CASE REPORT

# A RARE ENCOUNTER; DIAGNOSING LUTEMBACHER SYNDROME IN AN ADULT PATIENT AND LITERATURE REVIEW ADDRESSING IT'S CLINICAL INSIGHT

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**ABSTRACT:**

**BACKGROUND:** *A uncommon cardiovascular condition called Lutembacher syndrome consists of an atrial septal defect combined with mitral stenosis. The most well-known form combines congenital atrial septal defect with acquired mitral stenosis. In some people, the signs and symptoms of mitral stenosis may be mitigated or delayed because the atrial septal defect functions as a pressure-relieving gateway.*

*This case report details the case of a 45-year-old woman who was identified as having Lutembacher Syndrome and was experiencing worsening dyspnea and palpitations. Following an open surgical procedure, a significant secundum ASD and severe MS were effectively treated, according to the diagnostic evaluation. The scenario illustrates the difficulties in diagnosing this complicated ailment, the value of early diagnosis, and different therapeutic modalities.*

## INTRODUCTION:

Any congenital or iatrogenic atrial septal defect (ASD) combined with congenital or acquired mitral stenosis (MS) is known as Lutembacher syndrome (LS), an uncommon cardiovascular condition[1]. As previously said, it is an uncommon occurrence; in fact, the American Heart Journal reports that there is only one person in every billion worldwide who has this particular combination of illness[2]. Congenital MS is an uncommon condition that accounts for just 0.6% of congenital heart disease, whereas those with mitral stenosis have a 0.6% to 0.7% probability of having congenital ASD[3-5]. Because of this, congenital ASD with acquired MS as a result of rheumatic heart disease is a prevalent type of Lutembacher syndrome. Assuming a consistent prevalence of ASD, the incidence of LS is more common globally in areas with higher rates of rheumatic mitral stenosis, such as Southeast Asia and sub-Saharan Africa. However, this combination is uncommon in the west, where rheumatic mitral stenosis is less common. Rene Lutembacher, a French physician, initially provided a thorough description of these two abnormalities in a 61-year-old lady in 1916.[6]

The symptoms of Lutembacher Syndrome can manifest in a variety of ways. Individuals may have symptoms that range from mild dyspnea upon exertion to indications of pulmonary hypertension, right heart failure, and paradoxical embolism. The key to making a diagnosis is echocardiography, which gives vital information on MS hemodynamics, shunt severity, and ASD size[7]. Further imaging modalities to test pulmonary vascular resistance and further characterize anatomy may include cardiac catheterization or cardiac MRI.

A multidisciplinary approach is used in management techniques, which are customized based on the symptoms and hemodynamic condition of each patient. The goal of medical therapy is to control

related comorbidities and lessen the symptoms of heart failure. When necessary, the best course of action is a surgical or percutaneous intervention.

This paper provides a thorough case study of a 45-year-old woman who was eventually diagnosed with Lutembacher Syndrome after presenting with worsening dyspnea and palpitations.

**CASE REPORT:**

A 45-year-old woman with history of dyspnea and palpitations for six-months presented in cardiac emergency with complaint of worsening with exertion. She acknowledged having recurring respiratory illnesses as a child, but she denied having ever had a heart condition or having any serious medical conditions.

Upon evaluation, she did not have fever. Her pulse rate was 86 beats per minute. The blood pressure measured was 110/70 mm Hg, the respiratory rate was 20 breaths per minute, and the pulse rhythm was regular with sufficient volume. She had bipedal oedema and was pale. While examining precordium, apex beat was detected in left 5th intercostal space lateral to midclavicular line. Patient had palpable P2 accompanied by left parasternal heave but no palpable thrill was found on

