

Case Report

Hypoplasia or Absence of Posterior Leaflet: A Rare Congenital Anomaly of The Mitral Valve in Adulthood – Case Series

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Abstract

We present a case series of two adult patients with almost complete absence of the posterior mitral valve leaflet and who are asymptomatic or mildly symptomatic, with two different degrees of mitral regurgitation.

Keywords: Bicuspid aortic valve, congenital anomalies, mitral regurgitation, mitral valve, noncompaction cardiomyopathy

INTRODUCTION

Congenital abnormalities of the mitral valve are considered to be very rare cardiac anomalies. The absence of the posterior mitral valve leaflet (PMVL) is usually fatal in utero while deficient PMVLs typically present in childhood with symptomatic mitral regurgitation. A few cases have been described in asymptomatic adults.^[1] We present two cases in adult patients.

CASE REPORTS

Case A

A young man, 35-year-old, with tobacco abuse, hypertension, but no heart diseases in his clinical history, presented to our institution for palpitations. He had frequent ventricular premature beats on 24 h electrocardiography (ECG) Holter monitoring. At physical examination, we found an olosystolic murmur on the apex, not clinical signs of heart failure, blood pressure (BP) 110/70 and heart rate 80/min in sinus rhythm. Resting ECG and treadmill test were normal. The transthoracic echocardiogram (TTE) showed a clear picture of noncompaction cardiomyopathy. TTE demonstrated also an elongated and thickened anterior mitral valve leaflet with a moderate degree of prolapse. The posterior leaflet was extremely hypoplastic with just a short visible stump. The resultant coaptation line was eccentrically displaced with accompanying severe mitral regurgitation, directed posteriorly [Figures 1-3]. In other words, it was a functionally

unileaflet mitral valve. Transesophageal echocardiogram demonstrated also an anteroposterior bicuspid aortic valve [Figures 4 and 5] with accompanying severe aortic regurgitation. Real-time (RT) three-dimensional-transesophageal echocardiography (3D-TEE) showed very clearly the lack of posterior mitral leaflet [Figure 6]. The patient's clinical status impaired by 3 months with a severe heart failure development. The treatment was a mitral and aortic valve replacement with good long-term outcome.

Case B

A 21-year-old boy was referred to our cardiology unit by the family physician, who heard a cardiac murmur and suggested the need of a cardiac consult. The boy was asymptomatic, in good general condition and physically active. Physical examination revealed a mild systolic murmur at the apex. Lung fields were clear. BP was 120/70 mm Hg. ECG was entirely normal. TTE showed a marked hypoplasia of the posterior mitral leaflet [Figures 7 and 8], a wide anterior leaflet, with a very mild mitral regurgitation. Left ventricular and atrial volumes were normal as were ejective phase indices (left ventricular ejection fraction = 67%). Aortic, pulmonic and tricuspid valve did not show morphological abnormalities.

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Parato and Masia: Asymptomatic or mildly symptomatic adult patients with congenital mitral anomalies do exist

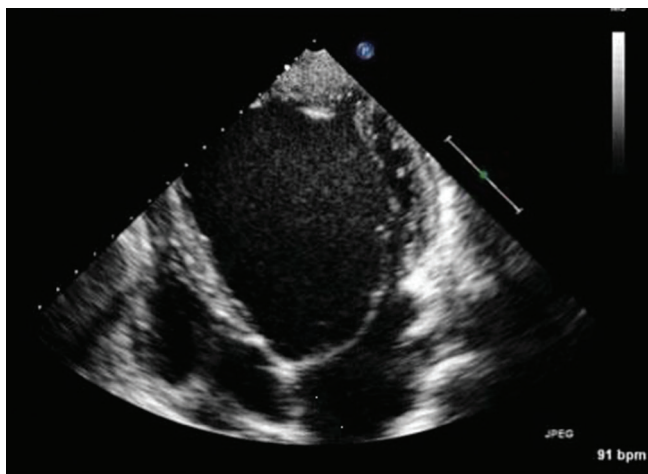


Figure 1: Case A. Transthoracic echocardiogram demonstrates an elongated and thickened anterior mitral valve leaflet. The posterior leaflet was extremely hypoplastic with just a short visible stump. A noncompaction picture of the left ventricle lateral wall is visible

