Mitral annular disjunction with atrial septal defect in children: An intriguing association

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ABSTRACT

Mitral annular disjunction (MAD) is defined as a separation between the mitral annulus and the left ventricular myocardium and is most often seen in association with mitral valve prolapse (MVP). MAD has been linked to ventricular arrhythmias in adults, independent of MVP. However, it has rarely been reported in children. We, hereby, report two cases of MAD associated with a large atrial septal defect (ASD). Thus far, there are no consensus guidelines for the management of MAD. The additional association of large ASD further complicates the decision-making in these patients. To the best of our knowledge, this is the first report of the association of MAD with ASD. We further discuss the challenges in the management of this condition.

Keywords: Congenital heart disease, mitral valve prolapse, pediatric cardiac intervention, pediatric cardiac surgery, sudden cardiac death

INTRODUCTION

Mitral annular disjunction (MAD) is a structural abnormality of the mitral valve that is commonly associated with mitral valve prolapse (MVP) and is defined by a separation between the mitral annulus and left ventricular (LV) myocardium.[1] MAD has been linked to ventricular arrhythmias and sudden cardiac death (SCD) in adults.[1,2] However, the prevalence, pathophysiology, and clinical implications of MAD in children are poorly understood. There are few reports of MAD with MVP in children,[3] but no reports with associated atrial septal defect (ASD) to the best of our knowledge. We report two cases of MAD with ASD and discuss the challenges in the management.

CASE REPORTS

Case 1

A 10-year-old male was referred for evaluation of a murmur detected during a routine school health check-up. On examination, the heart rate was 80/min with irregularity due to 3–4 ectopics/min. On auscultation, there was a midsystolic click followed by a grade 3/6 midsystolic murmur best heard at the apex. The rest of the examination was unremarkable. The electrocardiogram (ECG) showed sinus rhythm with a QTc interval of 470 ms and T-wave inversion in inferior leads. There were two ventricular ectopics of different morphologies [Figure 1]. Echocardiography showed an 18 mm secundum ASD with adequate rims, left-to-right shunt, dilated right-sided chambers, and no pulmonary hypertension [Video 1]. There was typical MVP with myxoid infiltration leading to leaflet thickness ≥5 mm (anterior mitral leaflet 7 mm, posterior mitral leaflet 14 mm) with bileaflet prolapse along with moderate mitral regurgitation (MR) [Video 2]. Furthermore, there was MAD with a systolic displacement of the lateral hinge point of the posterior mitral leaflet by 6 mm. The mitral annulus diameter was 30 mm and

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42 mm in diastole and systole, respectively. There was systolic curling of the LV base, with a base-to-mid-LV wall thickness ratio of 1.5. The Pickelhaube sign was positive [Figure 2], and LV function was normal (ejection fraction 65%). A 24-h Holter recording showed a ventricular ectopic burden of 8%, few runs of ventricular couplets, and bigeminy. The patient was advised to undergo cardiac magnetic resonance imaging (MRI) (± electrophysiology study) for further risk stratification. However, the patient was lost to follow-up.

Case 2

A 12-year-old asymptomatic female was referred to us with suspicion of congenital heart disease. The ECG showed sinus rhythm with no ventricular ectopics or ST-T changes. There was a 2% ventricular ectopic burden on a 24-h Holter study. Echocardiography revealed a fenestrated secundum ASD of 27 mm with a left-to-right shunt, dilated right-sided chambers, and no pulmonary hypertension. The mitral valve was myxomatous with leaflet thickness ≥5 mm (anterior mitral leaflet 5 mm, posterior mitral leaflet 9 mm) with bileaflet prolapse and associated moderate MR. Moreover, there was MAD with a systolic displacement of the lateral hinge point by 5 mm [Figure 3 and Video 3]. There was systolic curling of the LV base, with a base-to-mid-LV wall thickness ratio of 1.2. The Pickelhaube sign was positive [Figure 4], and LV function was normal (ejection fraction 68%). The patient was advised to undergo further investigations for risk stratification; however, this could not be done due to financial constraints.

