



LUTEMBACHER SYNDROME: A RARE CASE REPORT

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ABSTRACT

Lutembacher syndrome (LS) is a rare cardiac condition characterized by the coexistence of atrial septal defect (ASD) and mitral stenosis (MS). Although first described in 1916, its definition has since broadened to include combinations of congenital or acquired ASD with congenital or rheumatic MS. The hemodynamic interplay between these two lesions creates a distinctive clinical profile. Left atrial decompression via left-to-right shunting may delay symptoms of pulmonary congestion typically seen in isolated MS, while chronic right-sided volume overload contributes to early development of pulmonary arterial hypertension (PAH), right ventricular dilation, arrhythmias, and heart failure. We report the case of a 53-year-old woman who presented with progressively worsening dyspnea, pedal edema, and atrial fibrillation. Physical examination revealed signs of right-sided heart failure, including elevated jugular venous pressure and peripheral edema. Chest imaging demonstrated cardiomegaly, and echocardiography confirmed a large ostium secundum ASD with moderate MS, severe tricuspid regurgitation, severe PAH, and preserved left ventricular systolic function. Laboratory evaluation additionally revealed renal dysfunction and newly diagnosed hypothyroidism.

The patient's constellation of findings supported a diagnosis of Lutembacher syndrome. Given the advanced stage of PAH, right-sided dilation, arrhythmia, and multiorgan involvement, a multidisciplinary evaluation was undertaken to optimize clinical management. While medical therapy provided symptomatic relief, definitive management of both the ASD and MS was considered essential to prevent further clinical deterioration. This case underscores the importance of early recognition and comprehensive evaluation of patients with long-standing dyspnea, particularly in regions with high prevalence of rheumatic heart disease. Timely intervention can significantly improve outcomes and reduce complications such as right-heart failure and arrhythmias.

Keywords: Lutembacher syndrome, Atrial septal defect, Mitral stenosis, Pulmonary arterial hypertension, Atrial fibrillation, Rheumatic heart disease, Right-heart failure

Introduction

Lutembacher syndrome (LS) is a rare and complex cardiac condition defined by the coexistence of an atrial septal defect (ASD) and mitral stenosis (MS) [1]. First described by René Lutembacher in 1916, the nomenclature and criteria for this syndrome have evolved significantly over time [2]. Currently, LS is most broadly defined as the association of any ASD (which may be congenital or iatrogenic) with any MS (that may be congenital or acquired) [3]. Some authors have even expanded the spectrum to include ASD in conjunction with mitral regurgitation (MR), although MS remains the archetypal mitral lesion in LS [4].

Epidemiological data are scarce, but available literature estimates that MS is present in about 4% of patients with ASD, while conversely ASD is found in about 0.6%–0.7% of those with MS [5]. The condition is more frequently reported in regions with a high burden of rheumatic heart disease, such as Southeast Asia and other developing countries, which contributes to its clinical relevance in these populations [6]. Although the syndrome may manifest at any age, it is more commonly encountered in young adults and shows a female preponderance [3,7].

The hemodynamic interplay in LS is distinctive: the ASD offers a decompressing outlet for the elevated left atrial pressure caused by MS, which may delay the classic manifestations of left-sided heart failure. At the same time, the shunting of blood into the right heart places a volume load on the right atrium and ventricle, accelerates development of pulmonary arterial hypertension (PAH), and increases the risk of right-heart failure and arrhythmias such as atrial fibrillation [5,8]. In patients with a large, non-restrictive ASD and moderate MS, symptoms may initially resemble those of isolated ASD—whereas a restrictive ASD with severe MS often presents more like classical MS [9].

From a therapeutic standpoint, LS demands recognition of both lesions—the mitral valve pathology and the septal defect—to achieve optimal outcomes. While isolated MS may often be treated with balloon mitral valvuloplasty or mitral valve surgery, LS typically requires dual intervention: closure of the ASD along with repair/replacement or valvuloplasty of the mitral valve [10]. Timely diagnosis and management are essential to improve prognosis, reduce complications such as pulmonary hypertension, right-heart failure and paradoxical embolism, and to tailor the intervention strategy to the individual patient's anatomy and hemodynamics.

Lutembacher syndrome is a rare but clinically important entity, particularly in regions endemic for rheumatic mitral disease. Its recognition hinges on careful cardiac assessment and echocardiographic evaluation. Given its complex hemodynamics, a multidisciplinary approach is crucial for effective management and improved patient outcomes.

CASE PRESENTATION

A 53-year-old right-handed woman from Neemuch, Madhya Pradesh, presented to the Medicine Department of MB Government Hospital with complaints of progressively worsening shortness of breath for 16 years. The dyspnea initially occurred only on exertion but gradually progressed to occur even at rest during the past week. She also reported intermittent bilateral pedal edema for 5 years, worsening on prolonged standing and partially relieved with rest. Associated symptoms included a non-productive cough without wheeze or hemoptysis. There was no history of chest pain, syncope, or reduced exercise tolerance prior to the onset of respiratory complaints.

