Case Report

ANNALS OF MEDICINE & SURGERY

OPEN

Lutembacher syndrome with congenital atrial septal defect in an 18-year-old female: a rare case report

Sara Mohamed Astanbouli, MD[a], Abdulrahman Ahmad Othman, MD[a], Saleh Takkem, MD[b], Mohammad Yassin Al Aboud, MD[c], Hamza Yousef, MD[c], Haidar Yousef, MD[d], Judy Alabd AlRhman, MD[e], Mahmoud Mustafa Abbas, MD[f], Mahmoud Mohamad Khair Alhasan, MD[f], Khaled Taysir Jaamour, MD[c], Ahmad Ibraheem Buz, MD[g], Bilal Sleiay, MD[g,*]

Introduction: Lutembacher syndrome (LS) is a rare condition with congenital atrial septal defect (ASD) and mitral stenosis (MS), often post-rheumatic illness. Diagnosis uses Doppler echocardiography, and treatment may involve surgery or percutaneous options.

Case presentation: An 18-year-old female presented with worsening dyspnea, orthopnea, and potential undiagnosed rheumatic fever. Chest X-ray showed cardiomegaly. An echocardiogram revealed left atrial dilation and mitral stenosis with regurgitation, aortic valve showed thickening without stenosis, and the right ventricle was mildly dilated; an interatrial shunt was present. Mitral stenosis worsened left-to-right shunt.

Discussion: LS results from the balance of ASD and MS. Factors influencing prognosis include pulmonary resistance, ASD size, and mitral stenosis severity. Echocardiography is essential for diagnosis, which revealed left atrial dilation, normal left ventricular function, severe mitral stenosis, and pulmonary valve changes. ECG indicated right ventricular hypertrophy. CXR showed left atrial enlargement and right ventricle enlargement. Treatment included an open heart surgery which replaced the mitral valve and closed the ASD.

Conclusion: Lutembacher syndrome, a rare condition combining ASD and mitral stenosis, can cause cardiac failure and pulmonary hypertension if untreated. Given the patient's stable condition in the early stages of the disease, early surgical or percutaneous intervention is advisable.

Keywords: case report, Lutembacher syndrome, mitral stenosis, rheumatic illness

Introduction

Lutembacher syndrome (LS) is an uncommon condition characterized by a congenital atrial septal defect (ASD), usually of the ostium secundum kind, and mitral stenosis (MS), frequently resulting from rheumatic illness[1].

Epidemiological data on LS are limited, but some studies indicate a higher prevalence among women and a negative long-term development[2]. Recent studies indicate that a history of rheumatic fever is present in up to 40% of cases from developing countries. In general, patients with LS can go years without experiencing any symptoms. Clinical aspects are frequently caused by ASD, and variations in symptoms and signs depend on the magnitude of the ASD. When diagnosing LS, Doppler echocardiography is considered the gold standard method. It is accurate in diagnosing both ASD and MS, non-invasive, and available in various clinical settings[3].

Palpitations, ventricular overload, heart failure, and pulmonary congestion are all prevalent symptoms that might appear unexpectedly and infrequently. Certain symptoms occur later in life[4].

Even though surgery is still the preferred treatment in LS, percutaneous treatment has emerged as a viable and appealing alternative therapy in selected patients. The benefits of this style of treatment include its effectiveness and safety in selected patients, as well as its ability to prevent severe cardiac surgery and anesthesia issues[5].

We report a case of an 18-year-old female diagnosed with LS, its presentation, treatment modalities, and its follow-up.

Case presentation

An 18-year-old female patient presented to the clinic complained of exertional dyspnea (NYHA Class II) that was steadily worsening, as well as orthopnea. There was no history of associated chest pain, cough, palpitations, fever, or indications of upper respiratory infections. The patient had a history of pharyngitis

[a]Medizinische Fakultät der Universität des Saarlandes, Homburg, Germany, [b]Department of Cardiology, Alwatani Hospital, Hama, Syria, [c]faculty of medicine, Tishreen University, Latakia, Syria, [d]Faculty of Medicine, Al-Sham Private University, Damascus, Syria, [e]Faculty of Medicine, Damascus University, Damascus, Syria, [f]Faculty of Medicine, University of Aleppo, Aleppo, Syria and [g]Faculty of Medicine, University of Hama, Hama, Syria

Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article.

*Corresponding author. Address: Faculty of Medicine, University of Hama, Hama, Syria. E-mail: (bilalsleiaysleiay@gmail.com) (B. Sleiay).

