Lutembacher Syndrome Associated With Pulmonary Hypertension: The Importance of Early Diagnosis for Enabling Surgical Treatment

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Introduction

Lutembacher syndrome (LS) is defined by the simultaneous presence of an atrial septal defect (ASD) and mitral stenosis (MS). Although it is a rare condition, its clinical significance lies in the considerable hemodynamic impact and the potential for early progression to irreversible pulmonary hypertension (PH).

The exact prevalence of LS remains unknown. However, for estimation purposes, MS is considered the most frequent manifestation of rheumatic fever (RF) in young women,[1] and the prevalence of RF in developing countries ranges from 5 to 10 cases per 1,000 children. Furthermore, the incidence of congenital ASD in patients with MS is estimated at 0.6% to 0.7%.[2]

The hemodynamic effects of LS vary according to the size of the ASD, the severity of the MS, the compliance of the right ventricle (RV), and the degree of pulmonary vascular resistance (PVR). When MS is severe and the ASD has significant hemodynamic impact, there is a left-to-right shunt of blood flow from the left atrium (LA) to the right atrium (RA), which prevents a proportional increase in LA pressure relative to the severity of MS. In such cases, the development of pulmonary venous hypertension tends to occur more slowly. However, progressive dilation of the right heart chambers and pulmonary hyperflow are observed, as in the case described here. If left untreated, PVR tends to rise progressively, eventually leading to RV failure. Pulmonary arterial hypertension (PAH) in these patients is usually hyperkinetic, resulting from volume overload in the right chambers and pulmonary overcirculation, in contrast to the PAH seen in isolated severe MS, where the predominant mechanism is retrograde pressure overload. In cases where the ASD has no significant hemodynamic consequences, the shunt is minimal, and the clinical course resembles that of isolated MS.[3,4]

This case report was submitted to and approved by the Research Ethics Committee at Universidade de Brasília under protocol number 86333025.3.0000.5558.

Case report

A 54-year-old woman sought medical attention with complaints of dyspnea on minimal exertion, palpitations, dry cough, and lower limb edema. She reported a history of heart disease since 2017, with multiple hospital admissions over the years. Physical examination revealed signs of systemic and pulmonary congestion, a 3+/6+ rumbling diastolic mitral murmur, and a fixed splitting of the second heart sound.

Electrocardiography (ECG) showed atrial flutter and signs of right heart chamber overload (Figure 1). Chest radiography revealed enlargement of the right chambers and pulmonary congestion (Figure 2). Transthoracic echocardiography demonstrated an ostium secundum-type ASD measuring 25 mm, with a pulmonary-to-systemic flow ratio (Qp/Qs) of 2.5 and evidence of a bidirectional shunt. Right heart chamber enlargement was observed, with a right atrial (RA) volume of 92 mL/m^2, a right ventricular (RV) basal diameter of 63 mm, and a mid-ventricular diameter of 44 mm. There was also moderate RV dysfunction with diffuse hypokinesia (TAPSE: 15 mm; FAC: 25%). Severe MS was also identified, with a mitral valve area of 1.2 cm^2 by planimetry and a mean gradient of 9 mmHg. The Block Wilkins score was 11 (thickness: 2; calcification: 2; mobility: 3; subvalvular: 2). The estimated systolic pulmonary artery pressure (SPAP) was 60 mmHg. The diagnosis of LS was then established (Figure 3 and Video 1).

In addition, as part of the ongoing investigation of PH, a pulmonary artery computed tomography angiography was performed to rule out chronic pulmonary thromboembolism as a potential associated mechanism. The examination revealed enlargement of the pulmonary artery trunk, dilation of the right heart chambers, and no evidence of obstruction in the pulmonary artery or its branches (Figure 4).

After clinical optimization with diuretics, a beta-blocker, and sildenafil, the patient underwent right heart catheterization, which confirmed PH of mixed etiology. Hemodynamic findings included a mean pulmonary artery pressure of 49 mmHg, a pulmonary artery wedge pressure of 20 mmHg, a PVR of 4.18 Wood units, and a pulmonary diastolic pressure gradient of 10 mmHg. During the procedure, a favorable vasodilator response to nitric oxide was observed, indicating that the PH was not fixed. Surgical correction

Keywords

Lutembacher Syndrome; Pulmonary Hypertension; Mitral Valve Stenosis; Atrial Heart Septal Defects.