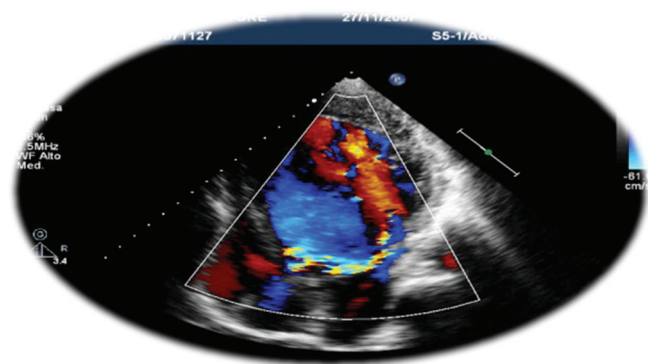


Figure 3: Case A. Transthoracic echocardiogram-4C color Doppler demonstrating an eccentric severe regurgitation jet

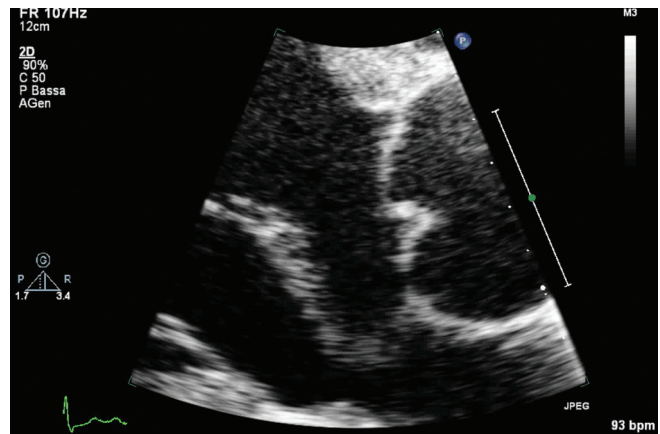


Figure 2: Case A. Transthoracic echocardiogram shows a marked hypoplasia of the posterior mitral leaflet and a normal sized anterior leaflet

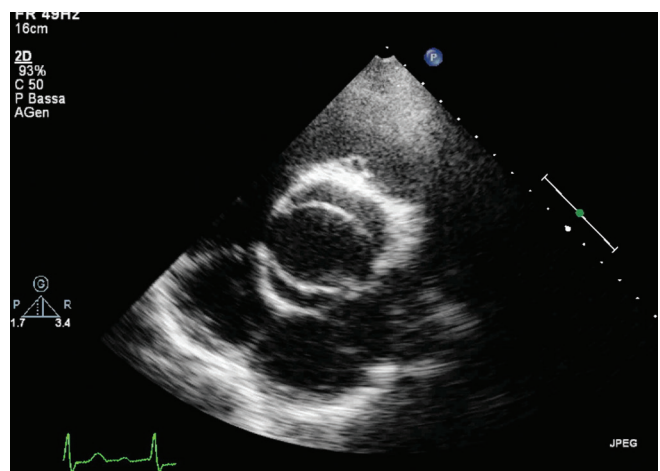


Figure 4: Case A. Two-dimensional transesophageal echocardiography demonstrating an anterior-posterior bicuspid aortic valve (opening phase)

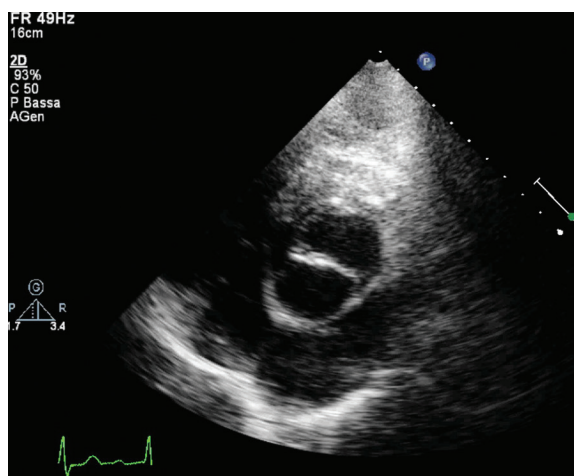


Figure 5: Case A. Two-dimensional transesophageal echocardiography demonstrating an anterior-posterior bicuspid aortic valve (closure phase)

