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ORIGINAL ARTICLE

High Prevalence of Scoliosis in Children with Mitral Valve Prolapse

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Abstract

OBJECTIVES: This study aims to evaluate the prevalence and characteristics of scoliosis in asthenic children diagnosed with mitral valve prolapse (MVP).
MATERIAL AND METHODS: This study aims to characterize scoliosis prevalence and examine relationships with mitral valve morphology, regurgitation severity, and skeletal phenotypes (pectus excavatum, hypermobility, skin laxity) in asthenic children with primary MVP based on echocardiographic findings. Individuals with known systemic connective tissue disorders were excluded. Scoliosis was assessed radiographically using Cobb angle measurements with a threshold of ≥10° for diagnosis. Statistical analysis was performed using SPSS 26.0.
RESULTS: The study included 49 asthenic children (25 females, 24 males) aged 9 to 17 years who were diagnosed MVP. The mean age of participants was 13.2 ± 2.2 years. Physical examination revealed pectus excavatum in 46.9%, joint hypermobility in 39%, and skin laxity in 16.3% of patients. Scoliosis (Cobb ≥10°): 47% (23/49, [95% CI: 33–61%]); potential scoliosis (5–10°): 27% (13/49, [95% CI: 16–40%]); any abnormal curvature: 73% (36/49, [95% CI: 59–84%]). Valve morphology: diffuse thickening 61% (30/49); localized elongation 39% (19/49). No significant association between scoliosis and valve morphology ($p > 0.05$) or MR severity ($p > 0.05$). Pectus excavatum significantly associated with elongated valve structure ($p = 0.001$). Pectus excavatum was significantly more common in patients with an elongated, straight valve structure ($p = 0.001$).
CONCLUSIONS: High scoliosis prevalence (47%) in asthenic MVP children is independent of cardiac severity, suggesting intrinsic skeletal tissue vulnerability. Neuroendocrine and immune mechanistic investigation warranted.

INTRODUCTION

Mitral valve prolapse represents a common cardiac condition characterized by the displacement of one or both mitral valve leaflets into the left atrium during systole, potentially leading to mitral regurgitation. The reported incidence of MVP is 2% to 5% in the pediatric population and the prevalence of MVP increases with age (Bouknight & O'Rourke, 2000). The underlying cause involves fibromyxomatous

changes affecting the mitral valve leaflets, leading to their displacement into the left atrium and subsequent late-systolic regurgitation (Vriz *et al.* 2022). One or both mitral valve leaflets bulge by at least 2 mm into the left atrium during systole and thickening of the involved leaflet to more than 5 mm supports the diagnosis. Some pediatric patients with characteristic body build and auscultatory findings of the condition, do not show the adult echo criterion of MVP; they may only show regional thickened mitral leaflets with systolic straightening or systolic superior doming and some posterosuperior displacement of the coaptation point of the mitral valve because MVP is a progressive disease (Boudoulas *et al.* 2020; Moller & Hoffman, 2020). Diffuse thickening of the valve is usually referred to as Barlow's valve, whereas regional thickening of the mitral valve often associated with elongated chordae tendineae is referred to as a fibroelastic deficiency valve (Boudoulas, 2024) .

Scoliosis is a three-dimensional spinal deformity that is often diagnosed in childhood and adolescence. It is characterized by lateral curvature and vertebral rotation. The Scoliosis Research Society's classification system, established in 1973, remains in use today (Terminology Committee,1976). Because it is often hidden by clothing, the symptoms and signs may go unnoticed, especially in the early stages. However, early detection and treatment are possible with regular screenings especially in high-risk patients. The risk of progression is also age-dependent, being higher at earlier ages and decreasing after menarche in girls.

Asthenic children are known to have a higher predisposition to both MVP and spinal deformities. Although MVP is commonly associated with connective tissue disorders, the isolated presentation of MVP in asthenic children has not been adequately investigated regarding scoliosis risk. This study aims to assess the prevalence and clinical associations of scoliosis in asthenic pediatric patients with MVP.