DISCUSSION

MVP is a common valvular disorder that has traditionally been considered benign. However, a recent meta-analysis suggested a link between SCD and MVP, independent of the severity of MR or LV dysfunction.[4] This MVP phenotype has been labeled as arrhythmic MVP phenotype, and it is independently and strongly associated with MAD, marked leaflet redundancy, and ST-T abnormalities on ECG.[4,5] Lately, the focus has been on identifying this high-risk group among MVP patients.

The exact mechanism underlying the association of MAD with ventricular arrhythmias and SCD remains unclear. As per Padua's hypothesis, MAD and systolic curling motion lead to regional hypercontraction of inferobasal LV, consequently increasing the localized wall stress, leading to hypertrophy and replacement-type fibrosis over time.[5] In addition, inefficient ventricular contraction due to the annulus and LV de-anchoring increases LV dimensions during the end-systole. Whether such LV enlargement, disproportionate to MR, warrants modification of surgical indications is uncertain.[2]

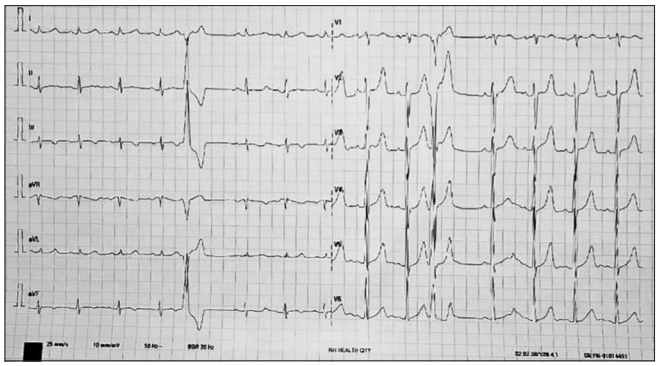

Figure 1: Twelve lead electrocardiogram showing sinus rhythm with T wave inversion in inferior leads and two ventricular ectopics of different morphologies

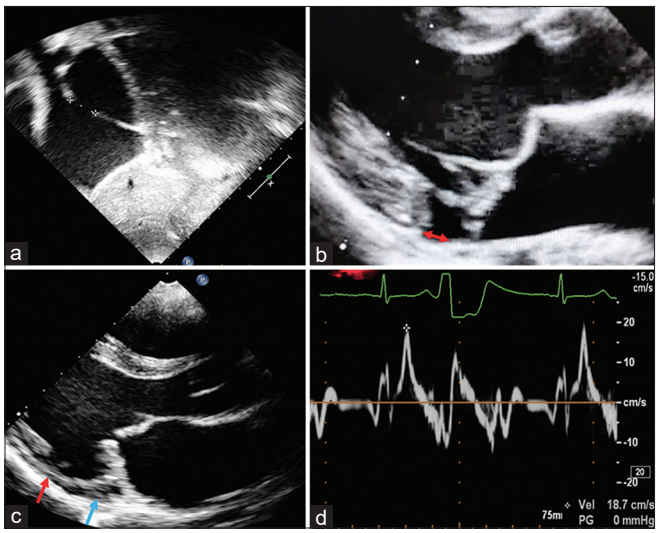

Figure 2: Transthoracic echocardiography of case 1. (a) Subcostal long-axis view showing the ostium secundum atrial septal defect measuring 18 mm. (b) The parastral long-axis view shows bileaflet mitral valve prolapse and the systolic displacement of the lateral hinge point of the posterior mitral leaflet (Red double arrow). (c) Parasternal long-axis view showing thickening of the posterior wall near annulus (blue arrow) and thinning of the mid posterior wall due to systolic curling of the left ventricular base (red arrow). (d) Tissue Doppler of the basal left ventricular myocardium showing a midsystolic peak of 18.9 cm/s (Pickelhaube sign)

