



CLINICAL SPECTRUM OF SKELETAL ABNORMALITIES AND MITRAL VALVE PROLAPSE AND THEIR CLINICAL IMPLICATIONS

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Fifty-six patients were diagnosed to have mitral valve prolapse (MVP) syndrome by auscultatory criteria, confirmed by cross-sectional echocardiography. Complete physical examination and x-ray of the bony thorax revealed bony deformities which were subdivided into four main groups according to the predominant deformity. Shallow chest with pectus excavatum, straight back, kyphoscoliosis and elliptical chest in marfanoid patients were reported in the cases studied. It is concluded that musculoskeletal abnormalities have to be considered as nonauscultatory features of MVP. Therefore, any patient with musculoskeletal deformity has to be screened for MVP by cross-sectional echocardiography to prevent life-threatening complications. *Ann Saudi Med* 1996;16(3): 266-268.

Over the past 25 years, mitral valve prolapse (MVP) has been extensively studied, and its echocardiographic, cine-angiographic, and clinical manifestations have become well-known. The prevalence of echocardiographic features and/or auscultatory signs of mitral valve prolapse has been high among the supposedly healthy populations among Western societies,¹⁻³ as well as among the Saudi population.^{4,5} The high prevalence confirms the statement, "For every symptomatic patient with MVP, there are hundreds of asymptomatic persons."⁶ Hunt and Sloman reported a high prevalence of MVP among females in early adulthood and concluded that this disorder has a familial tendency.⁷

It must be remembered that there is no single golden rule for diagnosing MVP. A combined approach using the clinical signs of systolic click and murmur together with diagnostic echocardiographic findings probably provides the clearest definition of prolapse of the mitral valve, and patients included in our study were selected according to this basis.

There are numerous observations associating MVP with generalized musculoskeletal and connective tissue disorders but few recent data are available. The present study has been carried out to determine the association of MVP among Saudi Arabs with musculoskeletal and connective tissue disorders and the clinical implications of such an association.

Patients and Methods

Fifty-six consecutive outpatients were referred for evaluation of a systolic click or a click with a late systolic murmur. All patients had a complete history, physical examination, and were investigated by electrocardiogram and two-dimensional echocardiography. Chest x-ray and the posteroanterior and lateral projections were done and the internal dimensions of the chest were recorded in the lateral view, from the anterior surface of the body of the 8th thoracic vertebra along a perpendicular line to the posterior cortex of the sternum in the posteroanterior view, whereas the transthoracic diameter was measured at the level of the diaphragm. The ratio of the anteroposterior to the transthoracic dimensions was obtained. There were 14 males and 42 females, with an average age of 23 years (range 15-47).

Echocardiographic examination was performed by two-dimensional modality in the parasternal long axis, as well as four-chamber views utilizing Hewlett Packard ultrasound imaging system with 2.5 MHz transducer of 0.5 inch diameter. Mitral valve prolapse was diagnosed according to the criteria suggested by Alpert et al.⁸ and Popp et al.,⁹ with arching or systolic billowing of one or both mitral leaflets posteriorly and superiorly into the left atrium after crossing the plane of the mitral annulus, and doming of the anterior mitral leaflet characterized by a helmet-shaped deformity during diastole. Recently, Levine stated that technological progress advances our understanding and ability, especially if two-dimensional images could be combined to display three-dimensional relationships, which allow quantitative description of the mitral valve leaflets' position relative to the annulus.¹⁰ In our study, echocardiographic studies, as well as x-ray films, were reviewed for the accurate diagnosis of MVP and the

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presence of musculoskeletal disorders, respectively. The patient's evaluation included the assessment and identification of the varying spectrum of musculoskeletal defects such as pectus excavatum, kyphoscoliosis, loss of physiological kyphosis of the thorax, and scoliosis.