Tab. 1. Clinical and Echocardiographic Characteristics of Patients

Parameter	n (%) / Mean ± SD
Gender	
- Female	25 (51%)
- Male	24 (49%)
Age (years)	13.2 ± 2.2
Physical Findings	
- Pectus excavatum	23 (46.9%)
- Joint hypermobility	19 (39%)
- Skin laxity	8 (16.3%)
Echocardiographic Features	
- Diffuse mitral thickening	30 (61.2%)
- Localized leaflet changes	19 (38.8%)
- Moderate/severe MR	4 (8.1%)
Spinal Curvature	
- Scoliosis (Cobb ≥10°)	23 (47%)
- Potential scoliosis (5–10°)	13 (26.5%)
- No scoliosis (<5°)	13 (26.5%)
Curve Severity (if scoliosis)	
- Mild (10–20°)	20
- Moderate (20–40°)	3
Curve Localization	
- Thoracic	12
- Thoracolumbar	12
- Lumbar	8
- Combined	4

MATERIAL AND METHOD

Participants

49 asthenic children (age 9–17 years) with echocardiographically confirmed primary MVP (pMVP, defined as ≥2 mm systolic displacement with or without ≥5 mm leaflet thickening). Asthenic habitus: BMI <10th percentile (WHO standards) + narrow thorax + long limbs + hyperlordotic posture. Exclusion: genetic testing confirmed absence of Marfan syndrome (FBN1), Ehlers-Danlos syndrome (COL3A1, COL5A1), or other syndromic connective tissue disorders Chest wall deformities, joint hypermobility, and skin laxity were recorded. Echocardiographic evaluation focused on mitral valve morphology (diffuse or localized thickening) and regurgitation severity. Standing anteroposterior spinal radiographs were used for scoliosis evaluation. Cobb angles were measured; angles of 5°–10° were considered potential scoliosis (because there may be progression with puberty), and angles ≥10° were defined as scoliosis (Figure 1,2,3). Scoliosis severity was classified as mild (10°–20°) or moderate (20°–40°), and replacement of scoliosis was classified as thoracic, thoracolumbar, lumbar, thoracic plus lumbar according to the curvature (6). Statistical analyses were performed using SPSS 26.0. Chi-square and Pearson correlation tests were used; $p < 0.05$ indicated statistical significance.

This study was approved by the Başkent University Institutional Review Board;Project No: KA 25/120.

RESULTS

The study cohort consisted of 25 females and 24 males, with a mean age of 13.2 ± 2.2 years. The clinical and echocardiographic characteristics of the patients are summarized in Table 1. The most common reasons for visiting the clinic were, in order of frequency, palpitations (36%), chest pain (20%), vasovagal syncope

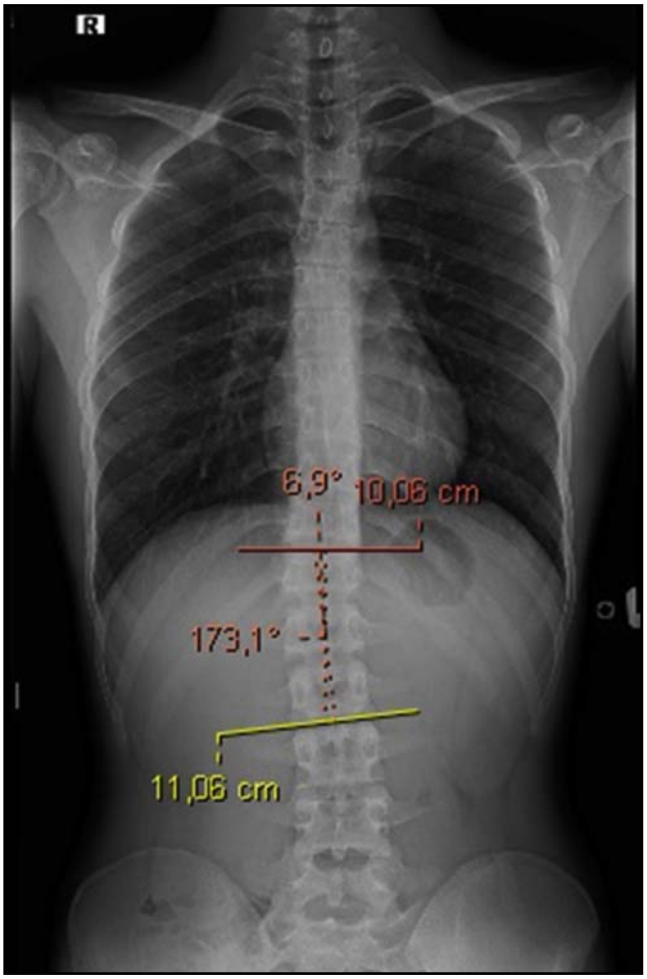

Fig. 1. Potential scoliosis with a Cobb angle of 6.9°, a potential for scoliosis in the thoracolumbar region.

