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Case Report

A Rare Association: Lutembacher Syndrome and Coarctation of Aorta

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Abstract

Lutembacher syndrome is defined by a congenital ostium secundum atrial septal defect associated to an acquired mitral valve stenosis. We present a similar case in a 24 year old female who came in with progressive shortness of breath. Examination revealed an elevated jugular vein distention. The first heart sound is loud in all four cardiac areas, a wide fixed split of second heart sound on inspiration and expiration and a grade 2/6 systolic murmur at of tricuspid regurgitation. Chest x-ray showed signs of pulmonary edema with bat wing pattern and cardiomegaly. Electrocardiography (ECG) revealed an electrical axis shifted to the right and right bundle branch block along and Transthoracic Echocardiography (TTE) showed a mitral stenosis with calcification and moderate tricuspid regurgitation, dilated cardiac chambers and coarctation of aorta. The patient was consequently treated with symptomatic drugs contain beta-blockers and diuretics and finally a scheduled for valvular and septal repair via open heart surgery. The purpose of this case report is to assist cardiologists in diagnosing this syndrome accurately on the basis of symptoms and investigations.

Keywords: *Atrial Septal Defect; Lutembacher Syndrome; Mitral Stenosis; Transthoracic Echocardiography*

Introduction

Lutembacher syndrome (LS) was first described in a letter by anatomist Johann Friedrich Meckel in 1750 [1,2]. Corvisart who later described the association of atrial septal defect (ASD) and mitral stenosis (MS) in 1811. However, the first comprehensive account of these two defects was reported by a French physician Rene Lutembacher in 1916, after whom this syndrome was eventually named. The first case of this syndrome was described in a 61-year-old woman with mitral valvular lesion associated to congenital mitral stenosis (MS). The definition of LS has changed many times since then. Opinion differs regarding what lesions the syndrome should include. Although defined as MS in combination with ASD, some authors also classify ASD with mitral regurgitation (MR) as a part of the LS spectrum. However, the current consensus defines LS as any combination of ASD (congenital or iatrogenic) and MS (congenital or acquired).

The exact prevalence of LS is not known [3]. It is more likely to prevalent in areas with higher prevalence of rheumatic heart disease (RHD). For this reason, it is reported more often in Southeast Asia. Furthermore, in such developing countries, 40% of patients with LS, had an history of rheumatic fever. The syndrome is usually more common in young adults but can present at any age. And there is a predilection for females more than men because ASD and rheumatic MS are both more prevalent in females.

Case Presentation

A 24 year-old female presented to Department of Cardiovascular Surgery of Ibn Rushd Hospital university, Mars 2019 with a 8 months history of progressive shortness of breath on minimal exertion, and awakening at night, palpitation and fatigue, which had worsened since the last few days. She also complained of orthopnoea since last 4weeks. She felt comfortable at rest but she report a shortness of breath during exercise or when lying flag. She had a history of tonsillitis repeatedly during childhood. There was no history of paroxysmal nocturnal dyspnoea or haemoptysis.

On physical examination she was afebrile with a rapid and irregular heartbeat with pulse of 95 beats/min. Her blood pressure was 110/70 mm of Hg and respiratory rate was 26 breaths/min. On precordial examination showed a signs of right sided heart failure with an elevated jugular vein distention and fluid retention in legs. The first heart sound is loud in all four cardiac areas, a wide fixed split of second heart sound on inspiration and expiration and a grade 2/6 systolic murmur at of tricuspid regurgitation. Crackles were heard on auscultation at the base of right lung. There was no evidence of hepatosplenomegaly and ascites.

A chest X ray showed signs of pulmonary edema with bat wing pattern and cardiomegaly.

Electrocardiogram (ECG) showed an electrical axis shifted to the right, right bundle branch block along and atrial fibrillation.

