitral valve was 1.2 cm in females, with a range between 0.7 and 2.4 cm. In males, the average length was slightly longer at 1.3 cm, with a range spanning from 0.8 to 1.8 cm. The free edge of the posterior leaflet is often divided into 3 or more scallops or described as lateral, middle, and medial or segments P1, P2, and P3.[9] Shree et al[10] conducted a study on 50 cadaveric hearts and demonstrated the presence of a variable number of scallops in the posterior leaflet, ranging from single, double, triple, quadruple, or tetra–scalloped, with the most notable configurations being 6 or hexa–scalloped. Although 3 scallops are most common, the scallops are not equal in size. Ranganathan et al[11] found the P2 scallop to be larger in the majority of hearts. However, although there are autopsy studies regarding the lengths of mitral valve scallops, there is no large-scale echocardiographic study determining the lengths of normal mitral valve scallops. In our study, we were also unable to evaluate the PMVL as separate scallops in patients with hypoplastic PMVL due to insufficient tissue.

Table 1. Baseline Clinical and Echocardiographic Features of the Patients with Hypoplastic Mitral Valves

Variables	n = 44
Baseline characteristics	
Age (years), median (IQR)	31 (23-44.5)
Gender (male), n (%)	22 (50)
HT, n (%)	4 (9.1)
DM, n (%)	3 (6.8)
Smoker, n (%)	4 (9.1)
Coronary artery disease, n (%)	1 (2.3)
Cerebrovascular disease, n (%)	1 (2.3)
Asymptomatic, n (%)	12 (27.3)
Symptomatic, n (%)	32 (72.7)
NYHA class I-II, n (%)	20 (45.4)
NYHA class III, n (%)	10 (22.7)
NYHA class IV, n (%)	2 (4.5)
Echocardiographic findings	
Ejection fraction (%), median (IQR)	65 (62.5-65)
LVEDD (cm), median (IQR)	4.5 (4.1-5.25)
LVESD (cm), median (IQR)	2.8 (2.2-3.45)
LAD (cm), mean ± SD	3.36 ± 0.72
IVS (cm), mean ± SD	1.05 ± 0.19
LVPW (cm), median (IQR)	1 (0.9-1)
Ascending aorta (cm), median (IQR)	3.1 (2.8-3.5)
Right ventricle base (cm), mean ± SD	3.78 ± 0.74
MR, n (%)	
None-mild	19 (43.2)
Moderate	14 (31.8)
Severe	11 (25)
Mitral stenosis, mild, n (%)	2 (4.5)
Aortic regurgitation, n (%)	
None	29 (65.9)
Mild	3 (6.8)
Moderate	3 (6.8)
Severe	9 (20.5)
Aortic stenosis, n (%)	
None	40 (90.9)
Mild	0 (0)
Moderate	2 (4.5)
Severe	2 (4.5)
Tricuspid regurgitation, n (%)	
None	15 (34.1)
Mild	28 (63.6)
Moderate	0 (0)
Severe	1 (2.3)
Pulmonary regurgitation, n (%)	
None	40 (90.9)
Mild	3 (6.8)
Moderate	1 (2.3)
Severe	0 (0)

(*Continued*)

Table 1. Baseline Clinical and Echocardiographic Features of the Patients with Hypoplastic Mitral Valves

Variables	n = 44
Characteristics of the mitral valve	
A1 length (mm), mean ± SD	28.95 ± 4.36
A2 length (mm), mean ± SD	31.52 ± 3.31
A3 length (mm), mean ± SD	29.43 ± 4.09
PML length (mm), median (IQR)	5 (5-6)
AML length (mm), mean ± SD	33.10 ± 3.70
AML prolapse, n (%)	8 (18.2)
A2P2 annulus diameter (mm), mean ± SD	32.38 ± 5.01
Mediolateral annulus diameter (mm), mean ± SD	34.14 ± 3.55
Additional characteristics of congenital heart conditions	
BAV, n (%)	13 (29.5)
Unicuspid unicommissural aortic valve	2 (4.5)
Secundum ASD, n (%)	7 (15.9)
Sinus venosus ASD, n (%)	1 (2.3)
Ventricular septal defect, n (%)	3 (6.8)
PAPVR, n (%)	2 (4.5)
PDA, n (%)	1 (2.3)
Hypertrophic cardiomyopathy, n (%)	2 (4.5)
Muscular formation, n (%)	10 (22.7)

AML, anterior mitral leaflet; ASD, atrial septal defect; BAV, bicuspid aortic valve; DM, diabetes mellitus; HT, hypertension; IQR, interquartile range; IVS, interventricular septum; LAD, left atrial diameter; LVEDD, left ventricular end-diastolic diameter, LVESD, left ventricular end-systolic diameter; LVPW, left ventricular posterior wall; PAPVR, partial anomalous pulmonary venous return; PDA, patent ductus arteriosus.