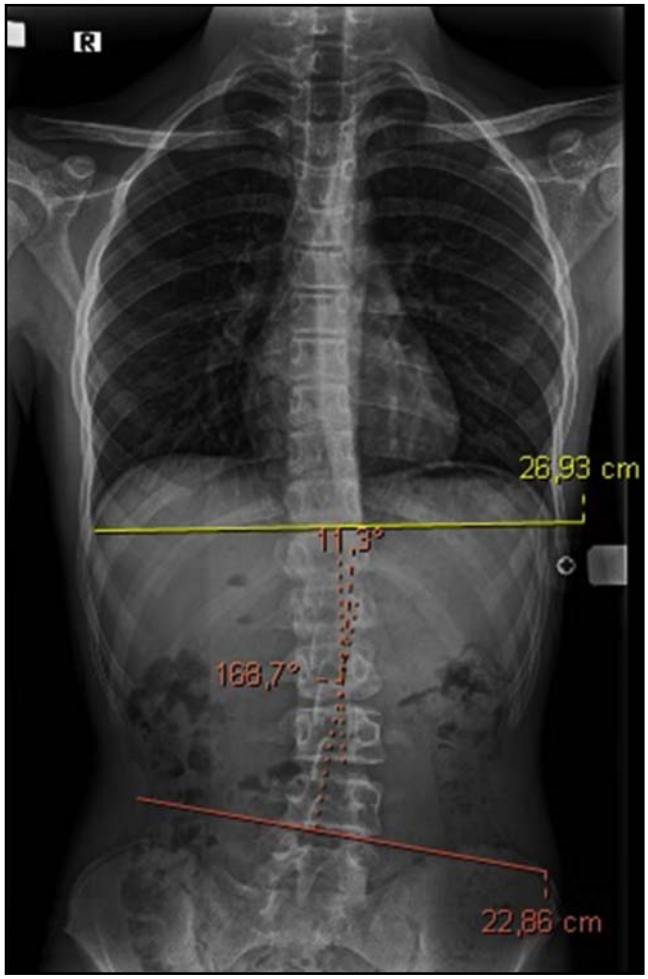

Fig. 2. Mild lumbar scoliosis with Cobb angle of 11.3°.

(16%), reduced exercise tolerance (14%), and breathlessness with exertion (12%). Pectus excavatum was observed in 23 patients (46.9%), joint hypermobility in 19 (39%), and skin laxity in 8 (16.3%). Diffuse mitral valve thickening was identified in 30 patients (13 patients had trivial, 8 had mild, 6 had moderate mitral regurgitation and 3 patients without regurgitation) and localized thickening with elongated chordae tendineae in 19 (8 patients had trivial mitral regurgitation and 1 patient had without regurgitation). The frequency of mitral valve prolapse was statistically higher in girls than in boys (p = 0,001). In 36 patients (73%), radiographs showed varying degrees of abnormal curvature of the spine. 13 patients had potential scoliosis (Cobb Angle between 5°-10°) and 23 patients (47%) had scoliosis (Cobb Angle ≥10°)). Among those with scoliosis, 20 had mild and 3 had moderate scoliosis. Moderately scoliotic patients were post-pubertal girls with a thick, bulging valve structure, with two requiring surgery and one undergoing physical therapy (Figure 4). The localization of scoliosis was thoracic in 12 patients, thoracolumbar in 12 patients, lumbar in 8 patients, and combined in 4 patients. Only 5 out of 36 patients' families were aware that their children had scoliosis. Families were informed that scoliosis may progress as prepubescent children grow older. They were also informed about the importance of sports and physical therapy, as well as the need for regular follow-ups. Bivariate associations tested: valve morphology and scoliosis (p > 0.05), MR severity and scoliosis (p > 0.05), pectus excavatum and scoliosis (p > 0.05), joint hypermobility and scoliosis (p > 0.05), skin laxity and scoliosis (p > 0.05). Only significant finding: pectus excavatum significantly associated with elongated valve morphology (p = 0.001; no correction for multiple comparisons). Note: univariate chi-square tests performed; no multivariate regression or adjusted analyses conducted.