Transthoracic Echocardiography (TTE) which showed severe mitral stenosis (mitral valve area calculated by planimetry was 1,4 cm^2, 1,48 cm^2 by pressure half time) (Figure 1), the peak pressure gradient was 16 mmHg and mean pressure gradient was 8 mmHg (Figure 1) with thickened and very calcified leaflets. There was a small congenital atrial septal defect of the ostium secundum variety measuring 6 mm (Figure 2) with dilated right atrium (Right atrial area at 22 cm^2), an enlarged right ventricle with volume overload. There was also a dilated left atrium of 45 mL/m^2 (Figure 3), the aortic valve was thickened but without an aortic stenosis or regurgitation. Left ventricle had a normal size with an ejection fraction of 62%.

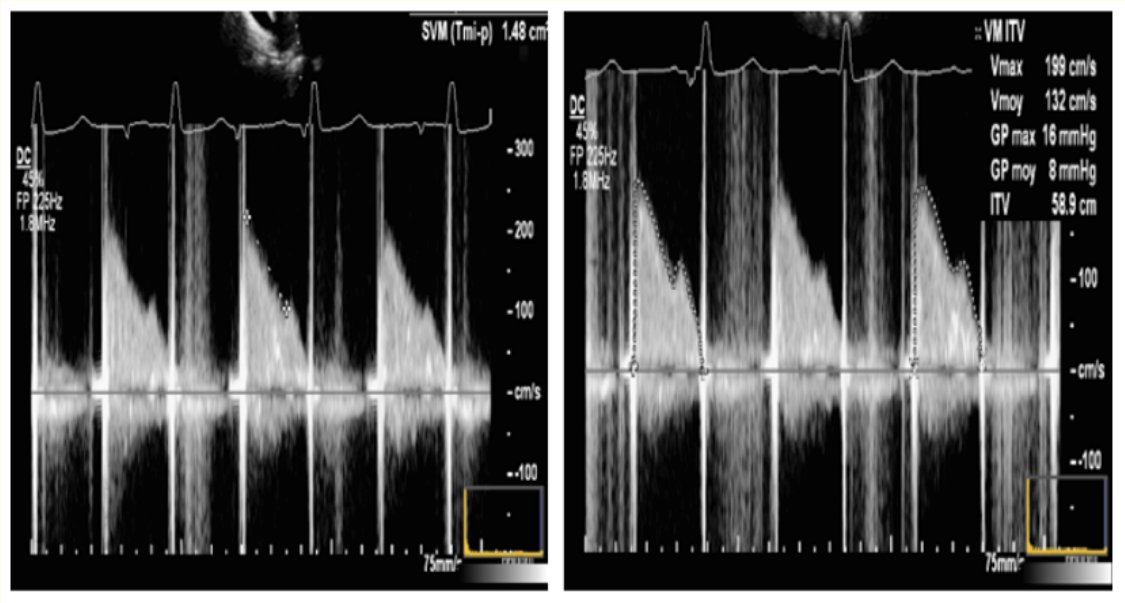

Figure 1: *Spectral Doppler envelope of mitral valve inflow shows calculated pressure half-time mitral valve area (MVA) of 1,48 cm^2 consistent with severe MS, peak pressure gradient (PPG) was 16 mmHg and mean pressure gradient (MPG) was 8 mmHg.*

