

Lutembacher syndrome with mitral valve calcification in a 31-year old male

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Abstract

Lutembacher syndrome is characterized by a congenital ostium secundum atrial septal defect and an acquired mitral valve stenosis. We present a similar case in a 31-year old male who came in with orthopnoea, central cyanosis and pedal oedema. Examination revealed cardiac murmurs in tricuspid and apical regions. Chest x-ray showed signs of pulmonary congestion and ventricular enlargement. Electrocardiography (ECG) revealed right axis deviation and right bundle branch block along with atrial fibrillation and Transthoracic Echocardiography (TTE) showed abnormal valves (mitral stenosis with calcification and tricuspid regurgitation) and dilated cardiac chambers. The patient was consequently treated with beta-blockers and diuretics and 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) is a rare cardiac clinical entity comprising of an unusual combination of atrial septal defect (ASD) and acquired Mitral valve stenosis (usually of the rheumatic nature).¹ LS is an infrequent disorder with a prevalence of 0.001 million per population, mostly occurring in females.^{2,3} The clinical presentation and prognosis of the disease varies depending on a multitude of factors; the most important one being the size of the defect while other factors include severity of stenosis and compliance of the right ventricle. Prognosis remains good if pulmonary hypertension and right sided heart failure does not develop. Surgical and percutaneous transcatheter therapies with balloon valvuloplasty and septal closure using an Amplatzer closure device have proven to be beneficial.⁴ Herein, we report a similar case of an adult male who was diagnosed with Lutembacher Syndrome

based on relevant clinical findings and investigations.

Case Report

A 31-year-old male presented to Cardiology Department of Civil Hospital, Karachi in June, 2016 with a 6-months history of progressive shortness of breath on minimal exertion, palpitation and fatigue, which had worsened since the last few days. He also complained of orthopnoea since last one month. He felt comfortable at rest but normal physical activity like walking aggravated his symptoms. There was no history of accompanying paroxysmal nocturnal dyspnoea, rheumatic fever or haemoptysis. On examination he was afebrile with a pulse of 111 beats/min (irregularly irregular). His blood pressure was 110/70 mm of Hg and respiratory rate was 24 breaths/min. He had cyanotic lips and tip of the tongue along with bilateral pedal oedema.

On precordial examination, there was a left parasternal heave along with a tapping impulse. Cardiovascular examination revealed an elevated jugular venous pulse (JVP). There was a loud S1 in all four cardiac areas and a wide fixed splitting of S2 on inspiration and expiration. It was associated with grade-IV pansystolic murmur which was heard at 5th intercostal space in tricuspid area and grade-IV mid-diastolic murmur which was heard at the

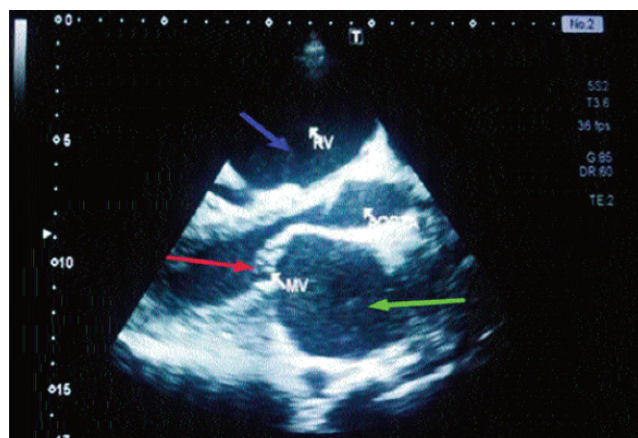


Figure-1: Parasternal long-view showing enlarged right ventricle (purple arrow), left atrium (green arrow) and calcified mitral valve (red arrow).

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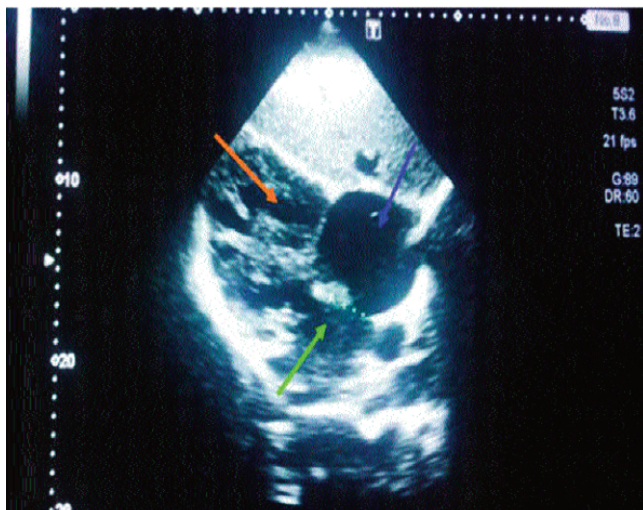


Figure-2: Subcostal view showing enlarged right ventricle (orange arrow), enlarged right atrium (purple arrow) and large atrial septal defect (green arrow).

apex. Crackles were also heard on auscultation at the base of right lung. There was no evidence of hepatosplenomegaly and ascites.