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Manuscript received May 17, 2025; revised May 19, 2025; accepted June 21, 2025
Editor responsible for the review: Andrea Vilela

DOI: https://doi.org/10.36660/abcimg.20250043i

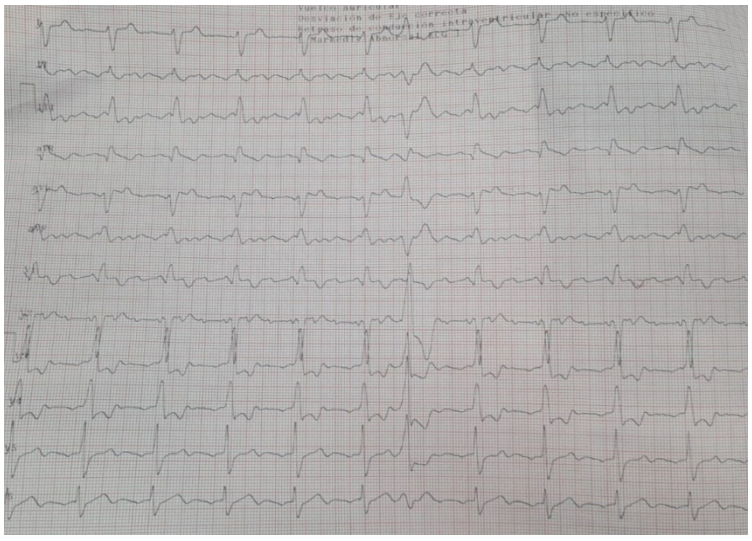


Figure 1 – *12-lead resting ECG showing atypical atrial flutter, right axis deviation, and right heart chamber overload. Source: Internal records of the Hospital Universitário de Brasília.*

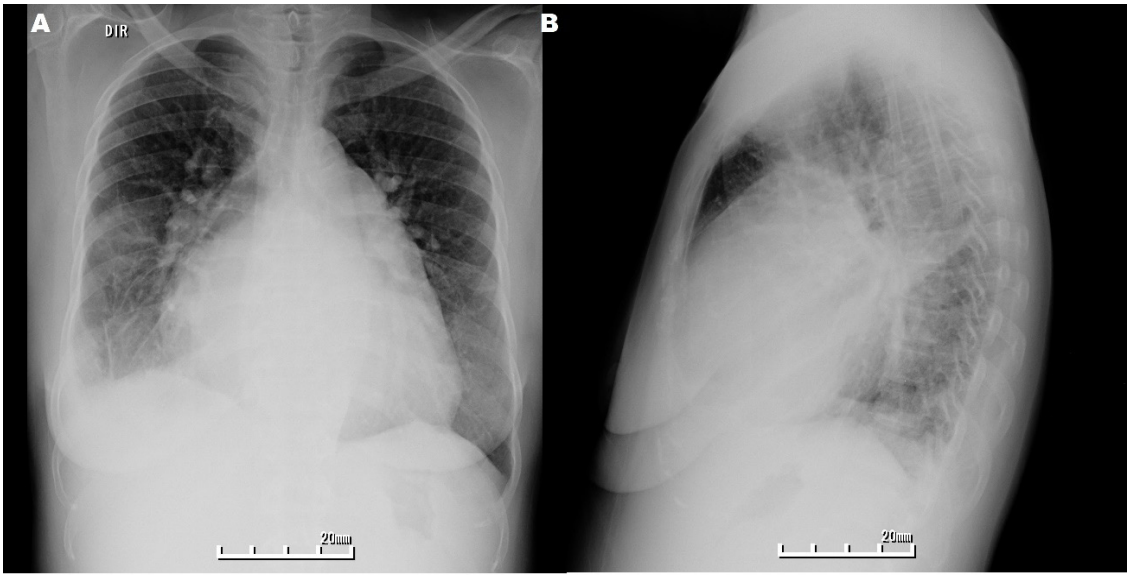


Figure 2 – *Chest radiography. A) Posteroanterior view and B) lateral view showing cardiomegaly and enlargement of the right heart chambers. Source: Internal records of the Department of Radiology, Hospital Universitário de Brasília.*

of the MS and ASD was therefore indicated. She is currently awaiting the procedure.