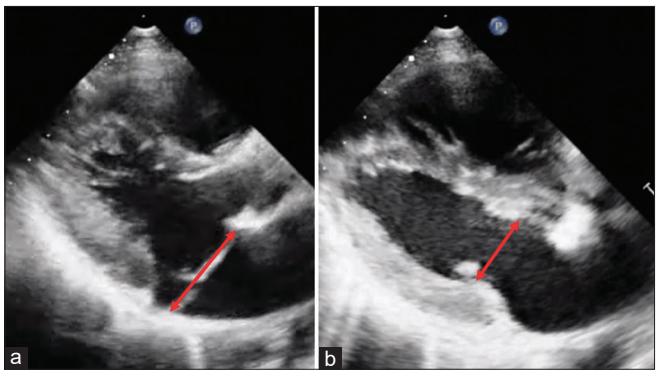

Figure 3: Parasternal long-axis images showing mitral annular diameter in (a). Systole (b). Diastole

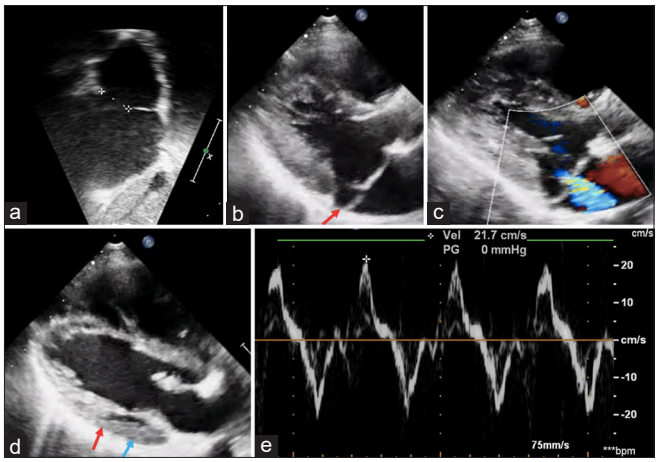

Figure 4: Transthoracic echocardiography of case 2. (a) Subcostal long-axis view showing the ostium secundum atrial septal defect measuring 19 mm. (b) The parastral long-axis view shows bileaflet mitral valve prolapse and the systolic displacement of the lateral hinge point of the posterior mitral leaflet (Red arrow). (c) The parsternal long-axis view with color Doppler shows moderate mitral regurgitation. (d) Parasternal long-axis view showing thickening of the posterior wall near annulus (blue arrow) and thinning of the mid posterior wall due to systolic curling of the left ventricular base (red arrow). (e) Tissue Doppler of the basal left ventricular myocardium showing a midsystolic peak of 21.7 cm/s (Pickelhaube sign)

The association of MVP has been seen in 10%–20% of patients with secundum ASD.[6] Most commonly, MVP in these patients is due to altered LV geometry resulting from right ventricular dilatation.[6] However, this MVP with associated MR usually spontaneously improves after ASD closure without the need to perform MV repair. On the other hand, patients with ASD and anatomical MVP (chordal elongation, leaflet redundancy) develop aggravation of MR after ASD closure.[7] Therefore, while evaluating patients with large ASD, careful evaluation of the mitral valve is crucial before planning intervention. Furthermore, with the increasing recognition of MAD and its clinical importance, it is essential to add it to the checklist when evaluating these patients. The prevalence of MAD in pediatric patients with MVP is likely to be lower than in adults with MVP (15%–86%) due to a lower incidence of myxomatous degeneration in children.[1,2]

The absence of consensus guidelines on the management of MAD makes decision-making in patients with MAD and large ASD challenging. The management options in MVP and MAD include medical therapy, catheter ablation, intracardiac defibrillator implantation, and mitral valve surgery.[2] The management needs to be individualized based on the risk stratification of these patients. Risk stratification is based on clinical presentation and initial tests, including ECG, echocardiography, diagnostic Holter, and further investigations such as cardiac MRI and electrophysiological testing.[2] Few studies have suggested that arrhythmic MVP could be treated surgically with a lower rate of postoperative arrhythmias, thus allowing antiarrhythmic drug discontinuation or reduction.[8]

In patients with MAD and ASD, a moderate-to-high risk SCD phenotype may necessitate a preference for surgical ASD closure and mitral valve repair over transcatheter ASD closure alone in case of device-suitable defects. Further systematic and large-volume prospective multicenter studies are needed to determine the threshold for preferring surgical repair over transcatheter repair in this unique and rare patient population. The documentation of such cases is important to draw attention toward the knowledge gaps in this area.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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