

## Case Report

# Total surgical correction of Lutembacher syndrome associated with partial anomalous connection of the pulmonary veins and tricuspid regurgitation

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**Abstract:** Lutembacher syndrome is characterized by the association of mitral stenosis and atrial septal defect (ASD), usually of the ostium secundum type. The association between superior vena cava-type ASD and partial anomalous pulmonary venous connection is unusual and there are few descriptions in the literature. We report this condition in a 24-year-old woman who was admitted to the hospital with a 1-year history of progressive dyspnea and describe the successful surgical repair following mitral commissurotomy and tunneling of the anomalous pulmonary veins to the left atrium, which determines the closure of the ASD and tricuspid repair.

**Keywords:** Lutembacher syndrome, mitral stenosis, atrial septal defect, anomalous pulmonary venous connection

### Introduction

Lutembacher syndrome is a rare combination of atrial septal defect (ASD), typically of the ostium secundum type, and mitral stenosis (MS) which is often caused by rheumatic disease. The clinical presentation varies according to the size of the ASD, severity of MS, and compliance of the right ventricle [1]. As a consequence of the left-to-right shunt caused by the combination of MS and ASD, right ventricle volume overload and progressive dilation of the tricuspid annulus lead to tricuspid functional regurgitation [2].

We report an unusual presentation of Lutembacher syndrome with superior vena cava-type ASD, which represents approximately 2-10% of all ASDs, associated with partial anomalous pulmonary vein connection and tricuspid functional regurgitation. Although approximately 10% of patients diagnosed with ASD present with partial anomalous pulmonary venous connection, the association with Lutembacher syndrome has rarely been described in the literature (Table 1) [3-5]. There is only one description of complete surgical correction of these concomitant structural heart disorders

[4]. We describe a case successfully treated with mitral commissurotomy, tunneling of the anomalous pulmonary veins to the left atrium, closure of the ASD, and tricuspid repair.

### Case report

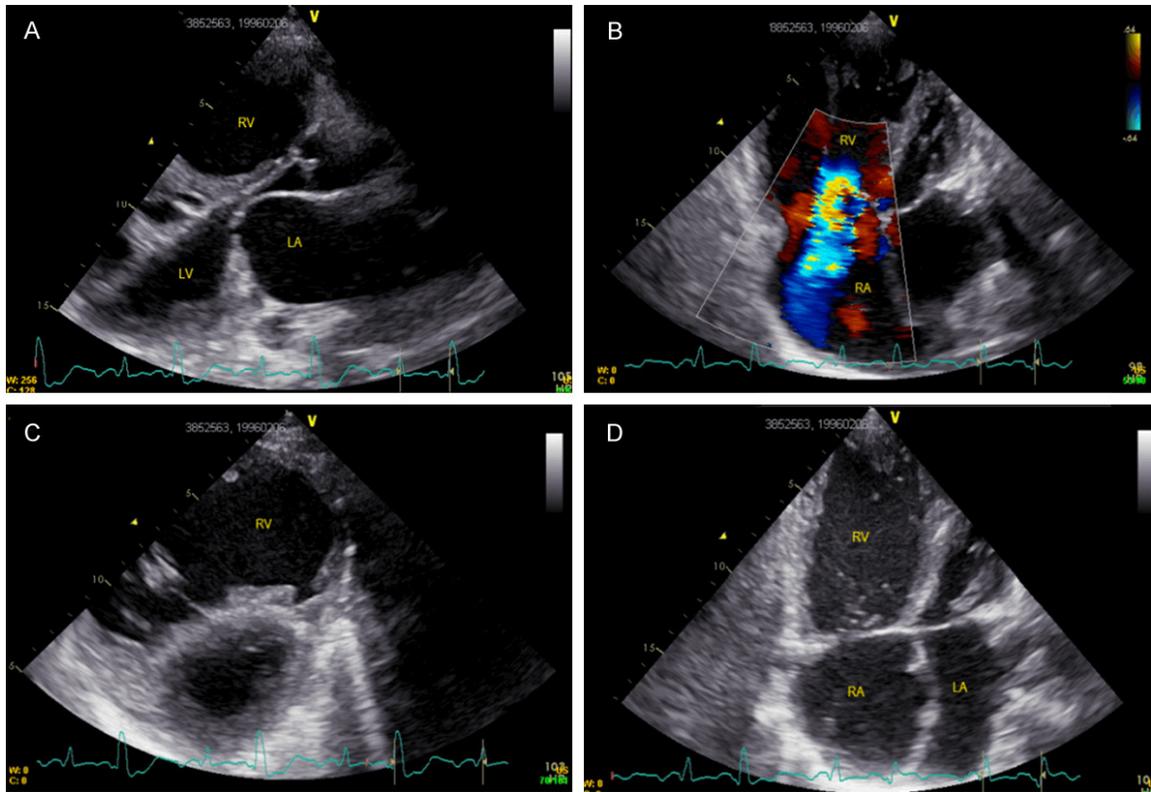
The patient was a 24-year-old previously healthy woman who presented because of experiencing dyspnea during daily physical activities (New York Heart Association Functional Class III). On physical examination, the patient was found to have a regular heart rhythm, normotensive blood pressure, hyperdynamic precordium with palpation of systolic impulses from the right ventricle and signs of right cardiac failure such as jugular turgescence and hepatojugular reflux. Auscultation revealed the presence of an early opening snap followed by a diastolic murmur with presystolic accentuation in the mitral focus, and regurgitative holosystolic murmur in the tricuspid focus with increased intensity after deep inspiration.