DISCUSSION

This study highlights the high prevalence of scoliosis in asthenic children with MVP, with nearly three-quarters of the participants affected. This rate is significantly higher than the expected prevalence in the reports of MVP and scoliosis coincident (Chan, 1983). Patients

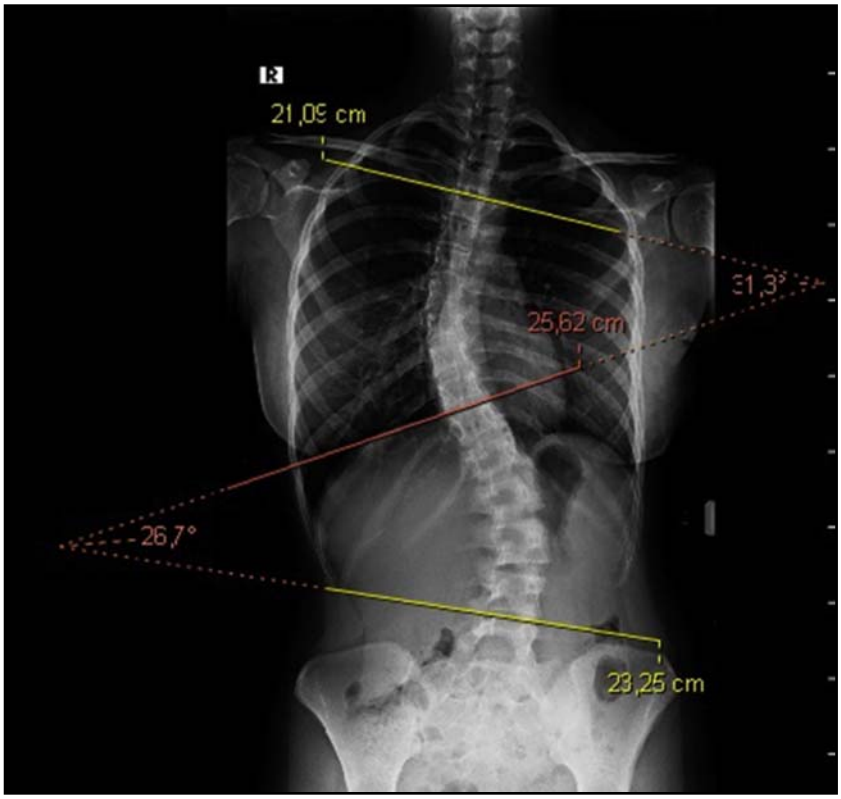

Fig. 3. Moderate thoracal + lumbar scoliosis with a Cobb angles of 31° (thoracic) and 26.7° (lumbar).