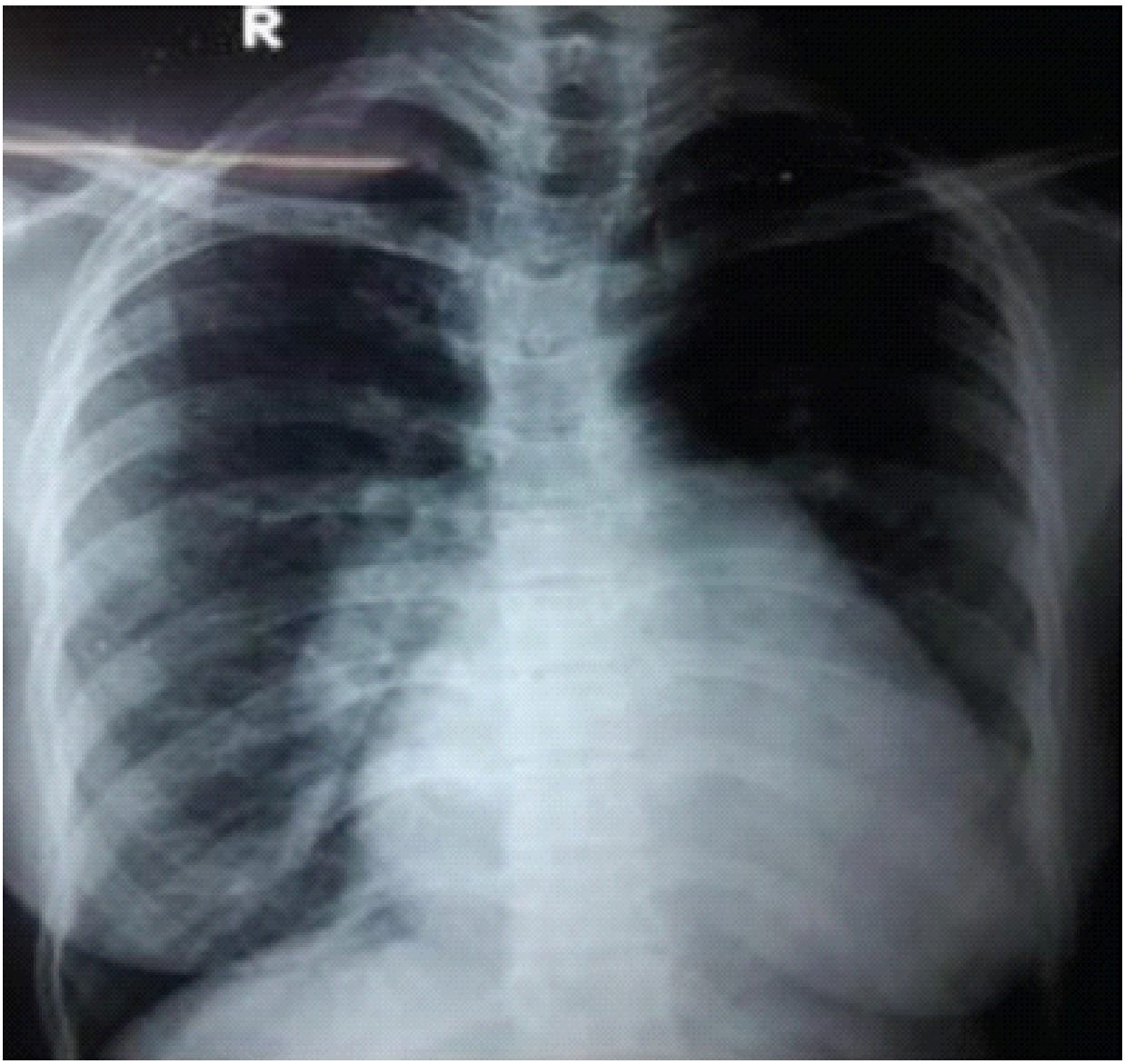


**Fig 1.** Chest X-ray showed cardiomegaly with RV type apex, dilated right pulmonary artery.

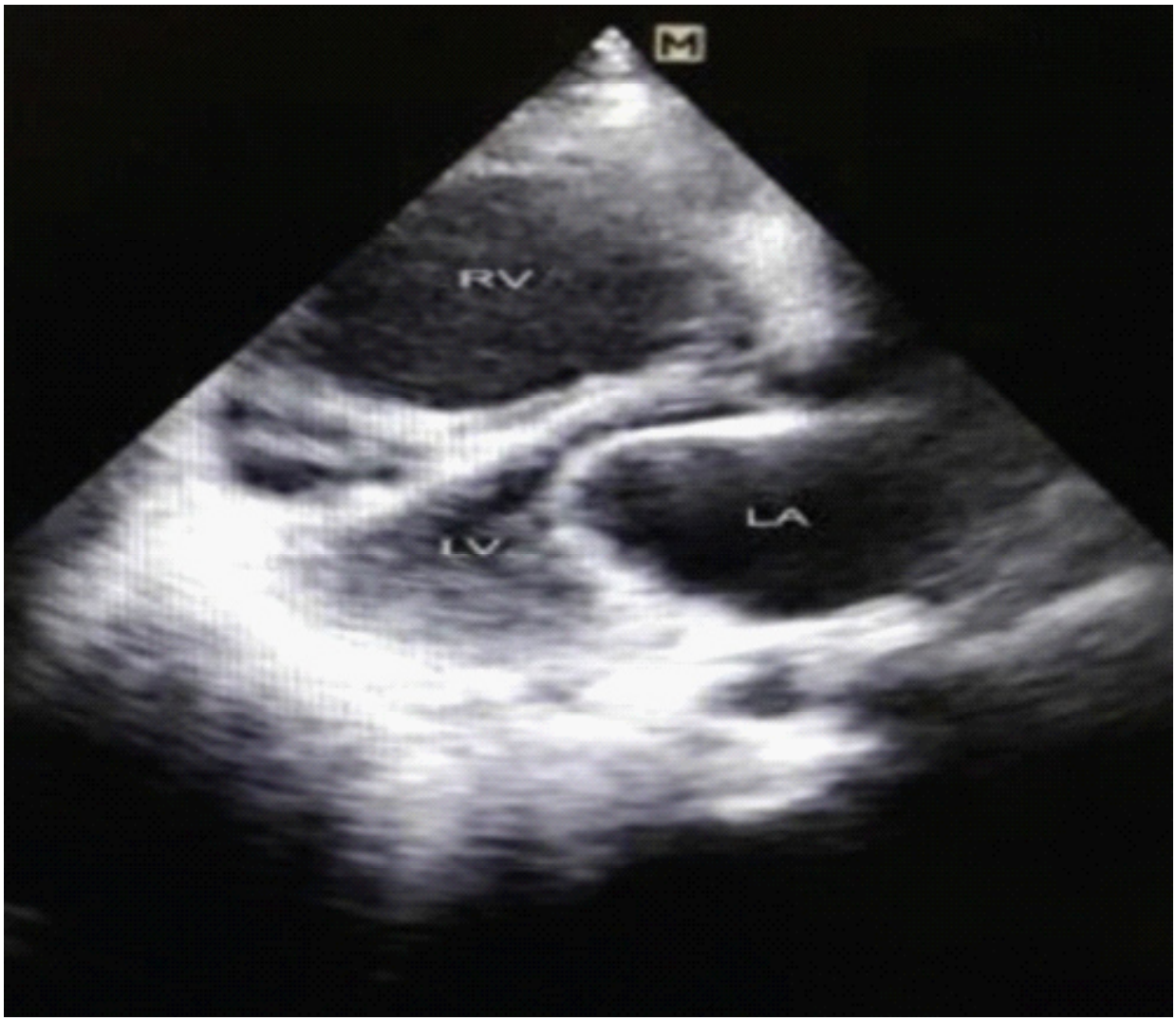


**FIG 2. Mitral Stenosis, dilated left atrium (LA) and right ventricle (RV)**

examination. On left auscultaltion, there was loud S1, and wide fixed splitting of pulmonic component of S2 with opening snap in mitral area. There was grade III mid diastolic murmur in mitral area and early systolic murmur in pulmonary area and a continuous murmur along lower left parasternal border. Respiratory system examination showed bilateral equal air entry and end-inspiratory fine crackles in bilateral lower lung fields. Abdominal examination showed no hepatomegaly or ascites.

Her chest X-ray showed cardiomegaly with RV type apex, engorged pulmonary conus, and dilated right pulmonary artery upon further investigation of the case. (Figure - 1)

Her echocardiography, which was performed later, showed that her right atrium (RA) and right ventricle (RV) were substantially dilated and significant stenosis of mitral valve. (Figure-2)

Mitral valve area (MVA) was 0.8 $cm^2$ on measurement via mitral valve planimetry and it showed thickened calcified mitral valve and subvalvular changes and the mean pressure gradient across the mitral valve was 9 mmHg. (Figure-3)

An atrial septal defect was present, with measurements of 13.43 mm in the subcostal view and 17.38 mm in the apical 4 chamber view. RV was dilated in the apical four chamber view, with a base diameter of 60 mm. (Figure-4)