Annals of Medicine & Surgery (2025) 87:1029–1033

Received 8 October 2024; Accepted 5 January 2025

Published online 21 January 2025

http://dx.doi.org/10.1097/MS9.0000000000002944

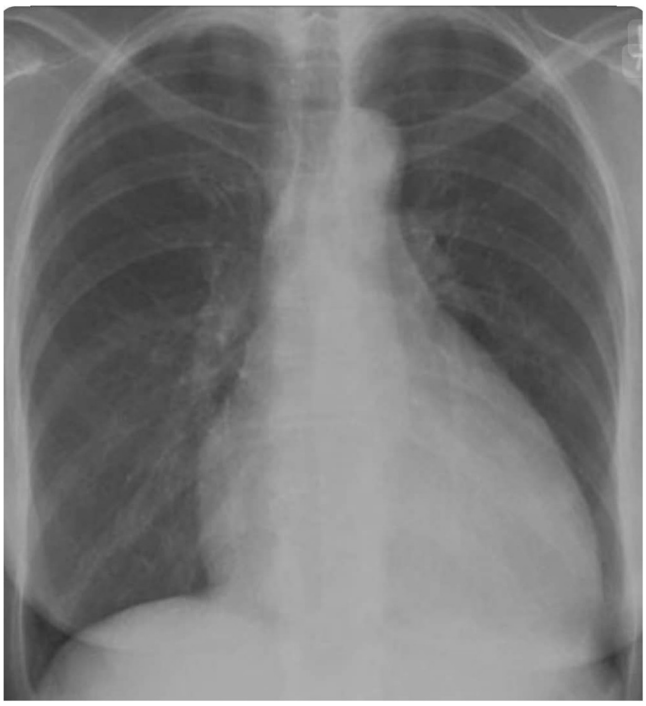

Figure 1. X-ray showed enlargement of the right ventricle.

and knee joint inflammation three years prior, and she may have an undiagnosed clinical history of rheumatic fever. Upon examination, a grade 3/6 apical diastolic rumble and a pulmonary systolic murmur were noted.

Her investigation results were as follows: blood pressure: 110/70 mmHg, pulse: 80 beats per minute, oxygen saturation: 97%, creatinine level (Cr): 0.7 mg/dL, urea: 15 mg/dL, hemoglobin (Hb): 12 g/dL, platelets: 250,000/µL, white blood cells (WBCs): 7,000/µL, sodium levels: 140 mEq/L, and potassium levels: 4.1 mEq/L.

Her chest X-ray (CXR) revealed features of cardiomegaly, such as straightening of the left cardiac border and enlargement of the right ventricle (Fig. 1). The electrocardiogram (ECG) showed a right-axis deviation and mild right ventricular hypertrophy, indicative of right heart pressure overload (RPH), as well as a partial right bundle branch block (RBBB) (Fig. 2).

A cardiac echocardiogram was performed . The left ventricle showed normal function with end-systolic and end-diastolic

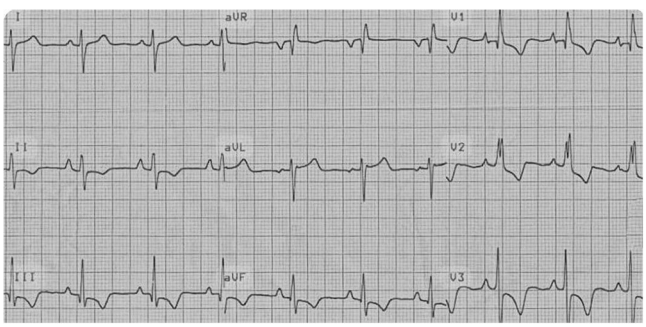

Figure 2. ECG admonstrated right bundle branch block.

diameters ranging from 35 to 47 mm, with an ejection fraction (EF) of 60%. There was thickening and fibrosis of the leaflets, with signs of rheumatic involvement, and a "hockey stick" appearance of the anterior leaflet, along with mild mitral regurgitation (1-2/4) and severe mitral stenosis. The mitral area was approximately 1.4 cm^2, with a mean gradient across the mitral valve of about 9 mmHg, and the pressure half-time (PHT) was measured at 153 ms (Fig. 3).

The aortic valve leaflets demonstrated thickening and calcification of the edges without significant stenosis or regurgitation. The right ventricle showed good function with mild dilation, and the right atrium was mildly to moderately dilated. There was increased flow across the pulmonary valve and mild regurgitation in the tricuspid valve (1-2/4) with a velocity of 3.3 m/s, while the pulmonary artery systolic pressure ranged from 40 to 45 mmHg.