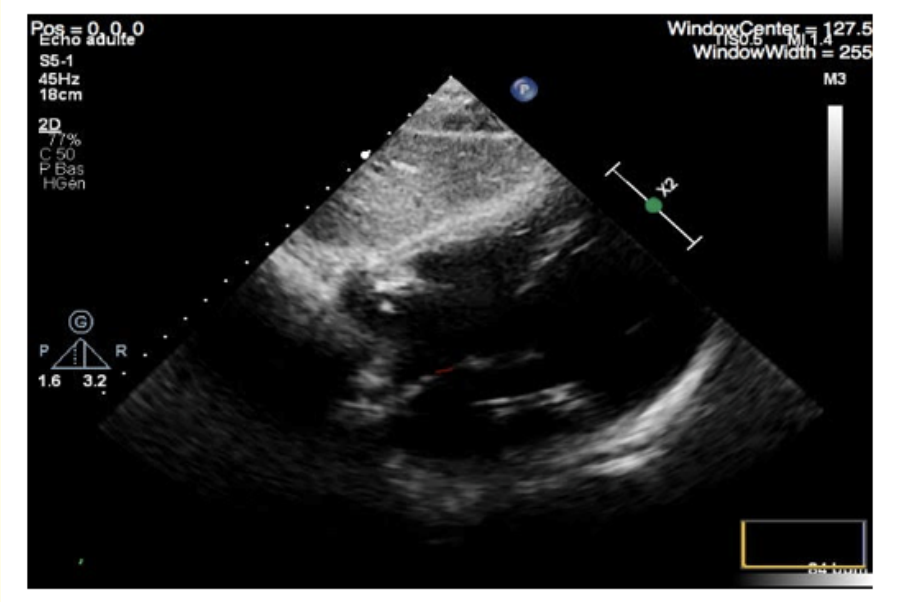

Figure 2: *Subcostal view at level of atria shows small left-to-right shunt (arrow) through ostium secundum septal.*

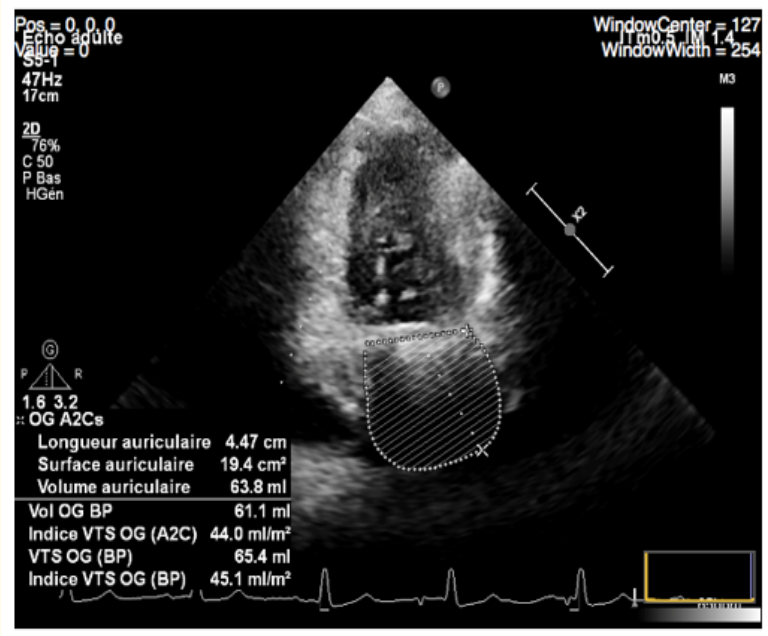

Figure 3: *Left atrial volume on apical tow chamber.*

The Qp/Qs flow ratio was calculated at 1,2/1,0.

The 2D transthoracic echocardiography suprasternal section shows a severe coarctation of aorta; the Doppler exam reveals a continuous flow with maximum of the velocity of 4,32 m/s (Figure 4).