Transesophageal echocardiography

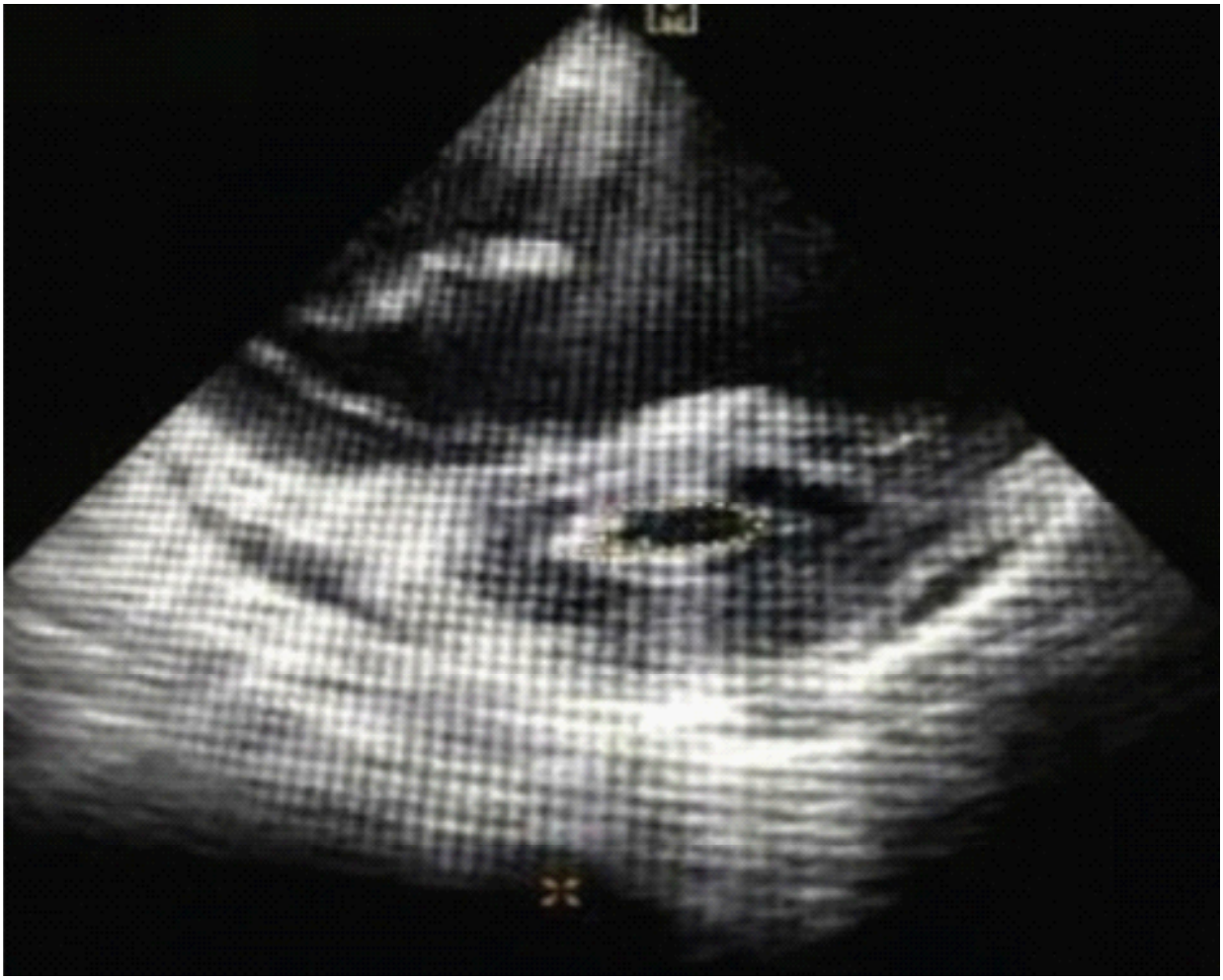

**Fig 3. Showed thickened calcified mitral valve**

(TEE) revealed limited leaflet movement and calcification compatible with severe MS, as well as confirming the existence of a secundum ASD measuring 20 mm in diameter. TEE offered a thorough morphological and hemodynamic evaluation.

Thus, in addition to receiving therapy for her presenting sickness, the patient received appropriate counseling of her medical status and was administered prophylaxis for rheumatic heart disease, low dose diuretics, and beta blockers. She eventually had her mitral valve replaced with a metallic prosthetic valve and had her ASD undergone patch repair.

Following the surgery, the patient's dyspnea and edema resolved, resulting in symptomatic alleviation. Three months later, a follow-up echocardiogram revealed no residual left-to-right shunt and a prosthetic valve operating well. The patient is still under routine observation for routine assessment of valve dysfunction, recurrent shunt, or other problems.