The absence of the PMVL typically leads to fetal demise in utero. Hypoplasia of the PMVL has been linked with stenosis of the supravalvular mitral ring.[12] Additionally, individuals with this anomaly may tolerate the condition into adulthood, with the gradual development of MR stemming from annular dilation. The prognosis for asymptomatic patients with hypoplastic mitral valves is uncertain. There is a potential risk of worsening MR, primarily due to annular dilation, which may lead to increased morbidity and mortality.[6] In this study, annular dilation of the mitral valve was found to be potentially associated with severe MR. The A2P2 distance was greater in patients with severe MR. Furthermore, AML prolapse, which may occur as a compensatory mechanism for annular dilation, was more commonly observed in the severe MR group. It was also found herein that the severe MR group had a longer AML.

Furthermore, in our study, differences were observed in left atrial diameter (LAD), left ventricular end-systolic diameter (LVESD), and left ventricular end-diastolic diameter (LVEDD) between individuals with severe MR and those without. However, it is possible that this difference may also be influenced by the duration of exposure to severe MR. Nevertheless, we do not have data related to this exposure duration. Additional studies are required to address this issue.

Distinguishing hypoplastic PMVL from rheumatic mitral valve disease is crucial. In hypoplastic PMVL cases, thickened and restricted leaflets may be observed, mimicking the appearance of end-stage rheumatic valve disease. However, in cases where rheumatic factors are implicated, it typically takes several decades for severe stenotic valvulopathy to develop after recurrent episodes of carditis.

Muscular formation is a cardiac pathology accompanying hypoplastic PMVL patients.[13-15] The muscular formation, which replaces the posterior leaflet, represents a primordial chordal apparatus adhering to the endocardium of the left ventricle posterior wall. During embryological development, the endocardial elements of the valvular primordia undergo infiltration and replacement by myocardial tissue. Concurrently, the sponge layer of the myocardium undergoes compaction and forms distinct papillary muscles. These myocardialized atrioventricular valvular leaflets and chordae then undergo collagenization, transforming into thin, delicate connective structures. Muscular formation may be attributed to a developmental arrest that occurs at this stage of transformation from muscle chords and leaflets

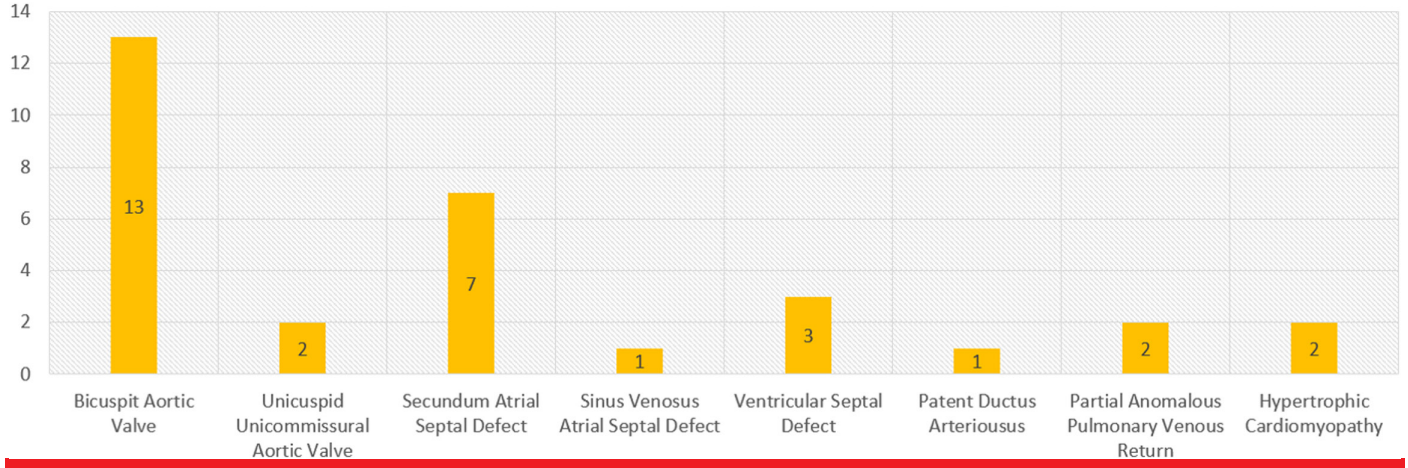

Figure 4. Distribution of concomitant congenital heart diseases in patients with hypoplastic posterior mitral valve leaflet.

