

Lutembacher Syndrome: Case Report and Literature Review

Susan Caroline Azevedo Aredes,¹ Gabriela da Silva Scopel,¹ Stella de Souza Carneiro,¹ Josana Azevedo Aredes,² Aline Valério de Lima,¹ Patrick Ventorim Costa¹

Hospital Universitário Cassiano Antonio de Moraes,¹ Escola Superior de Ciências da Santa Casa de Misericórdia de Vitória,² Vitória, ES - Brazil

Introduction

The Lutembacher syndrome (LS) is a rare condition, defined as a combination of mitral stenosis and an ostium secundum atrial septal defect (ASD). Both defects, ASD and mitral stenosis, may be congenital or acquired, and present a number of hemodynamic repercussions according to: disease evolution time, ASD size, and severity of mitral valvopathy. We report a case of this syndrome in a young oligosymptomatic patient, but with well-established cardiovascular disorders.

Case Report

Seventeen-year-old female patient, born and living in Espírito Santo – Brazil, previously healthy and asymptomatic. In September/2016, during physical activity at school, the patient had sudden dyspnea of moderate intensity, associated with intense sharp chest pain, lasting about 10 minutes, requiring interruption of the exercise.

After one (1) week of symptoms, she sought medical attention with a cardiologist. At physical examination, she was in good general condition, acyanotic, with respiratory auscultation without any disorders. Cardiac auscultation evidenced 3+/6+ systolic murmur more audible in the upper left sternal border, with fixed and wide split of the second sound. The precordium was hyperdynamic and it was possible to palpate right ventricular systolic impulses. Blood pressure 110/70 mmHg, heart rate 70 bpm, low amplitude and symmetrical peripheral pulses, no abnormalities in abdominal and lower limb examination. She had brought normal laboratory test results, such as hemogram, renal function, lipidogram and thyroid function.

Chest X-ray showed enlarged cardiac area and electrocardiography showed sinus rhythm with right axis deviation and overload of right chambers (Figure 1).

Transthoracic echocardiography was performed one month after the first consultation for diagnostic elucidation, which showed a 55% left ventricular ejection fraction, severe increase in right chambers (right ventricle with 56 mm basal, 44 mm

mean and 78 mm longitudinal dimensions), ostium secundum atrial septal defect (ASD) measuring about 2.0 cm, with a large shunt from the left atrium to the right atrium.

Mitral valve with reduced opening, no calcification or significant thickening of cusps, average gradient of 11 mmHg and valvular area calculated by the planimetry of 1.3 cm², representing moderate to severe mitral valve stenosis of probable congenital etiology (Figure 2). The test also revealed pulmonary hypertension with pulmonary artery systolic pressure estimated at 50 mmHg, associated with pulmonary artery trunk dilatation, measuring approximately 3.7 cm without evidence of pulmonary valve stenosis or significant insufficiency.

The patient remained oligosymptomatic throughout this period, maintaining some sporadic episodes of dyspnea on exertion (functional class II), and non-specific chest pain.

Based on the clinical and imaging findings, Lutembacher Syndrome (LS) was diagnosed and the patient was referred for consultation for pediatric cardiac surgery. Despite efforts to provide adequate treatment and thorough explanations of disease progression and prognosis, the patient and her family chose to remain under clinical treatment and routine outpatient follow-up. However, she goes to outpatient clinics irregularly, not properly continuing clinical treatment.

Literature Review

The Lutembacher Syndrome (LS) is a very rare disease whose incidence in a study published in the American Heart Journal in 1997 was 0.001/1,000,000.¹ It was first described in 1865 by Martineau, and revised by Lutembacher in 1916.²

It consists of a combination of ostium secundum atrial septal defect and mitral stenosis.¹ Opinions differ as to the type of mitral lesion that should be included in this syndrome. In some papers, it is simply described as acquired mitral stenosis. Today, however, the recognition of LS includes congenital or acquired disorders, both in mitral stenosis and in atrial septal defect (ASD).² This defect in LS may also be iatrogenic, secondary to transeptal puncture during mitral valvuloplasty.³ A study published in 2016 in India, a country with a high prevalence of rheumatic disease, assessed 44 autopsies of patients with Lutembacher syndrome and 54.5% of patients had non-rheumatic mitral valve disease.⁴