**DISCUSSION:**

One of the uncommon cardiovascular syndromes, lutembacher's syndrome, typically manifests as acquired mitral stenosis with congenital ASD[2]. This syndrome is more common in the female population, as ASD and MS are more common in female patients[8]. Although there have been multiple reports of familial cases of Lutembacher's syndrome, these have only been individual instances.[9]

The initial idea was that an atrial septal defect or left-to-right shunt was caused by the patent foramen ovale being stretched open by high LA pressure brought on by

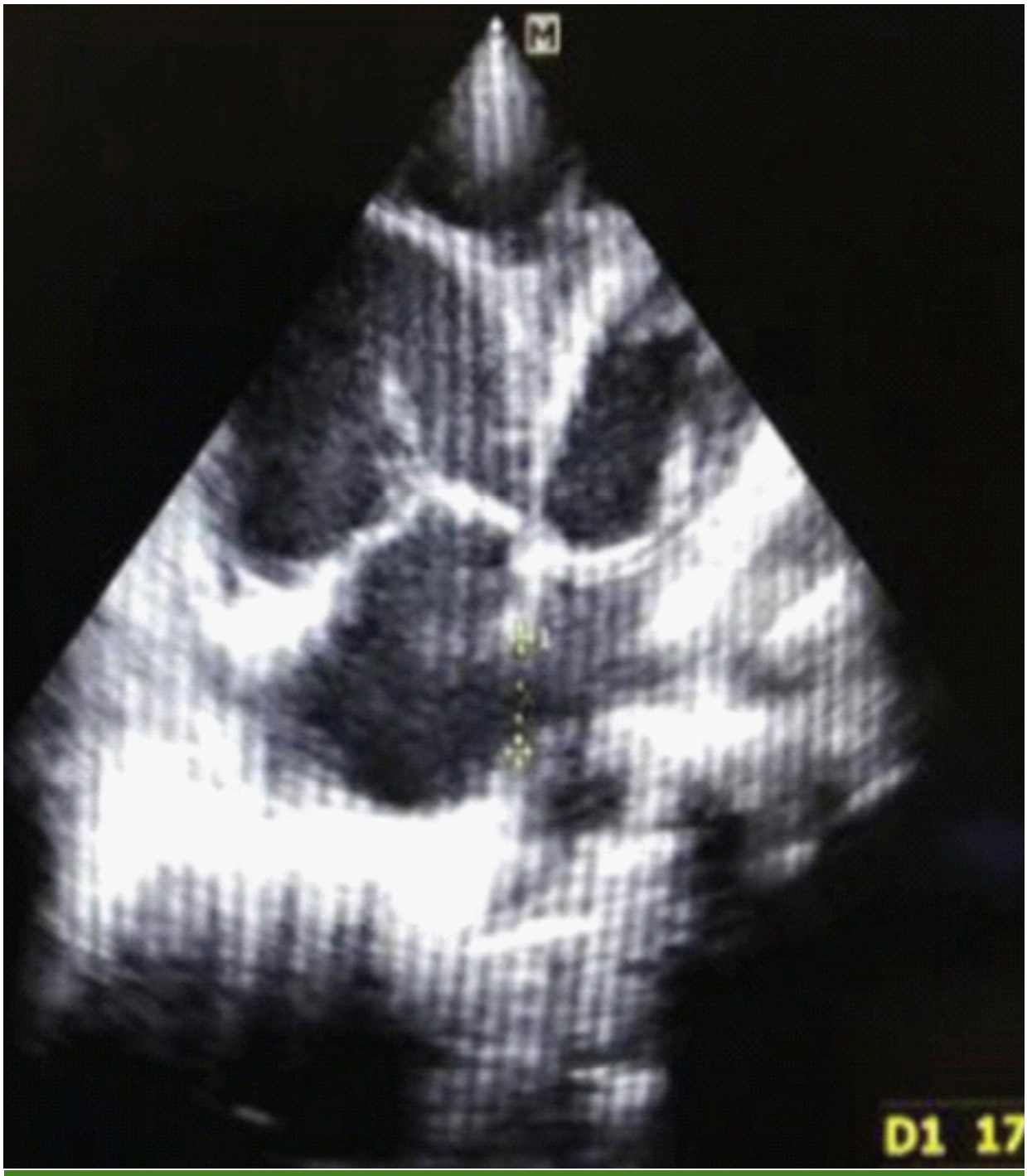


**Fig 4. Figure showed the atrial septal defect**

mitral stenosis. However, according to recent directions available, ASD may be congenital or acquired[10]. Some observations by Ananthasubramaniam et al. in 2001, they described a case of mitral valve replacement that further supported the idea that high LA pressure causes ASD. There was no longer any discernible left-to-right shunt following the valve replacement[11].