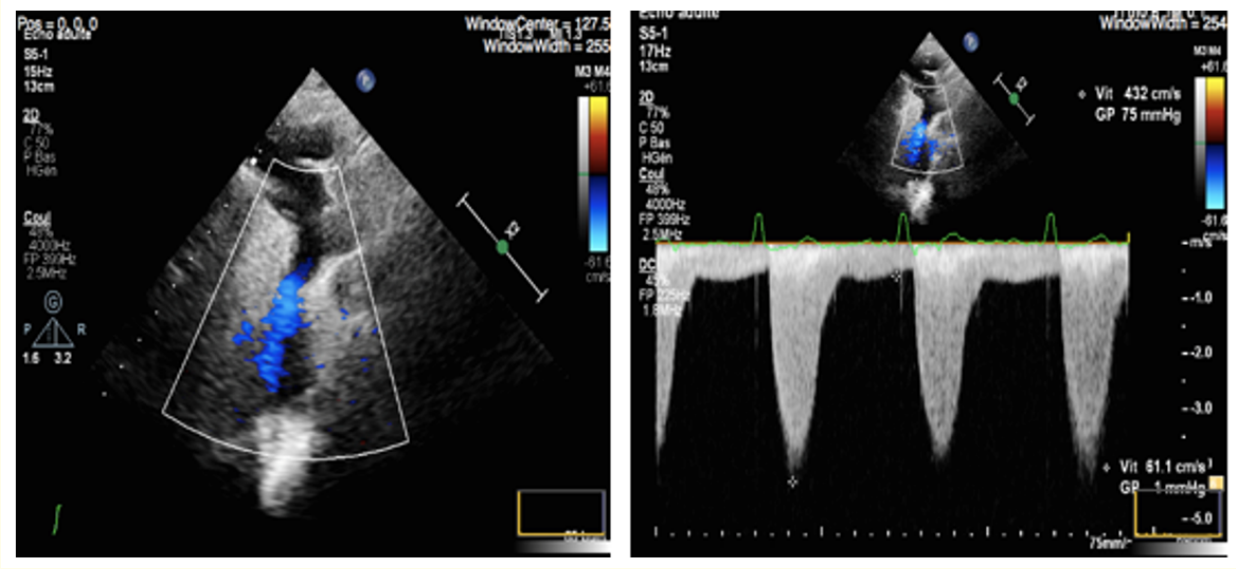

Figure 4: *Transthoracic echocardiography suprasternal section showes a severe coarctation of aorta; the Doppler exam reveals a continuous flow with maximum of the velocity of 4,32 m/s.*

On the basis of the above investigations, a diagnosis of Lutembacher's syndrome associated to severe coarctation of aorta was made. The patient was administered metoprolol, furosemide and scheduled for open heart surgery at a later date with stenting was the preferred approach. A Willkin's score of 10 and association to another congenital disease suggested that percutaneous transcatheter therapy was an unfavourable option in this scenario.

Written consent was understood and taken from the patient prior to the reporting of the case.

Discussion

The hemodynamic effects of this syndrome are the result of the interdependence between the relative effects of atrial septal defect and mitral stenosis. The hemodynamic features and the natural history of the patients depend upon the size of the ASD, severity of MS, compliance of the right ventricle and the degree of pulmonary vascular resistance. When MS is severe, and ASD is nonrestrictive, left atrium (LA) finds another exit through the septum in addition to the mitral valve (LA decompression). Therefore, LA pressure does not rise in proportion to the severity of MS. For this reason, pulmonary venous hypertension takes a long time to develop. However, this results in an increased left to right shunt across the ASD and progressive dilatation of right atrium (RA) and right ventricle (RV) with increased pulmonary blood flow. In untreated cases, the pulmonary vascular resistance continues to increase which eventually leads to right ventricular failure. The pulmonary artery hypertension (PAH) in these patients is usually hyperkinetic (because of increased left to right shunt) in comparison to patients with isolated severe MS where it develops due to direct back pressure transmission, reactive pulmonary vasoconstriction and obliterative changes in pulmonary arterioles. There occurs a reciprocal decrease in the left ventricular filling and

stroke volume. In contrast, if the ASD is restrictive, the shunt across the defect will be less, and hence, the patient will follow the course of isolated MS.

In contrast to an isolated ASD, the susceptibility to infective endocarditis is increased by the presence of MS.

The calcification of mitral leaflets is less common in LS, as there occurs decompression of LA, resulting in less turbulent flow across the mitral valve. Atrial dilatation in LS predisposes the patients to develop atrial fibrillation or atrial flutter.

A rare clinical entity, Lutembacher's syndrome is a rare association of mitral stenosis and atrial septal defect. This a rare syndrome which is often isolated but may be associated with other congenital diseases.

Both of these cardiac defects (MS and ASD) can be either congenital or acquired. Mitral stenosis is generally acquired in this syndrome as a consequence of rheumatic heart disease as was in our case. Atrial septal defect can be congenital, as was in our case and can be iatrogenic, secondary to cardiac interventional procedures like mitral valvuloplasty.

There has been a female preponderance in Lutembacher's syndrome cases reported throughout the literature. The increased prevalence of ostium secundum atrial septal defects and rheumatic heart disease in females explain the predominance of this defect in women [5]. In the study by Bashi., *et al*. [5], cute rheumatic fever has been reported to be the chief causative factor for mitral stenosis (as a component of rheumatic heart disease) in developing countries as demonstrated.