Table 2. Comparison of the Clinical and Echocardiographic Characteristics of the Patients with Hypoplastic PMVL According to the Presence of Severe MR.

Variables	MR Severe (+)	MR Severe (–)	*P*
	n = 11	n = 33	
Significant parameters			
Age (years), median (IQR)	40 (33-66)	31 (21.5-42.5)	**.025**
Ejection fraction (%), median (IQR)	60 (55-65)	65 (61.25-65)	**.032**
LVEDD (cm), median (IQR)	5.5 (4.5-6.3)	4.5 (4.1-5.2)	**.013**
LVESD (cm), median (IQR)	3.6 (3.1-4.4)	2.6 (2.4-3.45)	**.010**
LAD (cm), mean ± SD	4.26 ± 0.73	3.27 ± 0.64	**< .001**
AML length (mm), mean ± SD	37.22 ± 4.63	32.96 ± 3.48	**.007**
A1 length (mm), mean ± SD	33.89 ± 5.90	29.10 ± 3.80	**.009**
Anulus diameter-A2P2 (mm), mean ± SD	39 ± 5.09	32.68 ± 5.57	**.008**
AML prolapse, n (%)	6 (54.54)	2 (6.06)	**<.001**
Muscular formation, n (%)	7 (63.63)	3 (9.09)	**<.001**
Nonsignificant parameters			
Gender (male), n (%)	7 (63.63)	15 (45.45)	.296
HT, n (%)	2 (18.18)	2 (6.06)	.256
DM, n (%)	1 (9.09)	2 (6.06)	.588
Smoker, n (%)	2 (18.18)	2 (6.06)	.256
Coronary artery disease, n (%)	1 (9.09)	0 (0)	.250
Cerebrovascular disease, n (%)	1 (9.09)	0 (0)	.250
IVS (cm), mean ± SD	1.12 ± 0.16	1.11 ± 0.37	.970
LVPW (cm), median (IQR)	1 (1-1)	1 (1-1)	.858
Ascending aorta (cm), median (IQR)	3.6 (3-4.2)	3.1 (2.8-3.45)	.096
Right ventricle base (cm), mean ± SD	4.08 ± 0.44	3.72 ± 0.78	.340
A2 length (mm), mean ± SD	34.11 ± 4.26	31.56 ± 2.98	.059
A3 length (mm), mean ± SD	32.11 ± 6.79	29 ± 2.81	.215
PML length (mm), median (IQR)	4 (3-6)	5 (5-6)	.334
Anulus diameter-mediolateral (mm), mean ± SD	37.5 (36-40.5)	33 (31.5-38.5)	.081

AML, anterior mitral leaflet; DM, diabetes mellitus; HT, hypertension; IQR, interquartile range; IVS, interventricular septum; LAD, left atrial diameter; LVEDD, left ventricular end-diastolic diameter, LVESD, left ventricular end-systolic diameter; LVPW, left ventricular posterior wall; PDA, patent ductus arteriosus.

to thin connective structures. The presence of myocytes in biopsy specimens of the posterior valvular remnants substantiates this hypothesis.[15] In the current study, muscular formation appeared to be more common in the severe MR group.