Because MS and ASD coexist, the interaction of multiple factors, including the size of ASD, the severity of MS, and the distensibility of RV, affects hemodynamic expression and clinical characteristics. As

a result of blood shunting from LA to RA, increased LA pressure resulting from mitral stenosis in Lutembacher syndrome finds ASD as a second exit for LA blood. The size of ASD can control this blood shunt, which in turn mitigates the harmful effects of rising LA pressure. If there is no ASD, blood finds its way to the pulmonary veins as a result of elevated atrial pressure brought on by MS, which more frequently and quickly results in pulmonary congestion. ASD minimizes congestion and back pressure to the pulmonary vasculature in patients with Lutembacher syndrome. Similarly, symptoms of lung congestion are either delayed or lessened. These include hemoptysis, pulmonary edema, orthopnea, and paroxysmal nocturnal dyspnea. On the other hand, left to right shunt precipitates right heart overflow and the occurrence of dilatation of right sided heart chambers and pulmonary hypertension and right heart failure[12].

The physical examination of a patient with Lutembacher syndrome is significantly impacted by hemodynamic decompression of LA pressure accumulation caused by mitral stenosis. Pure MS clinical examination findings are either missing or very weak. Mid-diastolic murmur with presystolic accentuation, the opening snap, and the loud initial heart sound are usually absent. The high LA to low RA pressure difference through ASD that lasts the whole cycle typically results in a persistent murmur. Increased pulmonary blood flow over the pulmonary valve results in a loud ejection systolic murmur across the pulmonary region as more blood is shunted, in contrast to isolated ASD. Accompanying the condition might be a holosystolic murmur over the left lower sternal border, and pulmonary hypertension could result in a regurgitant tricuspid valve.

When it comes to verifying the diagnosis and determining the extent of ASD and MS, echocardiography is essential. Transesophageal echocardiography (TEE) and cardiac catheterization are examples of advanced imaging procedures that offer comprehensive anatomical and hemodynamic data that are essential for therapy planning.

A customized approach is required for the care of Lutembacher Syndrome, taking into account the degree of MS, the size and location of ASD, and the severity of symptoms. The goal of medical therapy for heart failure symptoms is to maximize circulatory function by lowering preload and afterload. In order to stop left-to-right shunting and avoid consequences like paradoxical embolism and pulmonary hypertension, the ASD can be surgically or percutaneously closed. In situations of severe stenosis, interventional MS techniques like as balloon valvuloplasty or surgical valve repair/replacement may be considered. Minimal invasive percutaneous procedures are preferred over open surgical repair for management of lutembacher syndrome[13].

Even while ASD lessens the severity of MS symptoms, more blood is shifted to the right side of the heart as a result of mitral valve stenosis, which increases blood flow through ASD and increases the risk of atrial fibrillation and right ventricular failure. These individuals have a dismal prognosis when they develop heart failure and pulmonary hypertension at the late advanced stage. If diagnosis is significantly delayed and severe pulmonary hypertension has developed, in such a case, conservative management including alleviating symptoms, managing heart failure and prophylaxis for bacterial endocarditis may be considered as management modality[14,15]. However, if indicated then resolving this dual issue with surgical closure of ASD and replacement of the mitral valve has a favorable prognosis and increases life expectancy. Lately, percutaneous transluminal mitral commissurotomy (PTMC) and closure of ASD with the Amplatzer device are commonly considered feasible interventional techniques[16]. These are basically minimal invasive interventional modalities that offer adequate alternative of extensive invasive surgical procedures and to avoid their relevant complications[17].

It is necessary to do further study to investigate new treatment approaches, improve diagnostic techniques, and clarify the long-term effects of various therapeutic measures in patients with

Lutembacher syndrome. To further enhance our knowledge of this uncommon but clinically relevant illness and to improve clinical outcomes for patients, collaboration between doctors, researchers, and industry stakeholders is essential.

**CONFLICT OF INTEREST:**

There is nothing to disclose.

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