The interatrial septum showed a secondary opening of approximately 6 to 7 mm in size (medium). This indicated a left-to-right shunt, decreasing left atrial pressure and underestimating the mean gradient across the mitral valve. In cases of severe mitral stenosis alone, the mean gradient is usually over 10 mmHg; however, if an interatrial shunt is present, blood flow direction can shift from the left atrium to the right, resulting in a decrease in the mean gradient across the mitral valve. This may lead one to mistakenly believe that the stenosis is moderate when it is severe, as indicated by a gradient of 8 mmHg in this case (Fig. 4).

The mitral stenosis exacerbates the left-to-right shunt, increasing the pulmonary blood flow (Qp) relative to systemic blood flow (Qs) and enhancing the shunt. Thus, a small opening between the atria may functionally act as a larger opening, forcefully increasing flow and raising pulmonary pressure more rapidly than expected.

Surgical intervention was recommended, resulting in the replacement of the mitral valve, which was deemed irreparable, along with surgical closure of the interatrial septal opening using a patch. Upon follow-up after one and a half years, the patient was in good general condition, on warfarin therapy, and doing well.

Discussion

LS is determined by the balance between ASD and MS. Numerous factors influence the prognosis, including pulmonary vascular resistance, RV compliance, ASD size, and MV stenosis severity, as well as the presence of secondary pulmonary hypertension and congestive heart failure, which are frequently linked with a poor prognosis. LS is an uncommon heart abnormality caused by the combination of congenital or iatrogenic ASD and congenital or acquired MS. The clinical characteristics and hemodynamic consequences[6]. The incidence rate of mitral stenosis in patients with atrial septal defect is 4%, while the rate of atrial septal defect in patients with mitral stenosis is 0.6% to 0.7%. The syndrome can occur at any age; however, it is most commonly seen in young adults. There is a predisposition for females because ASD and rheumatic MS are both more common in females[7].

Clinical suspicion of LS should trigger a thorough assessment using non-invasive and, when possible, invasive techniques. Echocardiography is the gold standard for diagnosing and

