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Asymptomatic Mitral Regurgitation Caused by an Isolated Mitral Leaflet Cleft in a Young Adult: A Case Report

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Abstract

We report the case of an 18-year-old young man without previous medical history, who was referred for evaluation of an asymptomatic cardiac murmur. Physical examination found holosystolic 4-5/6 apical murmur with normal ECG. Bidimensionnal (2D) echocardiography revealed severe mitral regurgitation with thin mitral leaflets. Three dimensional (3D) Echocardiography done for better assessment of mitral valve regurgitation mechanism revealed an isolated mitral leaflet cleft, without signs of endocarditis or traumatic lesion. Regarding the absence of symptoms and excellent maximal exercise tolerance at stress echocardiography, a repair surgery wasn't offered. Isolated mitral leaflet cleft is a rare congenital anomaly, in adults, the cleft may be an incidental finding that remains asymptomatic for years when the leak is well tolerated. 2D combined with 3D echocardiography is key for diagnosis and surgery guidance.

Keywords

Mitral Regurgitation, Isolated Mitral Cleft, Adult Congenital Heart Disease, Senegal

1. Introduction

The cleft mitral leaflet is a rare congenital anomaly [1], causing a mitral regurgitation. It is the most common cause of congenital mitral regurgitation (MR). Isolated cleft of the mitral valve, not associated with endocardial cushion defects, is uncommon. This anomaly is defined by a division of the mitral leaflet, due to a fusion

default of two components [1]. This first description was done in 1954 [2]. Evaluation of this congenital valvular anomaly is best performed by bi-dimensional echocardiography (2D) paired with three-dimensional (3D), viewing anatomic and morphological details, mitral regurgitation mechanism and quantification.

We here report a clinical case of a mitral regurgitation occurring secondary to an isolated mitral leaflet cleft, and the value of three-dimensional echocardiography for diagnosis and morphologic evaluation.

2. Case Presentation

A young 18-year-old male, with no medical or chirurgical history, was referred for evaluation of an asymptomatic cardiac murmur. Physical examination found a holosystolic murmur, prominent at the apex, radiating to the axilla, with 4-5/6 intensity. ECG 12-leads at rest were normal.

Bi-dimensional transthoracic echocardiography revealed a severe mitral regurgitation jet, (surface regurgitant orifice was 38 mm^2, regurgitant volume 67.8 ml and vena contracta 7 mm), eccentric directed towards the left atrium wall (**Figure 1**) and left atrium dilatation. Left ventricular size (end-diastolic diameter was 54 mm) and function were normal (**Figure 2**). Size of the mitral annulus

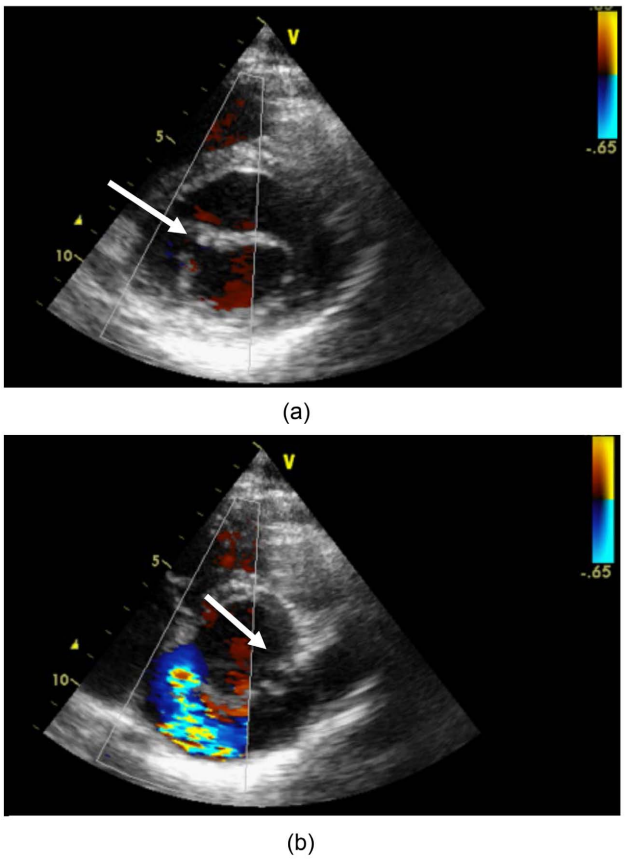

Figure 1. 2D (a) and color Doppler (b) echocardiography A: Short axis view at mitral valve level. Showing the isolated mitral cleft (white arrow).