Discussion

The initial course of LS is often oligosymptomatic, as the presence of the ASD relieves LA overload caused by MS, delaying the onset of symptoms and the development of PH. However, chronic shunting leads to pulmonary overcirculation and progressive overload of the right heart chambers, as observed in the present case.[4,5] A favorable vasodilator response during right heart catheterization suggests the potential reversibility of PH and serves as a key criterion for indicating surgical treatment.[6,7]

Isolated MS is characterized by thickening of the valve leaflets and restricted mitral valve opening, resulting in increased LA pressure, postcapillary PH, and overload of the right

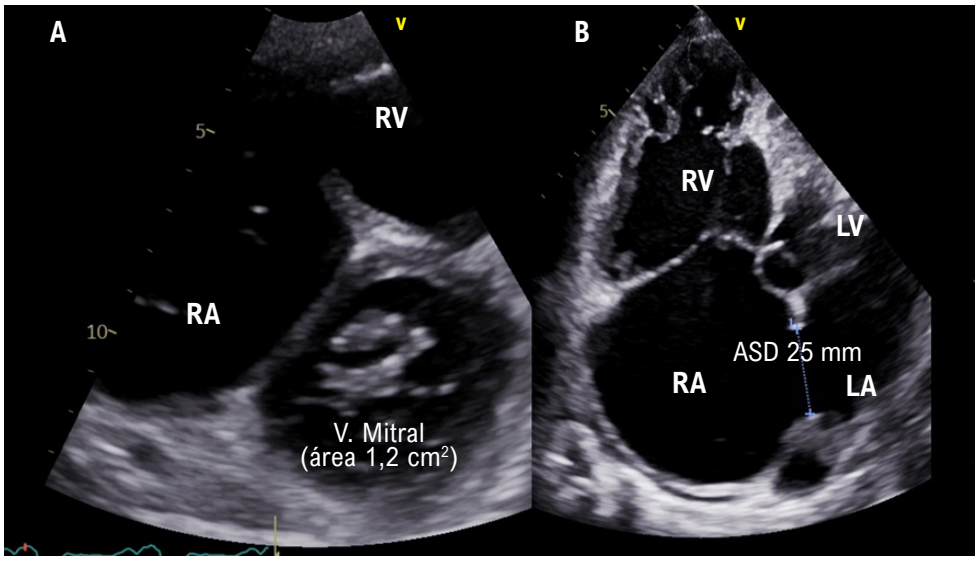
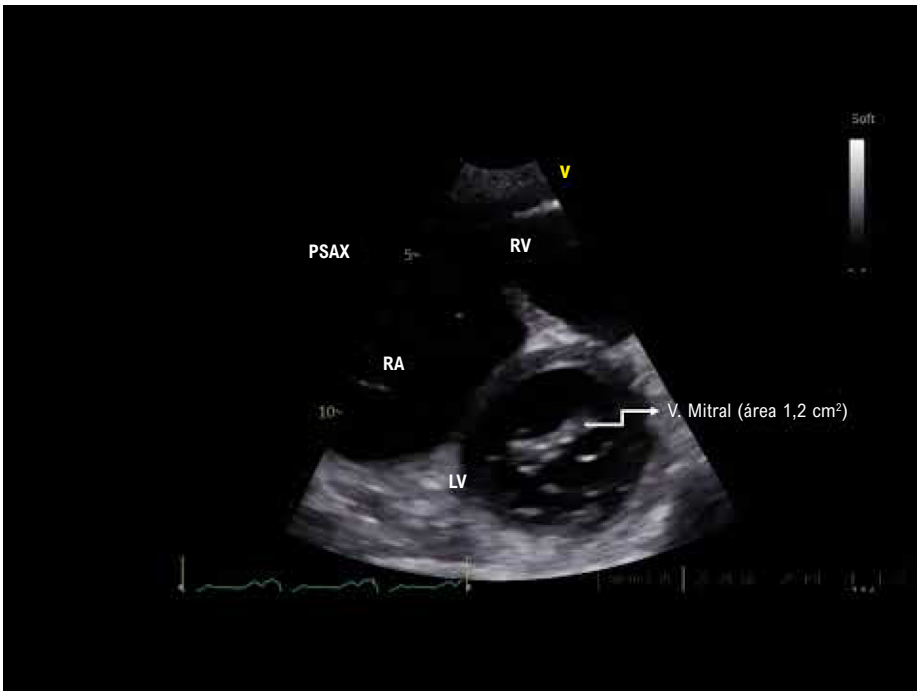


