Unileaflet Mitral Valve in Patient With Marfanoid Habitus

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INTRODUCTION

Congenital malformations of the mitral valve (parachute mitral valve, double orifice of mitral valve, cleft mitral valve, atresia, and unileaflet mitral valve) are extremely rare. One of the rarest forms of these congenital anomalies is a unileaflet mitral valve, which is usually incompatible with life as it is associated with severe mitral regurgitation.[1] There have been very rare reported cases where patients have been asymptomatic and this valvular anomaly was an incidental finding on echocardiogram.[2] The presence of a unileaflet mitral valve is commonly due to a severely hypoplastic posterior mitral valve leaflet.[3] We present a case of a patient with marfanoid habitus and a unileaflet mitral valve found incidentally on transthoracic echocardiogram (TTE).

CASE PRESENTATION

A 26-year-old man with a medical history of right traumatic tension pneumothorax due to a gunshot wound and a childhood heart murmur presented to the hospital with severe acute pleuritic left-sided anterior midthoracic chest pain associated with dyspnea and exacerbated by movement that began 1 hour prior to arrival in the emergency department.

Social history included marijuana and tobacco use, and the patient was not taking any medications. Family history included a brother with asthma. They endorsed having testing as a child for Marfan syndrome, without confirmation of diagnosis. Reported occasional episodes of palpitations were more pronounced when they were anxious.

On physical examination, the patient's height was 193.04 cm; weight, 65.2 kg, body mass index, 17.5; and respiratory rate, 22. The remaining vital signs were stable. Arachnodactyly of extremities, long limbs, and pectus excavatum were observed. Systolic early peaking grade 1 out of 6 murmur was auscultated over the left sternal border. Decreased breath sounds were heard on auscultation of the left lung. The rest of the physical exam was within normal limits.

Investigation showed normal complete blood count, basic metabolic panel, serial troponins, thyroid-stimulating hormone, alpha-1-antitrypsin, and albumin. Both alkaline phosphatase and total protein were elevated at 140 U/L and 8.5 g/dL, respectively. Electrocardiogram showed right bundle branch block (Figure 1). Chest x-ray showed large left pneumothorax with no mediastinal shift and a bullet projecting over the right upper quadrant. Chest computed tomography without contrast showed moderate bullous disease in the right lung apex. During hospitalization, the patient had both symptomatic and asymptomatic self-resolving supraventricular tachycardic episodes lasting approximately 5 minutes each with heart rates in the 160s. This was found on the cardiac monitor, and cardiology was subsequently consulted. Transthoracic echocardiogram was consistent with a unileaflet mitral valve (Video 1). There was a prominent, redundant, and thickened anterior leaflet and absent posterior leaflet (Videos 2 and 3). The posteromedial papillary muscle was probably absent or underdeveloped (Video 4), and the anterolateral papillary muscle was normal. There were redundant, elongated chordae (Video 5) without systolic anterior motion and pseudodisjunction of the posterior mitral annulus. No mitral regurgitation was appreciated on color flow Doppler (Videos 6 and 7). The ejection fraction was estimated at 55% to 60%. In the parasternal long-axis two-dimensional (2D) view from TTE, we did not visualize a posterior mitral leaflet (Figures 2 and 3), which was highlighted with M mode (Figure 4). The patient recovered well, with minimal oxygen supplementation and chest tube placement. Given that their congenital mitral valve abnormality was found incidentally and they had no significant cardiac implications (as evidenced by a lack of mitral valve regurgitation, normal left ventricular function, and normal aortic root size), the patient was recommended to follow up with cardiology routinely and to consider further genetic testing for Marfan syndrome.

DISCUSSION

Unileaflet mitral valve, an incidental finding in our patient, can be caused by aplasia of the posterior mitral valve. This valvular anomaly is often found in the neonatal period and is associated with severe mitral regurgitation.[4] In this setting, the condition is incompatible with life without surgical intervention. Mitral valve repair or mitral valve replacement with either a mechanical or bioprosthetic valve are the current available surgical options.[5] Our patient had an unknown type of murmur found as a child and to our knowledge was not diagnosed with a congenital heart disease. The symptomatic tachyarrhythmia along with physical examination findings of marfanoid habitus and suspicion for cardiac conduction defect suggested by right bundle branch block, which can also accompany congenital heart disease, led to cardiac workup. Marfan syndrome is a dominantly inherited connective tissue disorder affecting the eyes, heart, and skeleton.[6] It is associated with cardiovascular abnormalities such as mitral valve prolapse and aortic aneurysm.[7] Our case presents the hypothesis of a possible association between a defect in fibrillin and a unileaflet mitral valve. However, the patient's modified Ghent criteria score was 3 and did not meet the criteria of 7 points to make the diagnosis of Marfan syndrome.[8] Our patient with marfanoid habitus had conduction

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Keywords: Unileaflet mitral valve, Marfanoid features, Marfan syndrome, Pneumothorax, Congenital anomaly

Conflicts of Interest: None.