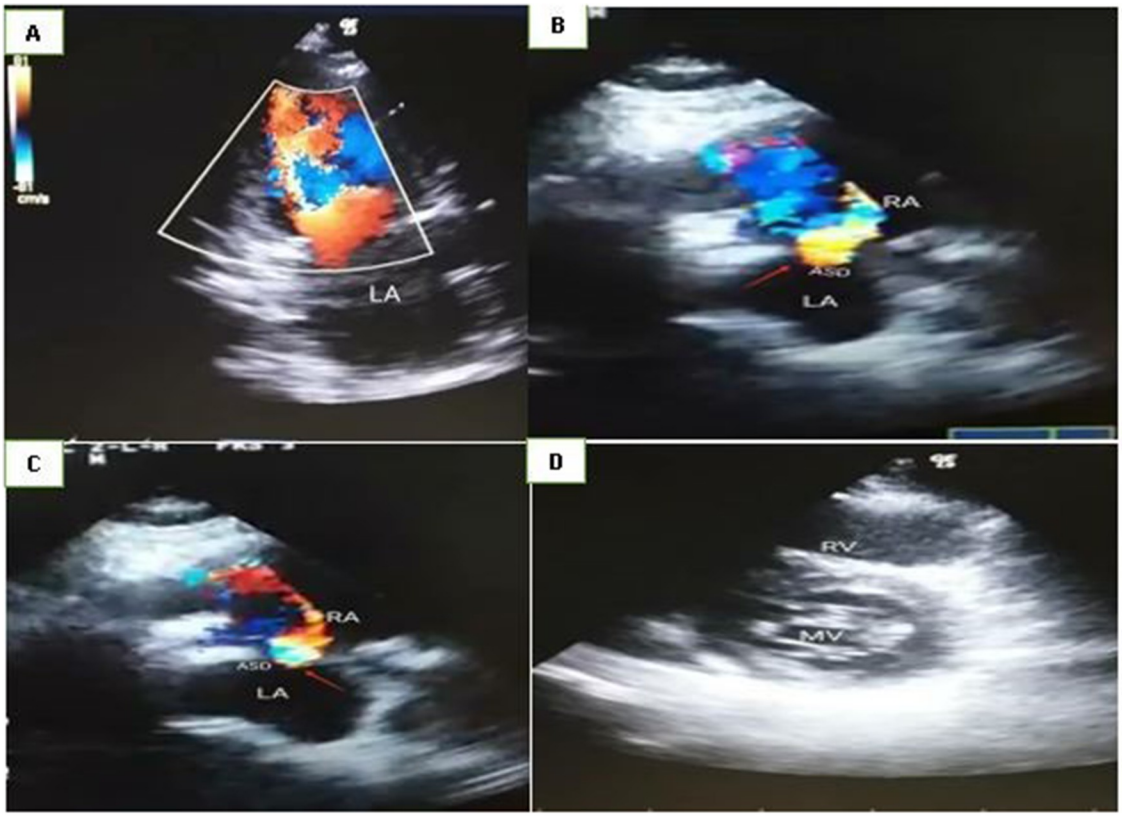

Figure 3. A: Apical two-chamber view shows color Doppler with aliasing due to stenosis at the level of the coronary artery. B: Subcostal view shows the interatrial septum with left-to-right flow through the opening between the atria using color Doppler. C: Subcostal view shows the opening between the atria with left-to-right flow through it using color Doppler. D: Short axis view at mitral valve level shows right ventricular enlargement (note RV) and reveals thickening and calcification of the mitral leaflets.

evaluating LS. Timely diagnosis is crucial for changing the natural course. At various phases of LS, 2-dimensional (2D) transthoracic echocardiography (TTE) reveals the following: left atrial enlargement, enlargement of right side cavities, ASD, pulmonary artery enlargement, and mitral valve stenosis. Color flow and Doppler imaging are also useful for confirming and

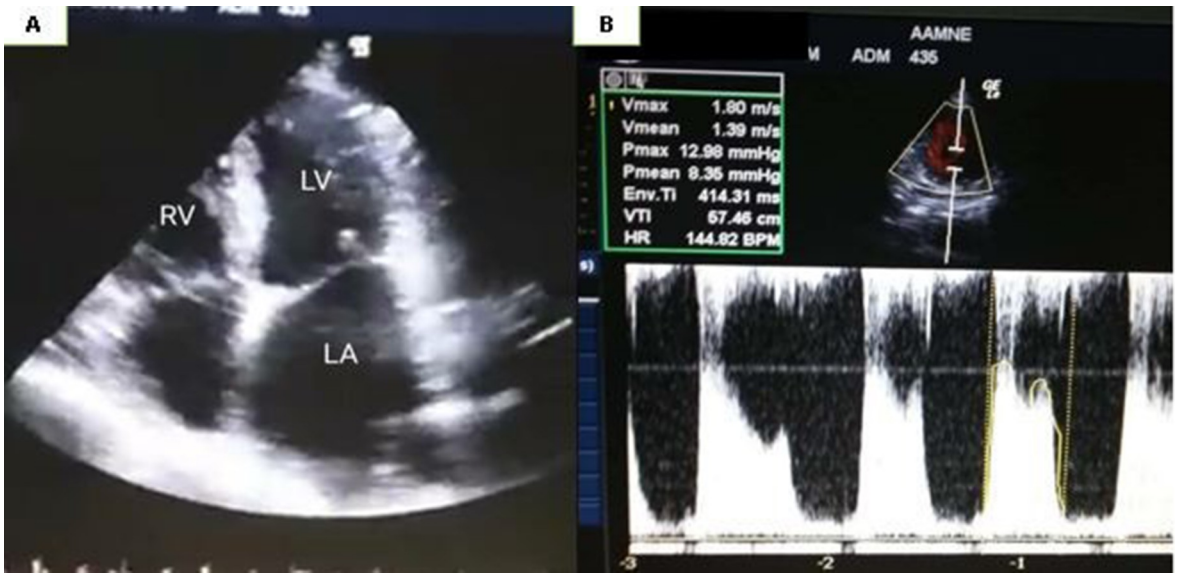

Figure 4. A: Apical four-chamber view without color Doppler shows left atrial enlargement and thickening of the mitral leaflets with signs of rheumatic involvement. B: Continuous wave Doppler across the mitral valve shows the mean pressure gradient, pressure half-time, and the valve area calculated by Doppler.

evaluating the severity of ASD, mitral valve stenosis and regurgitation, TR, and pulmonary pressure alterations[3]. In our case, the findings of echocardiography were a normal aortic root and significant dilation of the left atrium, measuring 5.5 × 6 cm. The left ventricle showed normal function with end-systolic and end-diastolic diameters ranging from 35 to 47 mm, with an EF of 60%. There was thickening and fibrosis of the leaflets, with signs of rheumatic involvement, and a "hockey stick" appearance of the anterior leaflet, and severe mitral stenosis. The mitral area was approximately 1.4 cm^2, with a mean gradient across the mitral valve of about 9 mmHg, and the pressure half-time (PHT) was measured at 153 ms. There was increased flow across the pulmonary valve and mild regurgitation in the tricuspid valve (1-2/4) with a velocity of 3.3 m/s, while the pulmonary artery systolic pressure ranged from 40 to 45 mmHg.