Electrocardiography revealed biatrial and right ventricular overload, and chest X-ray demonstrated enlargement of the cardiac area, mainly at the expense of the right chambers. Trans-

## Lutembacher syndrome and partial anomalous pulmonary venous connection

**Table 1.** Previous reports in current literature

Case	Source	Gender	Age	Association	Interventions	Postoperative
1	Anwar AM et al. [3]	F	28	Mitral stenosis + ostium secundum ASD + anomalous drainage of the left upper and lower pulmonary veins.	Open mitral commissurotomy + ASD closure with large Dacron patch + end-to-end anastomosis of vertical vein to left atrial appendage.	Sudden cardiorespiratory arrest two days after surgery.
2	Fradi El Faleh I et al. [5]	M	24	Mitral stenosis + superior vena cava-type ASD + anomalous drainage of pulmonary veins.	Mitral valve replacement + closure of ASD by PTFE patch + repositioning of pulmonary veins.	Postoperative 2D echo 10 days after surgery reveals normal functioning mitral valve with no residual shunt across the ASD patch and normal ventricular function.
3	Shende S et al. [4]	F	32	Rheumatic mitral stenosis + superior vena cava-type ASD + anomalous drainage of the right superior pulmonary vein.	Mitral valve replacement + simple patch closure by autologous untreated pericardial patch, which directs the pulmonary vein blood flow across the ASD into left atrium + modified DeVega's tricuspid ring annuloplasty.	Postoperative 2D echo reveals normal functioning mitral valve with no residual shunt across the ASD patch with no superior vena cava stenosis.



**Figure 1.** A: Two-dimensional echocardiographic view showing thickening and reduction of mitral valve opening; B: Important tricuspid regurgitation; C: Enlarged right ventricle; D: Enlarged right ventricle and right atrium. RA: right atrium; LA: left atrium; RV: right ventricle; LV: left ventricle.

thoracic echocardiography (TTE) revealed preserved left ventricular function, normal left ventricle dimensions, enlargement of the left atrium and right atrial dilation in association with severe pulmonary hypertension. Valvular analysis demonstrated functional tricuspid regurgitation and a mitral valve with rheumatic stigma (commissural fusion, thickening, and reduced cusp opening) leading to very severe MS (mitral valve area, 0.6 cm<sup>2</sup>; mean gradient 11 mmHg) with trivial regurgitation (**Figure 1**).