The hemodynamic repercussion resulting from this syndrome depends on the ASD size and the severity of the valve stenosis, which may lead to varying degrees of pulmonary hypertension, right chamber dilatation and tricuspid valve failure. Generally, since right ventricular compliance is lower, blood flow goes through the left atrial septal defect into the right atrium, leading to dilation and progressive dysfunction of the right chambers, as well as reduced blood flow to the left ventricle. Thus, in general, there will be no pulmonary congestion commonly found in isolated mitral stenosis,² and

Keywords

Lutembacher Syndrome/physiopathology; Heart Septal Defects, Atrial; Mitral Valve Stenosis; Echocardiography/diagnostic imaging; Stroke Volume; Hypertension Pulmonary.

Mailing Address: Susan Caroline Azevedo Aredes •

Rua Gelu Vervloet dos Santos, 950, ap. 1303, Edifício Green Residence.

Postal Code 29090-100, Bairro Jardim Camburi, Vitória, ES – Brazil

E-mail: susan_aazevedo@hotmail.com

Manuscript received November 30, 2017; revised December 13, 2017;

accepted February 15, 2018

DOI: 10.5935/2318-8219.20180016

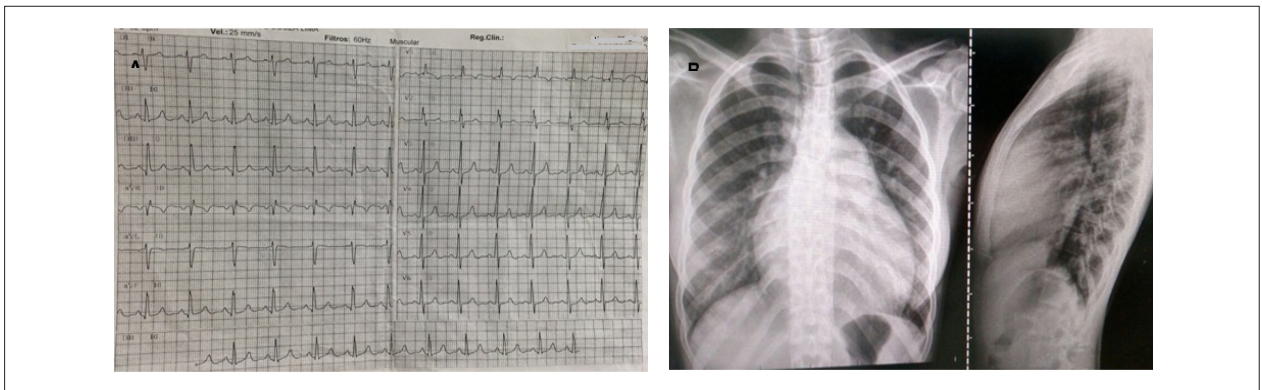


Figure 1 – Electrocardiography (A) showing sinus rhythm with right axis deviation and overload of right chambers. In B, chest X-ray with increased cardiothoracic index, bulging of the pulmonary artery trunk and obliteration of the retrosternal fat (finding compatible with enlarged right ventricle).