The concurrent existence of Mitral Stenosis and Atrial septal defect gives rise to particular haemodynamic manifestations. The stenosed mitral valve block blood flow into the main pumping chamber of heart (left ventricle) and the ASD shunts trapped blood from the left atrium into the right atrium, causes a right chambers dilatation, preventing pulmonary congestion however at the cost of diminished left ventricular outflow. Pulmonary oedema usually does not develop until late in the disease since the right ventricle is easily distensible as compared to the left ventricle due to which the blood shunts through the ASD instead of backing up into the pulmonary veins [4].

The clinical presentation of Lutembacher's syndrome is mainly dependent on three variables: size of ASD, severity of MS and compliance of right ventricle [6]. The shunting of blood from the left to the right side of the heart leads to progressive right ventricular overload and right-sided heart failure. Since anterograde blood flow into the left ventricle is reduced, there is fatigue on ordinary physical exertion which is usually the presenting complaint of the patient. In general, unless the ASD and mitral stenosis causing Lutembacher's syndrome is severe, symptoms may not appear until the second and third decade of the patient's life.

Palpitations as a result of atrial arrhythmias, blood flowing from left atrium to the right atrium causing a higher left atrial pressure and leading to mitral stenosis. Both atria will be dilated leading to future atrial arrythmias or atrial fibrillation [7].

Our patient had signs of right ventricular dysfunction. She also complained of orthopnea and his chest x-ray suggested pulmonary vascular congestion, further indicating that the right ventricle's compliance has diminished considerably enough to reduce the amount of shunting via atrial septal defect.

Without any medical or surgical intervention, the left-to-right shunt could convert into a right-to-left one, causes Eisenmenger syndrome. Another instance of development of a right-to-left shunt is Reverse Lutembacher syndrome, in which an additional cardiac anomaly i.e. severe tricuspid stenosis or pulmonary stenosis precipitates a constellation of signs like central cyanosis, digital clubbing, and hepatomegaly [8].

Transesophageal Echocardiography (TEE) is more performant to analyse atrial septum compared to transthoracic echocardiography (TTE), but TEE could not be performed in our case because it was refused. Planimetry has been shown to give more accurate measure-

ments of the mitral valve area as compared to Doppler half-time which tends to overestimate the calculations, thereby giving a false impression of the severity of mitral stenosis [9].

Symptomatic treatment is done with diuretics to relieve the symptoms of right-sided heart failure or pulmonary venous congestion if present. Beta-blockers and calcium channel blockers are prescribed for rate control in atrial fibrillation. Digoxin is a good alternative if beta blockers are contraindicated. Prophylaxis for infective endocarditis is recommended.

In recent times, percutaneous trans-catheter therapy has gained preference over more invasive surgical procedures due to its faster recovery time and decreased length of hospital stay [10,11]. It was found that combined percutaneous treatment (including balloon valvuloplasty for MS and Amplatzer septal occluder for closure of the ASD) has improved the patient's planimetric mitral valve area to 2.1 cm (as compared to the previous 1.5 cm), maximum diastolic gradient to 9 mmHg (compared to previous 17 mmHg), and mean diastolic gradient to 4 mmHg (as compared to previous 9 mmHg). An important contraindication to percutaneous therapies and the reason why our patient could not undergo such procedures is the presence of calcifications with a wilkin's at 10. Other contraindications include presence of left atrial thrombi, inadequate rim tissue surrounding the atrial septal defect and anomalous pulmonary drainage [7].

Conclusion

LS remains a rare entity and echocardiography assessment is the current diagnostic modality of choice and further helpful in excluding co-existent cardiac pathologies. Planimetry by Doppler echo remains the best method for assessing MVA. Whilst open heart surgery is frequently the treatment modality of choice in case of co-existent cardiac malformations, With most of these conclusions drawn essentially from case reports, we propose prospective multicenter registries to evaluate different therapies and its long term outcome in patients with LS.

Consent

Informed consent was obtained from the patient to reproduce his case in this report.

Disclaimer

The abstract has not been presented or published in any journal or conference.

Conflict of Interest

None to declare.

Funding Disclosure

None to declare.

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