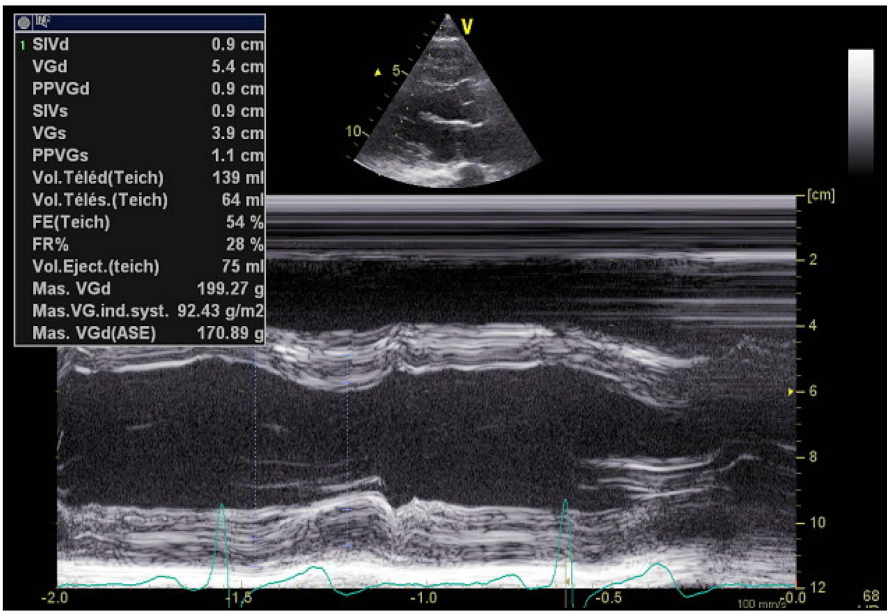

Figure 2. TM of left ventricle in transthoracic parasternal echocardiography view of the patient.

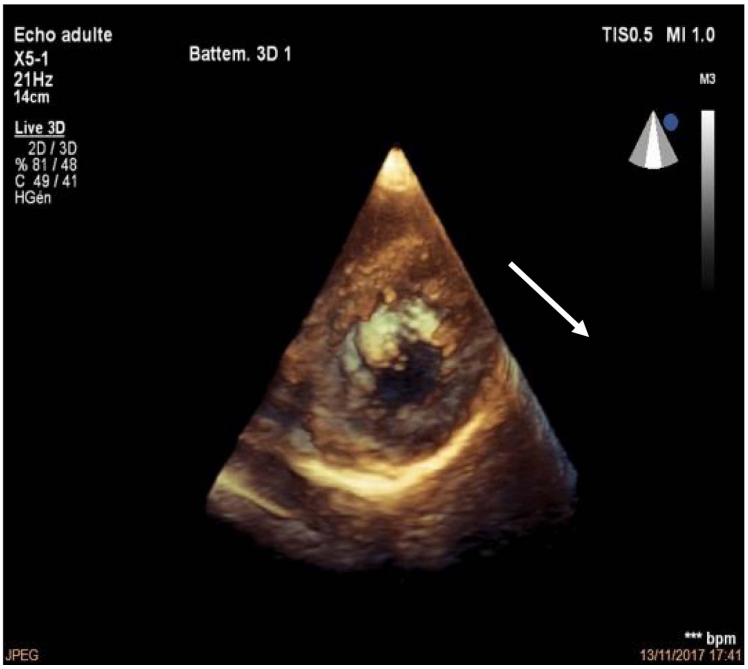

Figure 3. 3D echocardiography, short axis view at mitral level, showing the isolated mitral cleft (white arrow).

was also normal. Mitral leaflets were thin, there was no commissural fusion. Two D-echocardiographies did not show any other cardiac abnormality. For better assessment of mitral valve anatomy, a 3D echocardiography was performed (**Figure 3**). The examination revealed the isolated mitral leaflet cleft, and regurgitation at this place. There were no evidence of infective endocarditis, trauma or previous surgery, diagnosis of an isolated mitral leaflet cleft was retained.

Regarding the absence of symptoms and excellent maximal exercise tolerance at the stress echocardiography, a repair surgery wasn't offered to this patient.

3. Discussion

Isolated mitral leaflet cleft is caused by an incomplete fusion of two portions of the mitral leaflet's. This uncommon anomaly in adults causes a congenital mitral regurgitation [3] [4].

A mild mitral regurgitation, as a result of a mitral cleft, can remain asymptomatic for a long time making frequent its fortuitous discovery.

Echocardiography is the first-choice imaging technique to assess mitral valve congenital anomalies, giving details about mitral valve anatomy and morphology, mitral regurgitation mechanism and quantification. Sometimes, mitral cleft evaluation by 2D echocardiography examination may be difficult and 3D echocardiography may then be helpful to evaluate and get a sophisticated 3-dimensional structural analysis [5]. Three-Dimensional echocardiography gives a geometrical imaging of mitral leaflets and annulus, subvalvular apparatus and relationships with local structures. This technique shows a real-time display anatomy of the mitral valve.

Combining 2D and 3D echocardiography is useful for cleft diagnosis and repair surgery guidance. Repair surgery guided by both 2 and 3-dimensional echocardiography shows a success rate of 93% [6] [7].

In our case, echocardiography was the key for cleft diagnosis.

Some other imaging techniques may be useful to diagnose congenital valvular mitral anomalies, like cardiac magnetic resonance (CMR). However, CMR accessibility remains in our context a challenge due to its high cost.

Prognosis of surgically repaired mitral cleft is usually excellent, offering a significant survival improvement [8] [9].

In this case, mitral regurgitation was asymptomatic and the patient showed an excellent maximal exercise tolerance. Therefore, a repair surgery wasn't indicated. A clinical and echocardiographic follow-up was proposed to our patient.

4. Conclusion

Isolated mitral leaflet cleft is a rare congenital anomaly. In adults, cleft may be a fortuitous discovery that remains asymptomatic for years when responsible for a mild mitral regurgitation. 2D combined with 3D echocardiography is key for diagnosis and surgery guidance.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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