Sixteen years earlier, the patient recalled an episode of fever accompanied by generalized joint pain, which resolved after oral medications. Approximately four months later, she began experiencing exertional breathlessness that progressively deteriorated over the years. There was no history suggestive of tuberculosis, diabetes mellitus, hypertension, or substance abuse. She had been on regular cardiovascular medications, including digoxin, torsemide, and losartan, since 2007.

Physical Examination

On examination, the patient appeared cooperative but was in mild respiratory distress. Her pulse was irregularly irregular at 138 beats per minute with an apex-pulse deficit of 16. Blood pressure was 92/66 mmHg. Prominent distension of neck veins was noted predominantly on the left side, along with bilateral pitting pedal edema. Inspection revealed a precordial bulge and visible venous engorgement over the anterior chest wall (Figure 1).

Figure 1: Engorged neck veins over left side and prominence of veins over chest



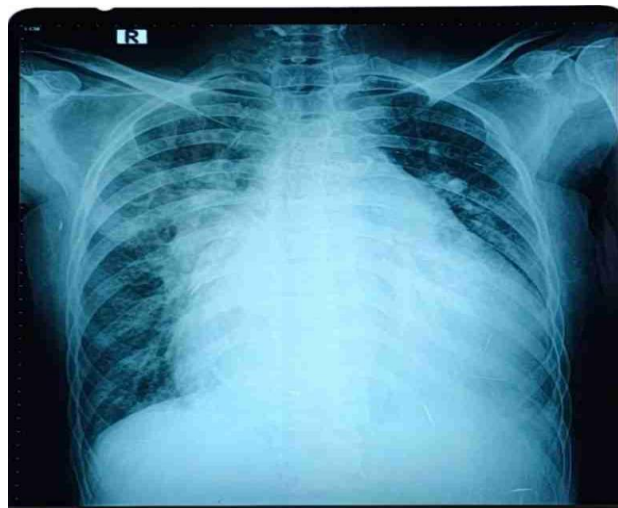
The apex beat was localized in the left 6th intercostal space, lateral to the mid-clavicular line, approximately 12.5 cm from the mid-sternal line, and was hyperdynamic. Auscultation revealed features suggestive of severe mitral regurgitation (MR), severe tricuspid regurgitation (TR), and pulmonary arterial hypertension (PAH). A high-pitched grade 3 ejection systolic murmur was audible in the left 2nd intercostal space, with increased intensity on leaning forward. A wide and fixed splitting of S2 was also noted. The aortic and neo-aortic areas were normal, with no additional sounds.

Investigations performed

Electrocardiography (ECG): Irregularly irregular rhythm with absent P waves; right axis deviation and right ventricular hypertrophy (RVH) with strain pattern, suggestive of atrial fibrillation with fast ventricular response and RVH.

Chest X-ray: Cardiomegaly with dilated left atrium, right atrium, and right ventricle. (Figure 2)

Figure 2: Chest Radiograph



2D Echocardiography: Atrial fibrillation during study; 34 mm defect in interatrial septum with echo dropout consistent with ASD. Moderate mitral stenosis, grade II MR, doming tricuspid valve, severe TR, severe PAH, and mild pulmonary regurgitation noted. Pulmonary artery was dilated. Dilatation of LA (4.3 cm), RA, and RV was present. Mild pericardial effusion was seen. Paradoxical interventricular septal motion and D-shaped left ventricular cavity were noted. No regional wall motion abnormality; LVEF was 50% with preserved biventricular systolic function. IVC was dilated (22 mm) and non-collapsible. No intracardiac thrombus or vegetation was visualized.

Laboratory Findings: Hemoglobin 14.5 g/dL; total leukocyte count 9,800/mm³; platelets 2.46 lakh/mm³. Urea 135 mg/dL; creatinine 2.2 mg/dL. Total bilirubin 0.7 mg/dL; SGOT 37 U/L; SGPT 28 U/L; ALP 126 U/L. Viral markers for HIV, HBV, and HCV were non-reactive. PT 16 s, INR 1.12. TSH elevated at 13 μ IU/mL; T3 1.1; T4 92. Troponin-T 0.05. ASO titers, rheumatoid factor, and VDRL were non-reactive.

Doppler evaluation of neck vessels revealed thrombus in the right internal jugular vein. The left internal jugular vein appeared normal.

Final Diagnosis: Acquired rheumatic heart disease with moderate mitral stenosis and moderate mitral regurgitation; severe tricuspid regurgitation; severe pulmonary arterial hypertension; atrial septal defect; atrial fibrillation; congestive heart failure (NYHA class IV) — consistent with Lutembacher syndrome, with newly diagnosed hypothyroidism.

DISCUSSION

Lutembacher syndrome (LS) is a rare but clinically significant entity arising from the coexistence of an atrial septal defect (ASD) and mitral stenosis (MS) [1]. In the prototypical form of LS, the ASD is congenital (commonly ostium secundum) and the MS is acquired (usually rheumatic) [2]. However, modern definitions recognise that both lesions may be congenital or acquired, and that iatrogenic ASDs (e.g., residual defects after balloon mitral valvuloplasty) may also form part of the spectrum [3,11].