In cases of severe mitral stenosis, the mean gradient is usually over 10 mmHg; however, if an interatrial shunt is present, blood flow direction can shift from the left atrium to the right, resulting in a decrease in the mean gradient across the mitral valve. This may lead one to mistakenly believe that the stenosis is moderate when it is severe, as indicated by a gradient of 8 mmHg in this case.

ECG is another helpful diagnostic technique that can reveal atrial fibrillation in specific circumstances, right ventricular hypertrophy with right chamber overload, and right bundle branch block[8]. The same we saw in our case, except for atrial fibrillation.

CXR findings of LS patients with a small or limited ASD are consistent with mitral stenosis. Pulmonary venous congestion and pronounced left atrial enlargement are typically indicative[3]. The findings in this case were straightening of the left cardiac border and enlargement of the right ventricle.

In the past, the preferred course of treatment was surgery. Currently, however, percutaneous transcatheter treatment is an option for both conditions[9].

The most often utilized today are the Amplatzer septal occluder for percutaneous closure of ASD and the Inoue balloon for percutaneous balloon mitral valvuloplasty (PBMV)[3].

Open heart surgery was the preferred procedure for patients with LS involving ASD closure and mitral commisurotomy or valve replacement. Surgical intervention was recommended with an open heart surgery, resulting in the replacement of the mitral valve, which was irreparable, along with surgical closure of the interatrial septal opening using a patch. Upon follow-up after one and a half years, the patient was in a good general condition, on warfarin therapy, and doing well.

Conclusion

Combining ASD and mitral stenosis, the rare illness known as Lutembacher syndrome has a preserved prognosis and can result in cardiac failure and pulmonary hypertension if left untreated. An early surgical or percutaneous approach should be taken into consideration, before the patient experiences major symptoms and complications.

Ethical approval

Not applicable.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images and videos. A copy of the written consent is available for review by the editor of this journal.

Sources of funding

All the authors declare to have received no financial support or sponsorship for this study.

Author's contribution

All authors were responsible toward writing – original draft and all authors approved the final manuscript.

Conflicts of interest disclosure

All the authors declare to have no conflicts of interest relevant to this study.

Research registration unique identifying number (UIN)

Not applicable.

Guarantor

Bilal Sleiay.

Provenance and peer review

Not commissioned, externally peer-reviewed.

Data availability statement

Not applicable.

Methods

The work has been reported in line with the SCARE criteria[10].

Acknowledgements

We hope to thank SMSR Team Lab. for their efforts and bringing our team together.

References

[1] Pomerantzeff PMA, Bichuette LD, Veronese ET, *et al*. Case report total surgical correction of lutembacher syndrome associated with partial anomalous connection of the pulmonary veins and tricuspid regurgitation. Am J Cardiovasc Dis 2021;11:499–503.

[2] Phan QT, Nguyen HL, Le TD, *et al*. Combined percutaneous procedure in patient with lutembacher syndrome: a case report and real-world experience review.Cardiol Res 2018;9:385–91.

[3] Aminde LN, Dzudie A, Takah NF, *et al*. Current diagnostic and treatment strategies for Lutembacher syndrome: the pivotal role of echocardiography. Cardiovasc Diagn Ther 2015;5:122–32.

[4] Yadav R, Sharma A, Pathak S. A case series on successful surgical management of patients presenting with lutembacher's syndrome to a tertiary healthcare center in Northern India. Cureus 2023;15: e44371.
[5] Johny D, Subramanyam K, Bhandary RM, *et al*. surgical treatment of Lutembacher syndrome: combined percutaneous transcatheter therapy. BMJ Case Rep CP 2022;15:1–5.
[6] Yang M, Zhang L, Tang H, *et al*. Surgical treatment of lutembacher syndrome with a huge right atrium: a case report. Heart Surg Forum 2021;24:E359–62.
[7] Report LSAC. Journal of Cardiology and Cardiovascular Medicine. J Cardiol Cardiovasc Med 2019;4.
[8] Aredes SCA, da S SG, Carneiro SDS, *et al*. Lutembacher syndrome: case report and literature review. Arq Bras Cardiol – Imagem Cardiovasc 2018;31:2017–19.
[9] Ho CL, Liang KW, Fu YC, *et al*. Transcatheter therapy of lutembacher syndrome. J Chinese Med Assoc 2007;70:253–56.
[10] Sohrabi C, Mathew G, Maria N, *et al*. The SCARE 2023 guideline: updating consensus Surgical CAse REport (SCARE) guidelines. Int J Surg Lond Engl 2023;109:1136.