Figure 3 – *Transthoracic echocardiography. A) Parasternal short-axis view showing commissural fusion of the mitral valve and reduced valve area. B) Apical four-chamber view focused on the RV, showing a 25 mm ASD, biatrial enlargement, and RV enlargement. ASD: atrial septal defect; LA: left atrium; LV: left ventricle; RA: right atrium; RV: right ventricle; V: valve. Source: Internal records of the Department of Echocardiography, Hospital Universitário de Brasília.*



Video 1 – *ASD: atrial septal defect; LA: left atrium; LV: left ventricle; PLAX: parasternal long-axis view; PSAX: parasternal short-axis view; RA: right atrium; RV: right ventricle; V: valve. Source: Internal records of the Department of Echocardiography, Hospital Universitário de Brasília.*
Link: http://abcimaging.org/supplementary-material/2025/3803/2025-0043_video_01.mp4

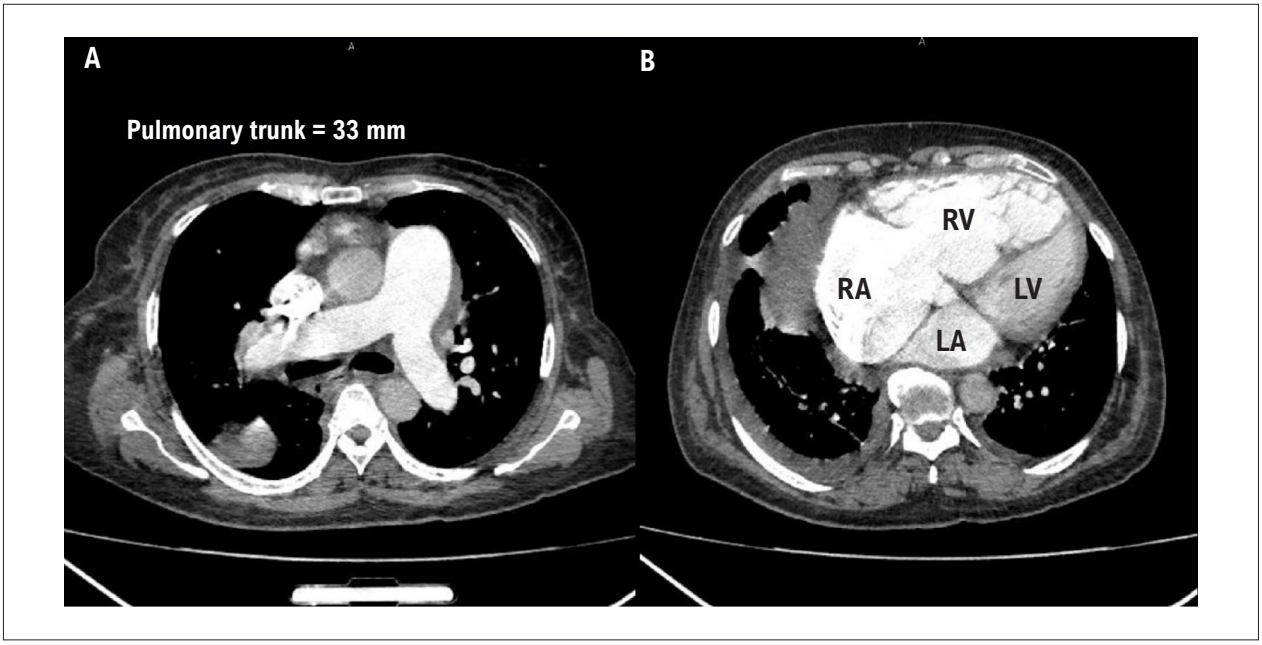


Figure 4 – *Pulmonary artery computed tomography angiography showing dilation of the pulmonary artery trunk and enlargement of the right heart chambers. LA: left atrium; LV: left ventricle; RA: right atrium; RV: right ventricle. Source: Internal records of the Department of Radiology, Hospital Universitário de Brasília.*