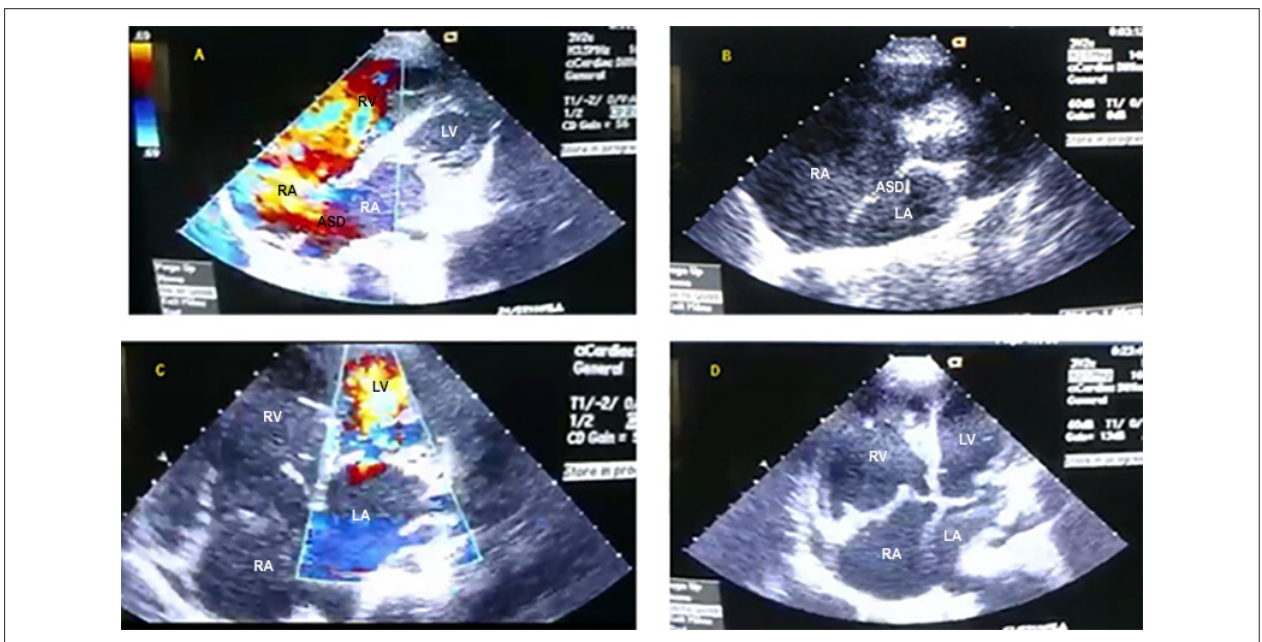


Figure 2 – Transthoracic echocardiogram showing in A and B an ostium secundum atrial septal defect with LA → RA flow. In C, swirling flow of mitral stenosis, and in D, overload of right chambers. RA: right atrium; LV: left ventricle; RV: right ventricle; ASD: atrial septal defect.

clinical presentations will most often be due to ASD, with variations in signs and symptoms depending on the ASD size. ASD in LS generally has a diameter greater than 1.5 cm, causing a severe left-right shunt, which can progress with progressive pulmonary hypertension and development of Eisenmenger's syndrome.³

Patients commonly report fatigue, exercise intolerance, palpitations, and are even more predisposed to develop atrial arrhythmias, of which atrial fibrillation (AF) is the most frequent one.³ Physical examination shows lower amplitude arterial pulses with regular rhythm and high jugular venous pulse (even in the absence of right heart failure, as well as absence of pulmonary hypertension). In addition to the classic findings of

mitral stenosis (hyperphonestic of the first heart sound, diastolic rhythm and opening click), it is common to hear the fixed split of the second heart sound (B2), which is typical of interatrial septal defect, as well as a proto or meso-diastolic murmur.³

As a workup, thorax radiography may show cardiomegaly and, occasionally, signs of pulmonary congestion. Electrocardiogram (ECG) may reveal right bundle branch block, right ventricular hypertrophy with right chamber overload and atrial fibrillation may be observed in some cases. Echocardiography is the gold standard method for diagnosing LS, with the benefit of not being invasive and widely available, as well as accurate for the evaluation of ASD and mitral valvopathy with its different severities.⁵

Early diagnosis with correction of mitral stenosis and closure of ASD, either percutaneously or surgically, indicate a good prognosis. However, if the diagnosis occurs late, already in the presence of severe pulmonary hypertension and heart failure, prognosis is reserved, generally recommending clinical treatment for relief of symptoms.¹

There are two well-described treatments of LS. Open surgical correction with ASD closure and mitral commissurotomy or valve replacement is traditionally performed. However, as interventional cardiology evolves, the approach has changed significantly, and now percutaneous therapy in eligible patients is the current treatment of choice with excellent success rates, performing balloon mitral valvuloplasty and septal occlusion with prosthetic device.^{6,7}

