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Mitral valve blood cyst with atrial septal aneurysm treated as rheumatic valvular heart disease: a case report and review of literature

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Abstract

Background Mitral blood cyst with atrial septal aneurysm is a unique instance of rare occurrence in medical literature. It is crucial for the discipline of cardiology to address the special difficulties that come with diagnosing and treating these two diseases together. The goal of this case report is to provide a comprehensive overview of the clinical presentation, diagnostic challenges, and treatment strategies for this uncommon cardiac disease in a resource-limited setting.

Case presentation This is a case involving a 50-year-old Ethiopian female patient referred to our hospital for cardiac evaluation. She presented with intermittent palpitation to a nearby health center, which was diagnosed as rheumatic heart disease, for which she was on intramuscular benzathine penicillin injection on a monthly basis. Otherwise, she had no history suggestive of acute rheumatic fever and no chest pain, body swelling, orthopnea, or paroxysmal nocturnal dyspnea, historically or on current presentation. Evaluations revealed that her chest was clear and resonant with good air entry, and flat jugular venous pressure, but she had a grade III pansystolic murmur best heard at the apex that radiates to the left axilla. No other pertinent physical examination findings were discovered. Echocardiographic evaluation revealed a tiny cyst at the apex of posterior mitral valve leaflet with interatrial septal bulging into the right atrium.

Conclusion Cardiac blood cysts are an uncommon congenital cardiac condition that can lead to stroke and other potentially fatal consequences, especially if they are situated on the mitral valve. With improved specialists and low-risk procedures, surgical removal is advised. Given the patient's state, a proper diagnosis was therefore essential to provide her with accessible management options in this setting while she waits for a definitive surgical treatment in case the need arises.

Key points

- Blood cyst is a rare congenital abnormality that can be found attached to heart valves. Individuals may present with heart failure symptoms and potentially with stroke. Treatment alternatives might be offered when a diagnosis is made early.

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- The rare occurrence of concomitant mitral valve blood cyst with atrial septal aneurysm posed a unique challenge that led to the treatment of the patient for rheumatic heart disease. This makes for an important topic for discussion in the field of cardiology.
- In a nation with limited resources such as ours, where rheumatic heart disease is the most prevalent problem, this poses a management conundrum. The management team opted to halt the patient's previous treatment regimen with planned follow-up after speaking with the patient, as the patient only exhibited intermittent palpitations, no other symptoms that would indicate heart failure, and no obvious therapeutic choices in the setting.

Keywords Mitral valve, Rheumatic heart disease, Blood cyst, Atrial septal aneurysm, Case report, Jimma Medical Center, Ethiopia

Introduction

Cardiac valve blood cysts are exceedingly rare benign cardiac lesions in adults that mostly originate from atrioventricular valves and, rarely, can accompany coronary artery disease [1]. Atrial septal aneurysm is another rare congenital disorder that can potentially be a source of neurological ischemic events [2]. Most individuals exhibit no symptoms, but, when present, manifestations depend on the tumor's size and location, including breathlessness, fainting, or embolic events that necessitate treatment. Echocardiography remains the primary diagnostic modality for cardiac blood cysts, akin to other heart tumors. Notably, cysts on the mitral valve carry a high risk of severe complications. Surgical excision is often advisable owing to its safety and effectiveness [3, 4].

Given its rarity, silent progression, and the widespread burden of rheumatic valvular disease in low-resource settings, especially in Africa and Ethiopia, clinicians often have a low suspicion index, resulting in frequent misdiagnosis or overlooked cases. As a result, no documented cases exist in Africa. This report emphasizes the importance of clinical vigilance and advanced imaging techniques to enhance detection, ensuring timely surgical intervention to prevent serious complications. In our case, the coexistence of the two disorders makes the case exceedingly rare, which poses a challenge in diagnosis and management. Here, we present a case of mitral valve blood cyst with atrial septal aneurysm in a 50-year-old female patient who came with a complaint of intermittent palpation being treated as rheumatic heart disease, primarily due to lack of expertise and, hence, proper evaluation with echocardiography. The patient was on treatment and follow-up, as the physical examination and primary screening echocardiogram performed by the radiologist suggested rheumatic heart disease, the most common cause of heart diseases in this setting.