heart chambers. Intervention is indicated when MS is severe and symptomatic, or in asymptomatic patients with complicating factors.[8] Anatomical severity is defined by a mitral valve area less than 1.5 cm^2, a mean left atrioventricular diastolic gradient greater than 10 mmHg, and SPAP exceeding 50 mmHg. The main complicating factors include the development of PH and new-onset atrial fibrillation.[8] In LS, the ASD decompresses the LA, which may lead to underestimation of the transmitral gradient. In such cases, mitral valve area assessment by planimetry is considered more reliable.

ASD accounts for 5% to 10% of congenital heart diseases and is more prevalent in females. The ostium secundum type represents 50% to 70% of cases, with the defect located in the central portion of the atrial septum. This communication between atria creates a left-to-right shunt, which results in pulmonary overcirculation.[6] Both components of LS — MS and ASD — can individually lead to the development of PH; however, when combined, their hemodynamic effects are amplified.[7,9] Early diagnosis and timely intervention in LS are essential to prevent the progression of PH to irreversible forms.[5]

Surgical intervention for ASD is indicated in the presence of a left-to-right shunt, dilation of the right heart chambers, and a Qp/Qs ratio greater than 1.5. In addition, SPAP must be less than 50% of the systemic systolic pressure, PVR must be less than one-third of systemic vascular resistance, and there must be no cyanosis at rest or during exertion.[10] Closure of the ASD is contraindicated in the presence of Eisenmenger syndrome, which is characterized by irreversible PH with reversal of the shunt to right-to-left.[9,10]

Clinical management of LS includes the use of diuretics to relieve congestive symptoms, as well as beta-blockers and calcium channel blockers for heart rate control, particularly in cases with associated atrial arrhythmias. Prophylaxis for infective endocarditis should also be considered.[4,7]

Regarding interventional correction, the classical approach has been open-heart surgery. However, with advances in percutaneous techniques, transcatheter therapy — through valvotomy for MS and ASD closure using septal occluder devices — has emerged as a viable alternative.[7] In the present case, despite the lower morbidity and mortality associated with percutaneous treatment, open surgical correction was chosen due to a contraindication for balloon valvuloplasty.[10]

Conclusion

LS is a rare, frequently underdiagnosed condition. LS should be considered in the evaluation of patients presenting with signs of MS, PH, and disproportionate right heart chamber overload relative to the severity of the valvular lesion. Early diagnosis, combined with thorough hemodynamic assessment, is essential to determine the optimal timing for intervention and to help prevent the progression of PH to irreversible forms.

Author Contributions

Conception and design of the research, acquisition of data, analysis and interpretation of the data, statistical analysis, writing of the manuscript and critical revision of the manuscript for intellectual content: Lopes ALC, Otto ME, Mello BCR, Sena MB, Gali WL.

Potential Conflict of Interest

No potential conflict of interest relevant to this article was reported.

Sources of Funding

There were no external funding sources for this study.

Study Association

This article is part of Ana Luiza Caldeira Lopes' graduation thesis at the University Hospital of Brasília.

Ethics Approval and Consent to Participate

This study was approved by the Ethics Committee of the Faculdade de Medicina da Universidade de Brasília under the protocol number 86333025.3.0000.5558. All the procedures in this study were in accordance with the 1975 Helsinki Declaration, updated in 2013. Informed consent was obtained from all participants included in the study.

Use of Artificial Intelligence

The authors did not use any artificial intelligence tools in the development of this work.

Availability of Research Data

The underlying content of the research text is contained within the manuscript.

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