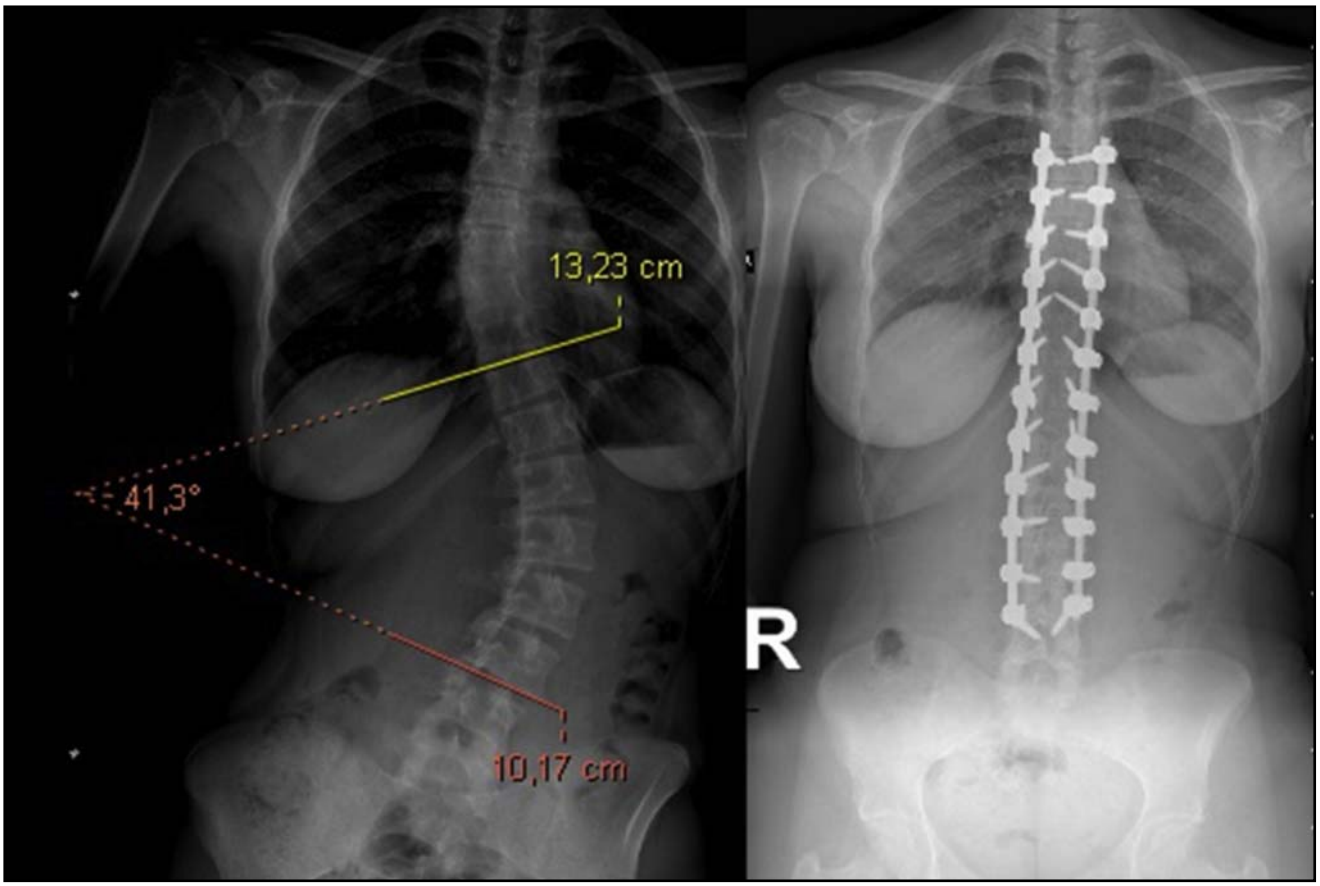

Fig. 4. Pre- and postoperative radiographs of a patient with severe scoliosis (Cobb angle 41.3°)

with mitral valve prolapse and asthenic build are usually asymptomatic, but a history of non-exertional chest pain, palpitation, and, rarely, syncope may be elicited. Pectus excavatum, pes planus, increased joint mobility, skin laxity and scoliosis have been reported as accompanying skeletal anomalies with primary MVP. The inclusion of radiographically confirmed early curvatures suggests the need for screening in at-risk populations.

Implicit in restructured Results; clarified as 47% (23/49) with 26.5% (13/49) potential scoliosis = 73.5% total abnormal curvature. This high prevalence was related to the early radiography scanning before the onset of externally apparent postural deformity (Hirschfeld *et al.* 1982; Kumar & Sahasranam, 1991).

Although no direct correlation was observed between mitral valve morphology or regurgitation severity and scoliosis, the coexistence of these conditions suggests an underlying connective tissue vulnerability. Studies by Freed *et al.* (Freed, 1999) and Yetman *et al.* (Yetman & McCrindle,1999) have reported an increased frequency of musculoskeletal abnormalities, including scoliosis, in patients with MVP, particularly in the context of non-syndromic presentations. The presented cases with figures highlight the different spectrums of scoliosis observed in asthenic patients with primary MVP. The early detection of mild and moderate curvatures is essential for curve progression risk in untreated scoliosis during growth (Lonstein & Carlson). Timely orthopedic referral and follow-up may prevent progression to severe deformities requiring surgical intervention.

Scoliosis localisation was not limited to the thoracic region, but was found in four different locations with similar frequency in patients with pectus excavatum. As children with pectus excavatum are assessed with regular chest radiographs in clinical practice, the possibility of a concomitant scoliosis should be kept in mind and the screening should be carried out to include all the spinal vertebrae.

Scoliosis appears to be more prevalent in asthenic children with MVP, underscoring the need for interdisciplinary management involving cardiologists and orthopedic specialists. In particular, the awareness of families and follow-up physicians is to be increased through scoliosis screening. Regular screening and early intervention may help prevent progression and associated complications, ultimately improving patient outcomes.

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ETHICAL APPPROVAL

Ethical approval was granted by the Baskent University Institutional Review Board (Project No: KA 25/120).

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