The pathophysiology and haemodynamics of LS are complex since the ASD and MS influence each other. Key determinants include the size and restrictiveness of the ASD, the severity of MS, right ventricle compliance, and pulmonary vascular resistance [9]. When the ASD is restrictive and MS is severe, the clinical picture resembles isolated MS: elevated left atrial pressure, pulmonary venous congestion, and left-sided heart failure dominate. Conversely, when the ASD is large and non-restrictive, left atrial decompression through the ASD mitigates left-sided symptoms, while the left-to-right shunt increases right atrial and right ventricular volume load, promoting earlier right-heart enlargement, pulmonary hypertension and right-sided failure [9,12].

In LS, the presence of a large, non-restrictive ASD reduces left atrial pressure and thus may delay the onset of typical MS symptoms (such as orthopnoea or PND), but this is accompanied by increased volume load in the right heart and accelerated pulmonary vascular damage [1,13]. The left-to-right shunt reduces left ventricular preload and systemic cardiac output, generating fatigue and exercise intolerance. It also causes atrial enlargement (particularly RA), which predisposes to atrial fibrillation—the very arrhythmia evident in our patient [7]. The combination of MS and ASD thus creates a milieu of mixed haemodynamics: decreased LV filling, right heart volume overload, early pulmonary hypertension and the risk of paradoxical embolism if shunt reversal occurs [9].

Our patient exhibits many of these hallmark features: a longstanding history of rheumatic MS, large ASD, atrial fibrillation, severe tricuspid regurgitation (secondary to RV dilatation), severe PAH and signs of right-heart failure. The decade-long progression without earlier intervention underlines the fact that LS may present late once right-sided consequences dominate, rather than the classic left-

sided symptoms of MS. This emphasises the need for heightened vigilance in patients with rheumatic MS or ASD, especially in regions with high rheumatic burden.

From a management perspective, LS poses unique challenges. Medical therapy (diuretics, rate control for AF, anticoagulation where indicated) is essential but cannot correct the underlying structural lesions. The definitive therapy requires addressing both components—the MS (via balloon mitral valvuloplasty [BMV], commissurotomy, repair or replacement) and the ASD (via surgical closure or percutaneous device closure) [3,14]. Historically, surgical repair—closing the ASD and performing mitral valve surgery—was the standard. Studies cite favourable long-term outcomes when both lesions are treated simultaneously [15]. However, in the modern era, percutaneous approaches have become increasingly feasible in selected patients: simultaneous or staged BMV plus ASD device closure (or hybrid approaches) have been reported with good success [11,13].

Key issues in decision-making include: (a) timing of intervention (early before irreversible pulmonary vascular disease sets in); (b) anatomical suitability for percutaneous intervention (septal rims, ASD size, mitral valve morphology, presence of calcification); (c) managing the sequence—some advocate BMV first then ASD closure, others perform both in single setting; (d) assessing and managing secondary pulmonary hypertension and right ventricular dysfunction, which may negatively impact outcomes [14,16]. When PAH and right ventricular decompensation are advanced, surgical therapy may carry higher risk and outcomes may be compromised.

In our case, the presence of severe PAH, right ventricular enlargement, tricuspid regurgitation, kidney dysfunction and atrial fibrillation increased the complexity. Optimal management required multidisciplinary input: cardiology, cardiac surgery/interventional cardiology, rhythmology and renal medicine. The decision to proceed with intervention must weigh operative risk against expected benefit; in some cases, staged percutaneous therapy may be preferred to surgical correction.

Prognostically, untreated LS carries a higher risk of right-sided heart failure, arrhythmias, thromboembolism and early mortality compared to isolated MS or ASD [3,17]. Early diagnosis and intervention are strongly associated with improved outcomes; indeed, echocardiography remains the gold-standard diagnostic modality to delineate anatomy, quantify shunt, assess mitral valve and right heart status [3,11]. In resource-limited settings or high-rheumatic-disease regions, earlier screening of MS patients for concurrent ASD (and vice versa) might facilitate earlier detection of LS before complication onset.

In summary, Lutembacher syndrome represents a “two-lesion” problem—one congenital, one acquired—that requires an integrated therapeutic strategy. In patients like ours presenting with advanced disease, timely recognition and individualised care planning are critical. The case emphasises the need for awareness of LS as a differential in patients with MS or unexplained right-heart failure, especially where rheumatic heart disease remains prevalent.

Conclusion

Lutembacher syndrome represents an uncommon but important combination of mitral stenosis and atrial septal defect, producing a unique hemodynamic profile that complicates diagnosis and management. Optimal outcomes rely on early recognition, detailed echocardiographic assessment, and timely correction of both lesions through surgical or percutaneous strategies. A multidisciplinary team approach is crucial, particularly in patients presenting with advanced pulmonary hypertension, arrhythmias, or right-heart failure.

This case highlights the need for heightened clinical suspicion in individuals with persistent cardiopulmonary symptoms, especially in regions where rheumatic heart disease remains prevalent. Greater awareness, earlier screening, and broader access to definitive interventions may significantly reduce morbidity and mortality. Continued research and systematic reporting of such cases will

contribute to improved diagnostic algorithms and management pathways for this rare but treatable condition.

Declarations

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