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VIDEO HIGHLIGHTS

Video 1: Parasternal long-axis 2D view from TTE with chrome highlighting the thickened redundant anterior mitral valve leaflet that is enveloping and closing the entire mitral valve opening. The left atrium appears narrow likely due to a combination of unconventional alignment as well as a prominent vertebral body posteriorly adjacent to the descending aorta and posterior left atrial wall in a thin-bodied man with probable thoracic spine bone deformity.

Video 2: Parasternal short-axis view at the mitral valve level, 2D view from TTE, showing prominent anterior mitral leaflet and absent posterior leaflet.

Video 3: Parasternal short-axis view at the mitral valve level, 2D view from TTE second view with absent posterior mitral leaflet.

Video 4: Parasternal short-axis view at the level of the papillary muscle, 2D view from TTE with chrome showing an absent versus severely hypoplastic posteromedial papillary muscle.

Video 5: Apical 4-chamber 2D view from TTE showing a markedly elongated and redundant anterior mitral valve leaflet occupying the entire mitral annulus. There are aberrant chord structures noted in the distal aspect of the left ventricle near the apex.

Video 6: Parasternal long-axis 2D view from TTE with color flow Doppler showing no evidence of mitral regurgitation.

Video 7: Apical 4-chamber 2D view from TTE with color flow Doppler showing no evidence of mitral regurgitation.

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anomalies presenting as episodes of both symptomatic and asymptomatic supraventricular tachycardia. Spontaneous pneumothorax, bullae, and emphysema can manifest as initial symptoms of undiagnosed Marfan syndrome.[9] Our patient was found to have right apical blebs and spontaneous left-sided pneumothorax. The patient's social history included use of an unknown amount of marijuana, which has been associated with bullous lung disease and can lead to spontaneous pneumothorax.[10] There also appeared to be the presence of pseudodisjunction of the posterior mitral annulus on echocardiogram (Figure 5). Pseudodisjunction of the mitral annulus is defined as the movement of the basal portion of the posterior leaflet against the left atrial wall in systole, giving the illusion of disjunction even though the posterior leaflet is still attached on the left ventricular crest. True mitral annular disjunction is defined as the dislocation of the hinge point from the crest of the left ventricular wall, in the direction of the left atrium.[11] This finding may be relevant to our case given the abnormal mitral valve anatomy, especially if our suspicion for Marfan syndrome were to be confirmed. Mitral annular disjunction in these cases can serve as a prognostic indicator, as it has recently been recognized as a substrate for ventricular arrhythmia irrespective of mitral valve prolapse. Obtaining a transesophageal echocardiogram, three-dimensional echocardiography, or cardiac magnetic resonance imaging may have allowed us to better visualize this area to evaluate whether the posterior leaflet was hypoplastic or missing.[12,13] There are currently no guidelines regarding follow-up studies in patients diagnosed with unileaflet mitral valve, as it is quite rare, and there are no known surgical treatment options for asymptomatic patients.[14] Valvular defects, conduction defects, and spontaneous pneumothoraces place patients at increased lifetime risk of adverse cardiopulmonary events. Our case report evaluated the possible association between a marfanoid habitus and unileaflet mitral valve.

CONCLUSION

Our case report demonstrates a rare unileaflet mitral valve in an adult. This patient is unique since there was no significant mitral regurgitation and the patient had many features of a marfanoid body habitus.