There are various other cardiac anomalies reported in hypoplastic PMVL patients. In the patients with hypoplastic PMVL in the present study, the most common accompanying congenital cardiac pathology was aortic valve diseases. Among aortic valve diseases, BAV is the most common, as demonstrated in many cases in the literature.[5,16,17] Additionally, a unicuspid unicommissural aortic valve was detected in only 2 of the patients. In the current study, the second most common accompanying pathology in patients with hypoplastic PMVL was ASD. Hypoplastic PMVL with ASD was also observed in studies by Pourafkari et al,[13] Heper et al,[18] and Bhardwaj et al.[19] The predominant type of ASD among patients is secundum ASD, although 1 patient was identified with a primum ASD. In 3 patients among the current cohort, ventricular septal defects were identified, and anomalies in pulmonary venous return were observed in 2 patients. These concomitant findings with hypoplastic PMVL have not been previously documented in the literature. Patients with hypoplastic PMVL are often associated with numerous hereditary pathologies, implying a hypothesis that genetic mutations occurring during fetal development may contribute to its etiology. Cases demonstrating familial clustering have also been documented, indicating a possible involvement of hereditary factors in the pathogenesis of the disease.[6,14] However, familial clustering was not observed within the current patient cohort. Moreover, in a considerable number of the patients in the present study with hypoplastic PMVL, there was no concomitant additional cardiac pathology.

In patients with hypoplastic PMVL, the most significant valve issue is severe mitral insufficiency. In the literature, valve replacement is commonly performed in many hypoplastic PMVL patients with severe MR.[3,20-22] In cases reported by Parato et al[21] and Joshi et al,[22] mechanical MVR was performed. However, Bacich et al[3] preferred biological MVR in elderly patients. In the literature, various surgical approaches for managing hypoplastic PMVL in pediatric patients have