Thoracic cage dimensions in 50 normal subjects of similar age have been previously examined for MVP and were used for comparison.^{4,5}

Results

Fifty-six patients diagnosed with mitral valve prolapse were classified into four groups according to the predominant musculoskeletal deformity on clinical grounds. Group 1: shallow chest with pectus excavatum and narrow anteroposterior diameter, in three males and 11 females (14 cases, 25%). Group 2: straight back with narrowed anteroposterior diameter in five males and 11 females (16 cases, 28.6%). Group 3: kyphoscoliosis and scoliosis were detected in four males and six females (10 cases, 17.8%). Group 4: elliptical chest with a marfanoid type of patient was detected in two males and 14 females (16 cases, 28.5%). Associated clinical abnormalities included spontaneous pneumothorax in six cases; tricuspid and aortic valve prolapse in two cases, respectively; bicuspid aortic valve in four cases; and atrial septal defect (ostium secundum) in four cases.

Analysis of measurements showed that 20 participants of the control group showed wider anteroposterior chest diameter and shorter arm span than did the group of patients with MVP, while the rest (30 participants) had no skeletal deformity to be noted.

Discussion

Abnormalities of the bony thoracic cage were relatively common in the adult population, although DeLeon et al. stated that their presence does not imply associated cardiac disease. They also claim that pectus excavatum and straight back syndrome have been cited as forms of "pseudoheart disease."¹¹ The conclusion drawn from these early studies on thoracic musculoskeletal abnormalities—that they were causes of cardiac murmurs and that their detection prevented further investigations—should be rejected in view of more recent reports attesting to their association with mitral valve prolapse.¹² On the contrary, thoracic cage abnormalities have been a point of interest in cases with systolic click thought to be of extracardiac origin.¹³ There are no previous reports about musculoskeletal abnormalities being assessed; furthermore, radiological stigmata for diagnosing such abnormalities have never been evaluated among a Saudi population, while the criteria employed for Caucasians may not be applicable.

The patients of this study were taken consecutively as they were referred for evaluation and thus represent a random sample of patients with systolic click-late systolic murmur syndrome. Our results indicated the presence of certain patterns of thoracic deformities which are commonly reported to be associated with MVP syndrome. The presence of these skeletal deformities may be an identifiable indicator for arousing suspicion of the presence of MVP syndrome.^{14,15} Pyeritz and McKusick stated that MVP had been found on clinical and echocardiographic grounds in more than 50% of patients of all ages with Marfan's syndrome.¹⁶ A higher incidence of thoracic musculoskeletal deformities in patients with MVP has been reported in several earlier studies, where scoliosis and kyphoscoliosis were reported in 39% of the cases studied.¹⁴ This was in agreement with our results, as these deformities were the most common among our patients studied.

But pectus excavatum was the most common deformity among cases of other studies,¹⁷ while in our study, pectus excavatum was reported in only 14 cases, in agreement with the results of Mallikarjun et al.¹³

The association of thoracic musculoskeletal deformities and mitral valve prolapse needs to be further clarified. As early as 1963, David speculated that an embryologic explanation might make this issue clearer.¹⁸

Since the primordia of the mitral valve undergo differentiation to their final form at the same time between the 35th and 42nd day of fetal life, the vertebral column, as well as the thoracic cage, begin their chondrification and ossification then. Therefore, any influence on growth patterns at this stage may affect both the mitral valve and the bony thorax.^{14,18} Mallikarjun and coworkers linked thoracic cage abnormalities and MVP to genetic influence.¹³ Furthermore, in our opinion, these associations may represent an expression of a systemic connective tissue disorder to which we attribute the development of spontaneous pneumothorax in six of the study cases.

Clinical Implications

It is clear that many cases with MVP have easily recognizable chest wall and thoracic spine deformities, which should alert the treating physician to consider MVP, where careful auscultation will invariably reveal the correct diagnosis. The suggestion has been made to consider chest and thoracic spine deformities as nonauscultatory features of MVP. Gibney and Maurer concluded that skeletal deformities may be considered as clinical markers, indicating a more severe form of MVP.²⁰ Therefore, identifying such patients is extremely essential, since potential fetal arrhythmias, infective endocarditis, and cerebral embolism are dangerous complications of MVP.¹⁹

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