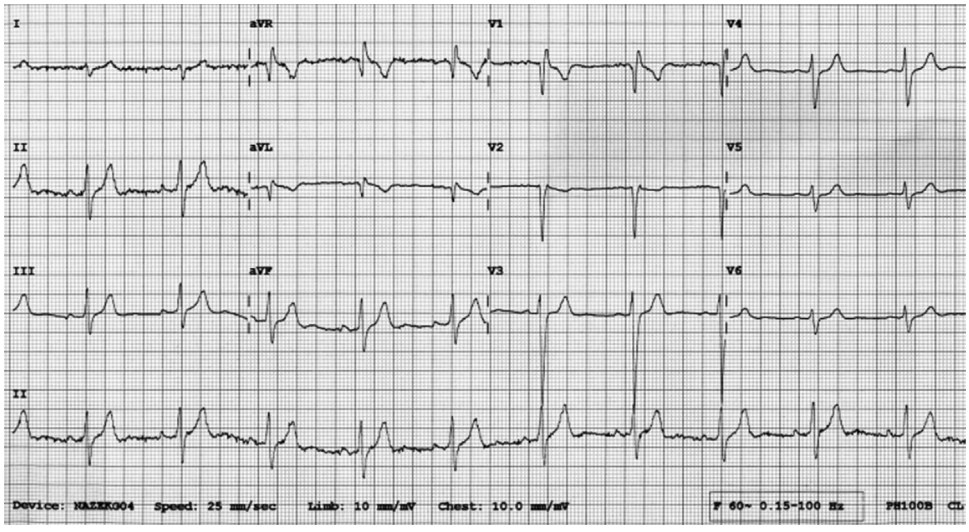

Figure 1 Electrocardiogram showing right bundle branch block.

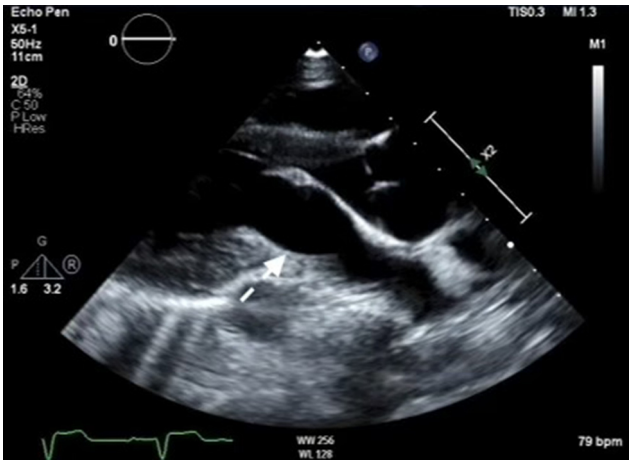

Figure 2 Parasternal long-axis 2D view from TTE showing mitral valve diastolic phase. The *arrow* points to the posterior mitral annulus with no visible posterior mitral leaflet.

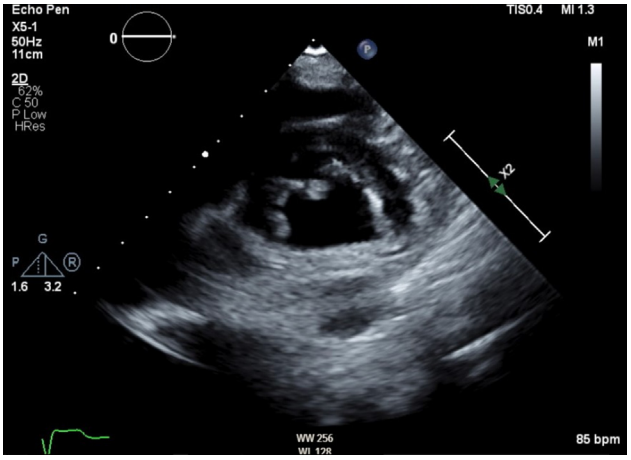

Figure 3 Parasternal short-axis 2D view from TTE showing mitral valve systolic phase with prominent anterior mitral leaflet and absent posterior leaflet.

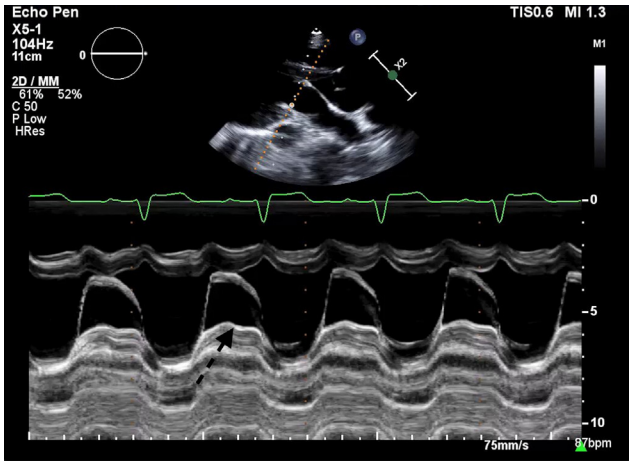

Figure 4 M mode depicting absence of posterior leaflet. The posterior mitral leaflet is typically seen in diastole below the anterior leaflet, which is visualized well in a high-quality M-mode imaging. The *arrow* points to the area of missing posterior leaflet. In addition, this M mode shows loss of E-F slope.