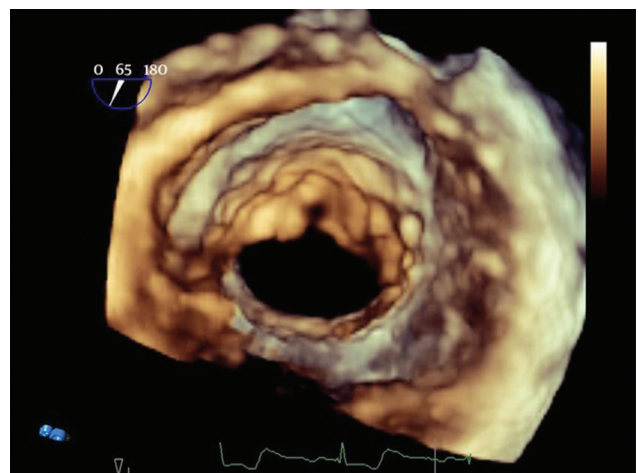


Figure 6: Case A. Three-dimensional real-time transesophageal echocardiography demonstrating the lack of posterior mitral leaflet

DISCUSSION

Congenital malformations of the posterior mitral leaflet are extremely rare and present with a wide spectrum. Hypoplasia

of the PMVL has been reported and a few cases of absent PMVL have been described.^[2] Hypoplasia of the PMVL is a rare congenital heart disease, usually presenting in infancy and

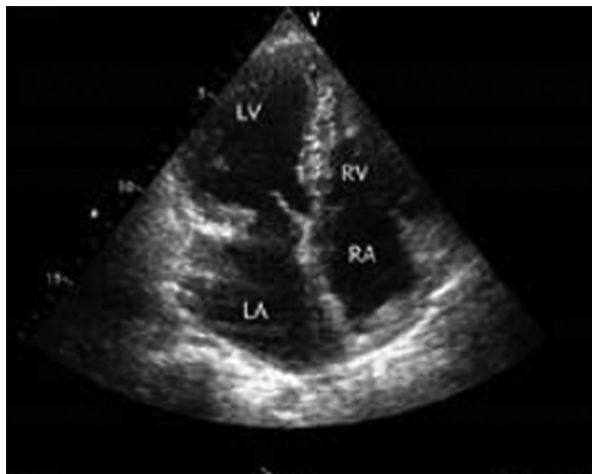


Figure 7: Case B. Transthoracic echocardiogram shows a marked hypoplasia of the posterior mitral leaflet and a normal sized anterior leaflet

childhood with severe mitral regurgitation, either in isolation or associated with other cardiac lesions.^[2] A prevalence of 1:8800 has previously been documented in preselected patient cohorts.^[3] The absence of PMVL is usually symptomatic, due to severe mitral regurgitation and coexisting abnormalities such as intracardiac shunt. Absent PMVL may be more prevalent in asymptomatic adults than is known. The reported cases demonstrate that asymptomatic patients with severe hypoplasia or complete absence of posterior mitral leaflet do exist. 3D-TTE may be the technique of choice to evaluate these cases.^[4] Saura *et al.*^[5] described a case of posterior leaflet hypoplasia associated with bicuspid aortic valve and evaluated by RT 3D-TTE. An association with noncompaction cardiomyopathy and bicuspid aortic valve, like case A, was not ever found.

Declaration of patient consent

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

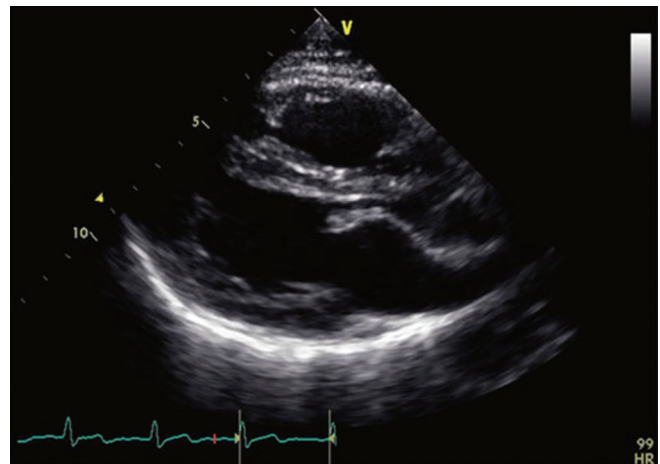


Figure 8: Case B. Transthoracic echocardiogram-Parasternal Long Axis (PLax) demonstrating a severe hypoplasia of mitral posterior leaflet

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Nil.

Conflicts of interest

There are no conflicts of interest.

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