Case presentation

This is the case of a 50-year-old Ethiopian female patient referred to our hospital for cardiac evaluation. She was a para 8 (all alive) who had been in her usual state of health until 2 years prior when she began experiencing intermittent palpitations. For this complaint, she visited a nearby health facility where she was prescribed monthly intramuscular benzathine penicillin injections for rheumatic heart disease, which she had been taking since then. However, she did not remember a history suggestive of acute rheumatic fever. She had no history of body swelling, orthopnea, or paroxysmal nocturnal dyspnea. She had no personal or family history of diabetes mellitus, hypertension, or cardiac disease.

Upon evaluation, she appeared well, with a blood pressure of 100/75 mmHg, a pulse rate of 76 beats per minute, and a respiratory rate of 18 breaths per minute. A thorough physical examination revealed clear, resonant lung fields with good air entry and a flat jugular venous pressure (JVP). However, she had a grade III pansystolic murmur best heard at the apex, radiating to the left axilla. No other significant physical examination findings were noted.

Echocardiographic examination revealed an interatrial septal bulge extending more than 10 mm into the right atrial cavity, consistent with a type 1R atrial septal aneurysm (ASA-1R; Fig. 1 and Video 1). In addition, a small cyst measuring 1.91 cm $\times$1.35 cm $\times$1.62 cm was identified at the apex of the posterior mitral valve leaflet (Fig. 2 and Video 1). Although a bubble study was not performed, no interatrial septal defect was observed. There was mild thickening of the mitral leaflet, while the left atrial anteroposterior (AP) diameter measured 3.9 cm in the M-mode parasternal long-axis view. The left ventricular internal dimensions were 5.2 cm in diastole and 3.1 cm in systole. The ejection fraction was normal (left ventricular ejection fraction

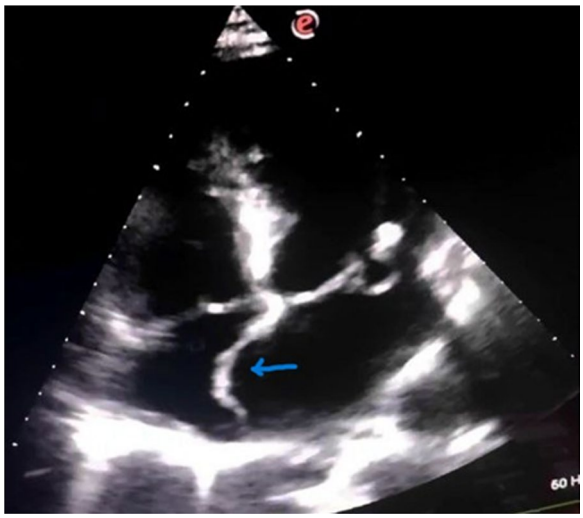


Fig. 1 Echocardiography of a 50-year-old female showing atrial septal aneurysm; *blue arrow* indicates an interatrial septal bulge extending over 10 mm into the right atrial cavity (apical four-chamber view)