A chest radiograph showed cardiomegaly, pulmonary plethora and severe left atrium and right ventricular enlargement. Electrocardiogram (ECG) showed right axis deviation, right bundle branch block (RBBB), right ventricular hypertrophy and atrial fibrillation.

Transthoracic Echocardiography (TTE) which showed an enlarged right ventricle of 29 mm with volume overload, a dilated left atrium of 48mm, a dilated right atrium and a normal-sized left ventricle with an ejection fraction of 55%. There was very severe mitral stenosis (mitral valve area calculated by planimetry was 0.8 cm²) with thickened and calcified leaflets. Across the mitral valve, the peak pressure gradient (PPG) was 29 mmHg and mean pressure gradient (MPG) was 19mmHg. The aortic valve was thickened and there was a large congenital atrial septal defect of the ostium secundum variety measuring 25 mm. Colour flow mapping (Doppler) showed evidence of moderate tricuspid regurgitation and bidirectional shunt mostly left to right. Pulmonary arterial hypertension accompanied tricuspid regurgitation as indicated by pulmonary artery systolic pressure (PASP) which was estimated to be 50 mmHg (Figure-1 & 2). A Wilkin's score of 12 in our patient suggested that percutaneous transcatheter therapy was an unfavourable option in this scenario.

On the basis of the above investigations, a diagnosis of Lutembacher's syndrome was made. The patient was administered metoprolol, furosemide, amiloride and

warfarin and scheduled for open heart surgery at a later date.

Formal informed and written consent was taken from the patient prior to the reporting of the case.

Discussion

A rare clinical entity, Lutembacher's syndrome is a combination of mitral stenosis and atrial septal defect. Both of these cardiac defects can be either congenital or acquired. Mitral stenosis is generally 'acquired' in this syndrome as a consequence of rheumatic heart disease. Atrial septal defect can be congenital, as was in our case and can be iatrogenic, secondary to cardiac interventional procedures like mitral valvuloplasty.

Though the index case was a male, there has been a female preponderance in Lutembacher's syndrome cases reported throughout the literature. This female predominance can be explained by the increased prevalence of ostium secundum atrial septal defects and rheumatic heart disease in females.⁵ Acute 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 in the study by Bashi et al.⁶

The concurrent existence of MS and ASD gives rise to peculiar haemodynamic manifestations. The stenosed mitral valve hinders blood flow from the left atrium to the left ventricle and the ASD shunts 'trapped' blood from the left atrium into the right atrium, thereby 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.⁵

The clinical scenario of Lutembacher is mainly dependent on three variables: size of ASD, severity of MS and compliance of right ventricle.⁷ The shunting of blood from the left to the right side of the heart leads to progressive dilatation and ultimately failure of the right ventricle. 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. Palpitations as a result of atrial arrhythmias can also result due to distension of the left atrium, especially if mitral stenosis is severe.⁸

Our patient had elevated jugular venous pressure and moderate tricuspid regurgitation, both of which are indicative of right ventricular dysfunction. He 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 ASD.

Should the disease progress without any medical or surgical intervention, the left-to-right shunt could convert into a right-to-left one, namely 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 precipitates a constellation of signs like central cyanosis, digital clubbing, limb and facial oedema and tender hepatomegaly.⁹

Transesophageal Echocardiography (TEE) is a superior imaging modality to transthoracic echocardiography (TTE), but TEE could not be performed in our case because the patient did not consent to its use. Planimetry has been shown to give more accurate measurements 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.¹⁰

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. Mitral commissurotomy using Inoue-Balloon catheter for MS and Amplatzer atrial septal occluder for plugging ASD are the commonly employed treatment modalities in this respect. An important contraindication to percutaneous therapies and the reason why our patient could not undergo such procedures is the presence of bicommissural calcification. Other contraindications include presence of left atrial thrombi, inadequate rim tissue surrounding the atrial septal defect and anomalous pulmonary drainage.⁸

Conclusion

It is absolutely imperative that Lutembacher syndrome be diagnosed correctly via transthoracic and transesophageal echocardiograms in order to provide adequate medical and surgical therapies. Early diagnosis

will lead to prompt treatments, thereby delaying and possibly preventing the onset of pulmonary hypertension and heart failure, and consequently improve survival rates. Appropriate surgical procedures, be it transcatheter procedures or open heart surgeries should be opted for, after an overall assessment of disease progression and cardiac anatomy.

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

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