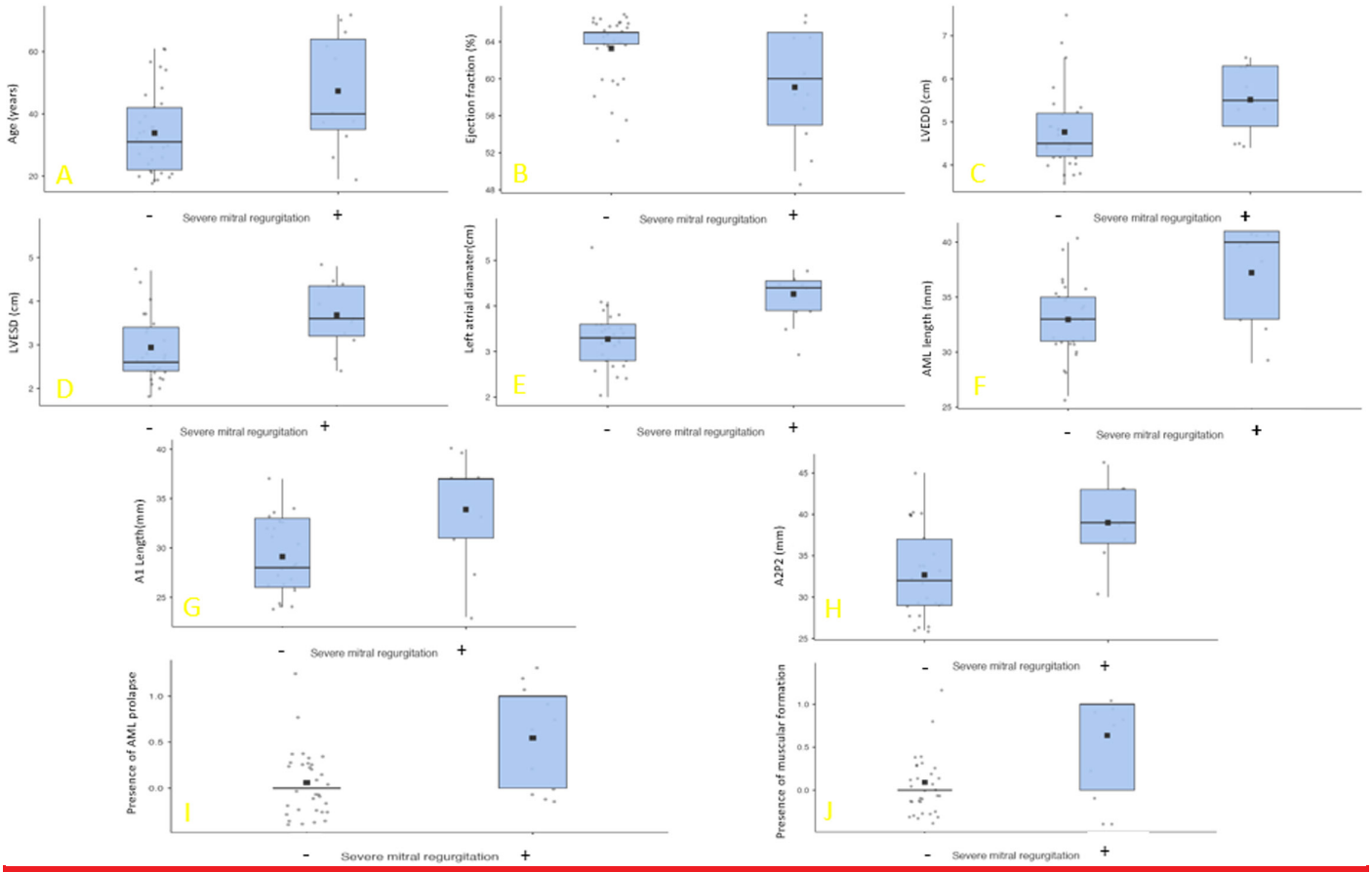

Figure 5. The significant results for severe mitral regurgitation in patients with hypoplastic posterior mitral valve were presented as a box plot graph.

been discussed. Kalangos et al[15] presented a case involving a 10-year-old girl who underwent annuloplasty, with a follow-up period of 12 months. Similarly, Caciolli et al[23] detailed the treatment of a 14-year-old girl with a comparable condition using annuloplasty, although no long-term follow-up was documented. Stojanovic et al[24] employed pericardial patches to augment the posterior leaflet area, in addition to annuloplasty. The patient, who was 29 years old, was the oldest among those who underwent surgical mitral valve repair due to hypoplastic PMVL. Despite favorable postoperative outcomes, only a 6-month follow-up was reported. Even with these findings, the limited evidence available does not strongly support the use of mitral valve repair in such cases, mainly because of the rarity of hypoplastic PMVL conditions, leading to insufficient description of long-term outcomes. Gupta et al[25] found that AML length is a strong predictor of mitral valve reparability in a rheumatic population. Similarly, in our view, achieving a successful repair may require that the anterior leaflet be longer than the anteroposterior annular diameter.

Currently, no genetic analyses are available for the patients included in our follow-up. However, we recognize the importance of genetic studies in understanding the potential hereditary factors and mutations associated with hypoplastic PMVL. This area represents a promising direction for future research. Although all of our patients underwent TTE and TEE, cardiac magnetic resonance imaging (MRI) could provide a more comprehensive assessment of hypoplastic PMVL and associated cardiac pathologies. Since no studies have been conducted using cardiac MRI for this purpose in the literature, this represents an area for potential future investigation.

Study Limitations

This study had several limitations. First, being a single-center cross-sectional observational study, there was the possibility of selection bias. Therefore, the results should be validated in larger, more diverse populations. Second, our center serves as a reference hospital due to its status as a high-volume cardiac center. Therefore, the incidence of hypoplastic PMVL cannot be extrapolated to the general population, and our patient selection is not consecutive. Prospective and multicenter large-volume studies are imperative to elucidate the anatomical characteristics of the mitral valve in patients with hypoplastic PMVL and to understand the associated cardiac pathologies within this patient cohort. Another limitation of our study is the potential impact of interobserver and intraobserver variability on the precision of cardiac measurements, despite efforts to minimize it by averaging the measurements taken by 2 experienced cardiologists. An additional limitation is the lack of cardiac MRI, which could provide a more comprehensive assessment of muscular formation.

CONCLUSION

In conclusion, hypoplastic PMVL is a rare but significant anomaly that causes mitral regurgitation. While it can coexist with numerous congenital conditions, the most frequent associations include BAV and, secondly, ASD. Severe MR is particularly observed in cases accompanied by dilated mitral annulus, AML prolapse, and muscular formation.

Data Availability: The data that support the findings of this study are available upon request from the corresponding author

Ethics Committee Approval: This study was performed in line with the principles of the Declaration of Helsinki. Approval granted by the Ethics Committee of Koşuyolu Heart Training and Research Hospital on 02.07.2024 (decision number 2024/12/853).

Informed Consent: Written informed consent was obtained from all participants who participated in this study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – G.K., A.K.; Design – Z.B., M.S.; Supervision – A.K., G.K.; Resources – C.Y., İ.B.; Materials – M.A., A.K.; Data Collection and/or Processing – İ.B., C.Y.; Analysis and/or Interpretation – M.A., M.S.; Literature Search – C.Y., İ.B.; Writing – Z.B., M.S.; Critical Review – G.K., A.K.

Declaration of Interests: The authors have no conflicts of interest to declare.

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