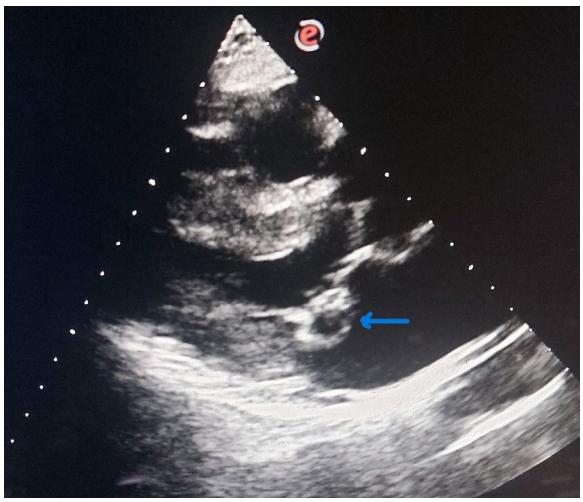


Fig. 2 Echocardiography of a 50-year-old female showing blood cyst at posterior mitral leaflet apex and associated atrial septal aneurysm; *blue arrow* indicates a cyst located at the tip of the posterior mitral leaflet

(LVEF): 63% with fractional shortening of 34%, measured in two-dimensional (2D) mode).

Additional qualitative Doppler imaging demonstrated transmitral regurgitation with a maximum velocity (V_{max}) of 4 m/second (directed anteriorly), a vena contracta width of 0.32 cm, and an estimated color flow jet area of 20–40%. The proximal isovelocity surface area (PISA) radius was less than 9 mm, and pulmonary vein systolic flow reversal was absent. These findings confirmed

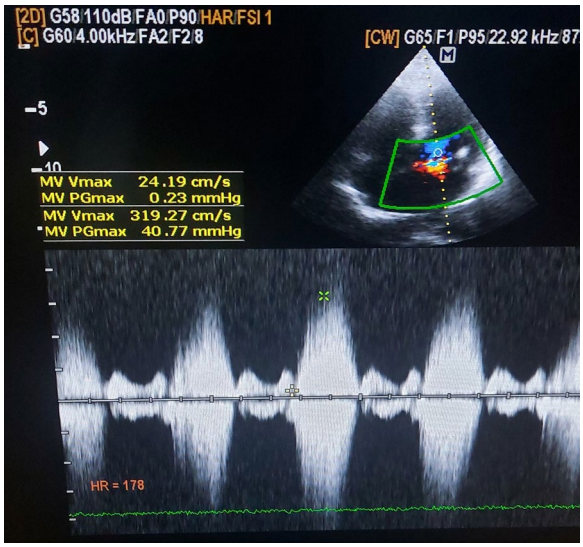


Fig. 3 Echocardiography of a 50-year-old female showing transmitral regurgitation with a maximum velocity of 4 m/s, vena contracta width of 0.32 cm and estimated color flow jet area between 20% and 40%, suggestive of moderate eccentric mitral regurgitation (directed anteriorly; Doppler study)

moderate mitral regurgitation (MR) (Fig. 3 and Video 1). Further evaluation with transesophageal echocardiography was not performed owing to the unavailability of this service at our facility.

Additional laboratory tests, including C-reactive protein (CRP), random blood sugar, liver and kidney function tests, and complete blood counts, were all within normal ranges. Anti-Streptolysin O (ASO) titer and serologic markers for syphilis and retroviral infections were negative. A posteroanterior (PA) chest X-ray showed no signs of lung congestion and a normal cardiothoracic ratio. Electrolyte levels and electrocardiogram (ECG) findings were also unremarkable.

On the basis of these findings, the initial diagnosis of rheumatic heart disease was reconsidered and ultimately abandoned. The final diagnosis was atrial septal aneurysm and a blood cyst associated with the apical portion of the posterior mitral valve leaflet. Since there was no surgical indication based on echocardiographic findings and no facility available for potential surgery, the patient was counseled, placed on follow-up, and found to be doing well during subsequent visits. She was advised to seek surgical referral if the need arose in the future.

Discussion

Blood cysts on heart valves were originally reported by Elsasser in 1844 and then by Luschka in 1857 [3]. While blood cysts of the heart valves are most commonly found in infants, they are uncommon in adults. The majority of

these cysts are asymptomatic, but they can occasionally result in systemic and cardiac problems, such as being a nidus for embolization and valve dysfunction, mitral regurgitation due to incomplete coaptation of the valve leaflets, and left ventricular outflow tract obstruction, especially when the cyst is on anterior mitral valve leaflet and large enough, prolapsing into the left ventricular outflow tract [4]. In addition to being a congenital condition, acute trauma, inflammatory processes, and complications from cardiothoracic surgery can all lead to cardiac blood cysts [5].