The percutaneous procedure of LS patient was performed for the first time in 1992 by Ruiz et al.⁸ This treatment, however, is challenging because of the anatomical characteristics of LS – small left atrium with mitral stenosis, and right atrium larger than the left one – making it difficult to handle the balloon in the correction of mitral stenosis.⁹

The most severe complication during the procedure is embolization of the septal device.¹⁰ Due to such difficulties, careful evaluation of the device size is recommended, with measures taken by transesophageal echocardiography, also considering the hemodynamicist's experience for the success of the procedure.⁷

References

1. Ali SY, Rahman M, Islam M, Barman RC, Ali MY. "Lutembacher's Syndrome" – A Case Report. *Faridpur Med Coll J.* 2011;6(1):59-60.
2. Kulkarni SS, Sakaria AK, Mahajan SK, Shah KB. Lutembacher's syndrome. *J Cardiovasc Dis Res.* 2012;3(2):179-81.
3. Aminde LN, Dzudie A, Takah NF, Ngu KB, Sliwa K, Kengne AP. Current diagnostic and treatment strategies for Lutembacher syndrome: the pivotal role of echocardiography. *Cardiovasc Diagn Ther.* 2015;5(2):122-32.
4. Vaideeswar P, Marathe S. Lutembacher's syndrome: is the mitral pathology always rheumatic? *Indian Heart J.* 2017;69(1):20-3.
5. Aminde LN, Dzudie A, Takah NF, Ambassa JC, Mapoh SY, Tanchou JC. Occurrence of Lutembacher syndrome in a rural regional hospital: case report from Buea, Cameroon. *Cardiovasc Diagn Ther.* 2014;4(3):263-6.
6. Goel S, Nath R, Sharma A, Pandit N, Wardhan H. Successful percutaneous management of Lutembacher syndrome. *Indian Heart J.* 2014;66(3):355-7.
7. Vadivelu R, Chakraborty S, Bagga S. Transcatheter therapy for Lutembacher's syndrome: the road less travelled. *Ann Pediatr Cardiol.* 2014;7(1):37-40.
8. Ruiz CE, Gamra H, Mahrer P, Allen JW, O'Laughlin MP, Lau FY. Percutaneous closure of a secundum atrial septal defect and double balloon valvotomies of a severe and aortic valve stenosis in a patient with Lutembacher's syndrome and severe pulmonary hypertension. *Cathet Cardiovasc Diagn.* 1992;25(4):309-12.
9. Kamana VK, Shetty R, Krishnan AM, Chowdary RK, Malpe UP. Is transoesophageal echocardiography necessary for the percutaneous management of Lutembacher Syndrome: a case report. *J Clin Diagn Res.* 2016;10(10):OD08-OD09.
10. Wilson NJ, Smith J, Prommete B, O'Donnell C, Gentler TL, Ruygrok PN, et al. Transcatheter closure of secundum atrial septal defects with the Amplatzer septal occluder in adults and children – follow-up closure rates, degree of mitral regurgitation and evolution of arrhythmias. *Heart Lung Circ.* 2008;17(4):318-24.

Conclusion

The Lutembacher syndrome is a rare disease characterized by a combination of mitral stenosis and ASD which, if not diagnosed and treated early, may lead to heart failure and pulmonary hypertension with a reserved prognosis. Therefore, early surgical or percutaneous approach should be considered, while the patient does not present any serious complications and can benefit from the procedure.

Authors' contributions

Data acquisition: Costa PV; Data analysis and interpretation: Costa PV; Manuscript drafting: Aredes SCA, Scopel GS, Carneiro SS, Aredes JA, Lima AV; Critical revision of the manuscript for important intellectual content: Aredes SCA, Scopel GS, Carneiro SS, Aredes JA, Lima AV, Costa PV.

Potential Conflicts of Interest

There are no relevant conflicts of interest.

Sources of Funding

This study had no external funding sources.

Academic Association

This study is not associated with any graduate program.