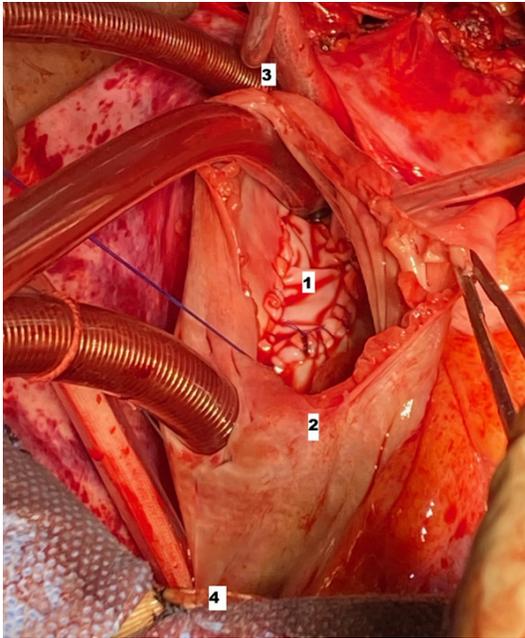
The patient underwent a surgical procedure with a median sternotomy and cardiopulmonary bypass. Intraoperative findings revealed a dilated right atrium associated with the right upper pulmonary vein in an unusual position, close to the opening of the superior vena cava. An extended vertical transatrial septal approach (Guiraudon incision) was used to expose of the mitral valve.

Both mitral valve commissurotomy and papillotomy were performed with an increase in the valvular orifice area. An intraoperative saline

test found no reflux. After closure of the atrial septum, we evaluated the right atrium and tricuspid valve, which had normal leaflets and central regurgitation secondary to the annular dilation. In addition, an incidentally partial anomalous connection of the right superior pulmonary vein associated with superior vena cava interatrial communication was noted.

A bicuspidization (Kay procedure) of the tricuspid valve was performed, and an improvement in cusp coaptation was observed in the intraoperative saline test. Due to the wide opening of the superior vena cava, the right upper pulmonary vein was tunneled to the left atrium using a bovine pericardium patch to ensure the closure of the superior vena cava-type communication following the reconstruction of the superior vena cava (**Figure 2**). Successful weaning of the cardiopulmonary bypass was achieved with no intraoperative complications.

Post-procedure TTE revealed preserved biventricular function, a mitral valve area of 1.8 cm<sup>2</sup>, trivial tricuspid regurgitation, and an absence



**Figure 2.** Intraoperative image after correction of anomalous connection, in addition to mitral and tricuspid plastics. (1) Pericardium patch correcting anomalous connection. (2) Incised right atrium. (3) Cannulated and looped upper vena cava. (4) Cannulated and looped inferior vena cava.

of pulmonary hypertension. Furthermore, a corrected connection of the right upper pulmonary vein to the left atrium without flow restrictions or atrial septum residual shunt was confirmed. The patient was discharged on the eighth postoperative day following an excellent recovery and remained asymptomatic during outpatient follow-up.

### Discussion

Lutembacher syndrome is a rare clinical condition that is more common in women, with an incidence of approximately 0.001 cases per 1,000,000 [6]. Described in 1916 by the French cardiologist, René Lutembacher, the syndrome refers to the association between ASD and MS, which is commonly difficult to diagnose, since the presence of an ASD alters the hemodynamic and clinical characteristics of the MS and vice-versa [7]. Classically, Lutembacher syndrome is represented by ostium secundum ASD and MS of rheumatic etiology, although any association may be present. Vaaidesar et al., in an autopsy series of 44 cases of non-primum ASD with mitral valve dis-

ease, identified a rheumatic etiology in 46% of cases [8].