In our review of English-language case reports (2004–2024) using the keyword "mitral valve blood cyst," we identified 56 documented patients with mitral valve blood cysts [6–8]. The mean age at presentation was 48 ±18 years. While the majority (85.7%, $n=$ 48/56) were asymptomatic, 12.5% ($n=$ 7/56) exhibited clinical manifestations, including cerebrovascular symptoms (e.g., transient ischemic attack-like weakness), syncope, or chest pain mimicking acute coronary syndrome. Transthoracic echocardiography (TTE) and transesophageal echocardiography (TEE) were the primary diagnostic modalities.

Cyst localization predominantly involved the anterior mitral leaflet (68%, $n=$ 38/56), with rare involvement of the anterior papillary muscle (14%, $n=$ 8/56), posterior mitral leaflet (11%, $n=$ 6/56), and posterior papillary muscle (7%, $n=$ 4/56). Doppler imaging revealed moderate-to-severe mitral regurgitation in 82% ($n=$ 46/56) of cases. Cyst dimensions ranged from 10 to 40 mm (median: 22 mm). Surgical excision was performed in 78.6% ($n=$ 44/56), while the remainder ($n=$ 12/56) underwent conservative monitoring owing to comorbidities or patient preference. Our case represents the first reported instance from Africa and is unique for its concomitant mitral valve blood cyst and atrial septal aneurysm—a combination not previously documented in literature. This association underscores the need for comprehensive imaging to exclude coexisting cardiac anomalies in such rare presentations.

Atrial septal aneurysm is also another rare yet well-known cardiac abnormality, which can occur as a result of interatrial pressure variations. It can also be the result of primary abnormality affecting the fossa ovalis or the whole interatrial septum. As a result, it is mostly associated with interatrial shunts, in particular patent foramen ovale [9]. Though it has no clearly known clinical significance, it can sometimes be related to systolic clicks and atrial arrhythmias [10].

Unlike the common association of atrial septal aneurysm with septal defects as described above in our case, it is associated with mitral valve blood cyst, which is exceedingly very rare. This creates a dilemma in management, especially in a resource-limited country such as ours, where rheumatic heart diseases are the commonest problem of society, and there is lack of facilities and limitation of skilled manpower to provide these patients with optimal care in most of the rural areas of the country. As the patient has only intermittent palpitations, with no other symptoms suggesting heart failure, and also considering the lack of facilities providing optimal care for this particular case and the limited manpower in our setting, the patient was placed on follow-up.

Conclusion

Cardiac blood cysts are an uncommon congenital cardiac condition that can lead to stroke and other potentially fatal consequences, especially if they are situated on the mitral valve. With improved specialists and low-risk procedures, surgical removal is advised. Given the patient's state, a proper diagnosis was therefore essential to provide her with accessible management options in this setting while she waits for a definitive surgical treatment in case the need arises.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s13256-025-05292-1.

Supplementary Material 1: Video 1: Echocardiography of a 50-year-old female showing blood cyst at posterior mitral leaflet apex and associated atrial septal aneurysm

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Author contributions
ETA, KNT, KTB, and TGW made contributions to the first draft's conception, design, investigation, analysis, and writing. ETA and KNT took part in the supervision, analysis, and edition of the data, as well as data curation. The manuscript was modified and reviewed by all authors before being approved in its final form.

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Availability of data and materials
All the necessary data are available within the document.

Declarations

Ethical approval and consent to participate
Ethical approval was not needed; however, the confidentiality of the participant's information was kept secret and was used only for this publication without the patient's identification.

Consent for publication
Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests
The authors declare that they do not have any competing interests regarding this manuscript.

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