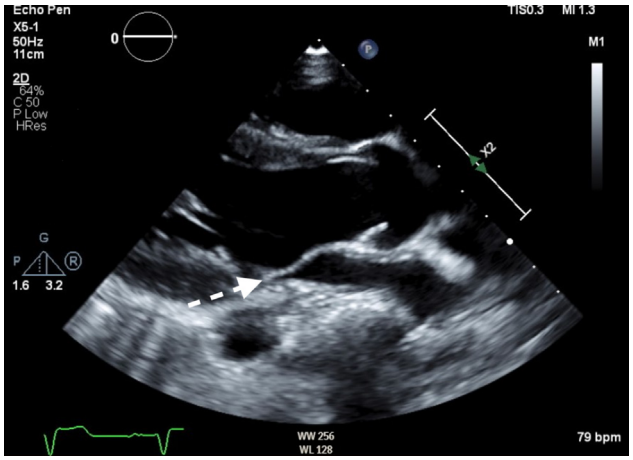

Figure 5 Parasternal long-axis 2D view from TTE showing mitral valve systolic phase. The *arrow* points to the closed anterior mitral leaflet that is redundant and covers the mitral annulus plane with no posterior leaflet seen at the coaptation line.

SUPPLEMENTARY DATA

Supplementary data to this article can be found online at https://doi.org/10.1016/j.case.2022.07.005.

REFERENCES

1. Bär H, Siegmund A, Wolf D, Hardt S, Katus HA, Mereles D. Prevalence of asymptomatic mitral valve malformations. Clin Res Cardiol 2009;98:305-9.
2. Kanagala P, Baker S, Green L, Houghton AR. Functionally uni-leaflet mitral valves in a family: a case series. Eur J Echocardiogr 2010;11:E27.
3. Roelandt R, Coeman M, Dehem J, Nollet A, De Keyser J. A rare case of a uni-leaflet mitral valve in a 65-year-old woman. Acta Cardiologica 2020;75:366-7.
4. Pourafkari L, Baghbani-Oskouei A, Toufan M, Ghaffari S, Nader ND. Hypoplastic posterior mitral valve leaflet: a case report and review of the literature. Echocardiography 2018;35:1052-5.
5. Arasaratnam K, Tomlinson S, Dahiya A, Lo A, Jalali H, Prasad SB. Surgical repair of a unileaflet mitral valve: a rare congenital abnormality and a novel surgical approach. CASE (Phila) 2020;4:420-8.
6. Child AH, Aragon-Martin JA, Sage K. Genetic testing in Marfan syndrome. Br J Hosp Med (Lond) 2016;77:38-41.
7. Espinola-Zavaleta N, Escalante-Armenta R, Rei-Cassab DL, Castellanos-Vizcaíno LJ, Alexanderson-Rosas E. Marfan syndrome with unileaflet mitral valve, moderate mitral regurgitation and atrial septal defect: an uncommon association. Clin Res Trials 2020;6.
8. Salik I, Rawla P. Marfan syndrome. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing, https://www.ncbi.nlm.nih.gov/books/NBK537339/ updated January 26, 2022.

9. Hao W, Fang Y, Lai H, Shen Y, Wang H, Lin M, et al. Marfan syndrome with pneumothorax: case report and review of literatures. J Thorac Dis 2017;9:E1100-3.
10. Mishra R, Patel R, Misbahuddin K. Cannabis-induced bullous lung disease leading to pneumothorax. Medicine 2017;96:e6917.
11. Punjabi PP, Rana BS. Mitral annular disjunction: is MAD "normal." Eur Heart J Cardiovasc Imaging 2021;22:623-5.
12. Espinola-Zavaleta N, Vargas-Barrón J, Keirns C, Rivera G, Romero-Cárdenas A, Roldán J, et al. Three-dimensional echocardiography in congenital malformations of the mitral valve. J Am Soc Echocardiogr 2002;15:468-72.
13. Bezgin T, Elveran A, Karagöz A, Canga Y, Yılmaz F. Mitral valve with a single leaflet. Turk Kardiyol Dern Ars 2014;42:80-2.
14. Shah J, Jain T, Shah S, Mawri S, Ananthasubramaniam K. Rare case of unileaflet mitral valve. J Cardiovasc Ultrasound 2016;24:168-9.