The clinical presentation varies according to the dimensions of the ASD and the severity of MS, with varying degrees of pulmonary hypertension, dilation of the right-sided chambers and tricuspid regurgitation. Due to the compliance of the right ventricle, blood flow is shunted from the left atrium to the right atrium, generating progressive dilation of the right chambers and reduced flow to the left ventricle. Therefore, as presented in this case report, the patient did not develop signs of pulmonary congestion, a classic finding in isolated MS [1]. Therefore, symptoms such as orthopnea, paroxysmal nocturnal dyspnea, hemoptysis and pulmonary edema are replaced by fatigue due to reduced left ventricular filling and low cardiac output, in addition to systemic congestion secondary to right ventricular dysfunction [9].

Our patient presented with superior vena cava-type ASD with an anomalous connection of the right upper pulmonary vein, a rarely described association [3, 4]. Partial anomalous connection of the pulmonary vein into the superior vena cava resulted in an additional increase in pulmonary blood flow, generating a greater overload of the right-sided chambers. Severe dilation of the right cardiac chambers and a non-significantly enlarged left atrium, despite significant MS, represented preoperative signs of a structural disease coexisting with a mitral valve disease, not seen on TTE.

Treatment is classically surgical, and percutaneous interventions might be considered in cases with favorable anatomy of the mitral valve and atrial septum, using mitral percutaneous balloon valvuloplasty associated with an ASD occluding prosthesis [10]. Early diagnosis and treatment can lead to favorable prognosis. However, when the clinical presentation includes advanced pulmonary hypertension and heart failure, such as in Eisenmenger syndrome, the prognosis is worse [11].

Despite the rare combination presented in this case report, it highlights the importance of thorough and detailed imaging examinations in patients presenting with Lutembacher syndrome.

**Disclosure of conflict of interest**

None.

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**References**

- [1] Budhwani N, Anis A, Nichols K and Saric M. Echocardiographic assessment of left and right heart hemodynamics in a patient with Lutembacher's syndrome. *Heart Lung* 2004; 33: 50-4.
- [2] Varsha AV, George G and Sahajanandan R. Lutembacher syndrome: dilemma of doing a tricuspid annuloplasty. *Ann Card Anaesth* 2017; 20: 456-458.
- [3] Anwar AM, Nosir YF, Ajam A, Galal AN, Ashmeg A and Chamsi-Pasha H. Partial anomalous pulmonary venous connection associated with Lutembacher's syndrome. *Echocardiography* 2008; 25: 436-439.
- [4] Shende S, Inamdar A and Inamdar S. Sinus venosus atrial septal defect with severe mitral stenosis: a rare presentation. *Med J D Y Patil Vydiapeth*; 8: 392-394.
- [5] Fradi El Faleh I, Ezzar MT, Zaroui A, Boussaada R and Mechmeche R. Percutaneous mitral commissurotomy associated to sinus venosus atrial septal defect and partially anomalous pulmonary venous connection: a case report. *Ann Cardiol Angeiol (Paris)* 2014; 63: 95-98.
- [6] Ali SY, Rahman M, Islam M, Barman RC, Ali MY and Mmsu I. Lutembacher's syndrome-a case report. *Faridpur Med Coll J* 2011; 6: 59-60.
- [7] Lutembacher R. De la sténose mitrale avec communication interauriculaire. *Arch Mal Coeur*; 237-260.
- [8] Vaideeswar P and Marathe S. Lutembacher's syndrome: is the mitral pathology always rheumatic? *Indian Heart J* 2017; 69: 20-23.
- [9] Olivares-Reyes A and Al-Kamme A. Lutembacher's syndrome with small atrial septal defect diagnosed by transthoracic and transesophageal echocardiography that underwent mitral valve replacement. *J Am Soc Echocardiogr* 2005; 18: 1105.
- [10] Cheng TO. Coexistent atrial septal defect and mitral stenosis (Lutembacher syndrome): an ideal combination for percutaneous treatment. *Catheter Cardiovasc Interv* 1999; 48: 205-6.
- [11] Aminde LN, Dzudie A, Takah NF, Ngu KB, Sliwa K and Kengne AP. Current diagnostic and treatment strategies for Lutembacher syndrome: the pivotal role of echocardiography. *Cardiovasc Diagn Ther* 2015; 5: 122-132.