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Lutembacher Syndrome- A Case Report and Reappraisal

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Abstract

Lutembacher syndrome is defined as a combination of mitral stenosis (mostly rheumatic) with a left to right shunt at the atrial septal level. It is a rare disease with a higher prevalence in areas, where rheumatic heart disease is common. Diagnosis on physical examination can be challenging and therefore echocardiography is generally required to make the diagnosis. If not diagnosed and treated early, patient may develop right heart failure and arrhythmic complications, which bear a bad prognosis. We present a case-based reappraisal of Lutembacher syndrome in a 23-years old female, who presented with symptoms of pulmonary hypertension and heart failure.

Introduction

In medical literature the first description of Lutembacher syndrome was mentioned in a letter by anatomist Johann Fredrich Meckel to Albert von Hallerin in 1750 [1]. In 1811, Corvisart first described the Association of Atrial Septal Defect (ASD) and Mitral Stenosis (MS) [2]. Rene Lutembacher in 1916 published the first comprehensive account of these two defects. He described the first case in a 61-year-old woman and attributed the mitral valvular lesion to be congenital MS [2]. The definition of Lutembacher syndrome has undergone many changes but the current consensus is that Lutembacher syndrome consists of a congenital defect in the atrial septum together with an acquired mitral stenosis, most commonly of rheumatic origin [3]. In a typical Lutembacher syndrome the ASD is usually more than 1.5cm in size [4].

Case Report

A 23-years old female was admitted with history of recurrent lower respiratory tract infections, easy fatigability and dyspnea since childhood. Since past 2 weeks her symptoms had progressed and she complained of dyspnea at rest along with orthopnea. There were complaints of fever with mucopurulent sputum production and swelling bilateral lower limbs since 2 weeks. On examination, she was having tachycardia (pulse rate was 100/minute and regular) and her blood pressure was 110/56 mm Hg. She was tachypneic with a respiratory rate of 42 breaths per minute and oxygen saturation was 88 to 93% on room air. She had bilateral pitting pedal edema and Jugular Venous Pressure (JVP) was raised. There was no evidence of cyanosis on clinical examination. On precordial examination the apex beat was displaced laterally in the 5th intercostal space in anterior axillary line. There were prominent pulsations in the left parasternal area with a grade 3 parasternal heave. S1 was normal, wide splitting of the second heart sound with loud P2 was present. A long low pitch mid-diastolic rumbling murmur was heard in the mitral area along with a pansystolic murmur grade 3/6 along the left lower parasternal border. There was also, an early to mid-ejection systolic murmur grade 2/6, audible in the pulmonary area. Respiratory system examination revealed bilateral basal crepts and rhonchi. On abdominal examination, tender hepatomegaly was present and liver was palpable 5 centimeter below the right costal margin and shifting dullness was present .A provisional diagnosis of Rheumatic Heart Disease with severe mitral stenosis, moderate tricuspid regurgitation with severe pulmonary venous hypertension, severe pulmonary arterial hypertension, congestive cardiac failure (NYHA class 4) in sinus rhythm with lower respiratory tract infection was made. Lutembacher syndrome was also kept as a differential diagnosis. Investigations revealed hemoglobin of 11.4 g/dl, total leucocyte count of 12,200/mm3, erythrocyte sedimentation rate of 26 mm in 1st hour and a positive C-reactive protein. Her ASO titers were negative. Rest all laboratory investigations were normal. Her chest x ray revealed plethoric lung fields. There was evidence of cardiomegaly with a right ventricle type of apex. Right atrial enlargement and prominent pulmonary conus were also apparent. Her electrocardiogram showed peaked P waves, normal PR intervals, with right axis deviation of the QRS, and rSR complex in right chest leads. Transthoracic echocardiography revealed a large ostium secundum ASD of size

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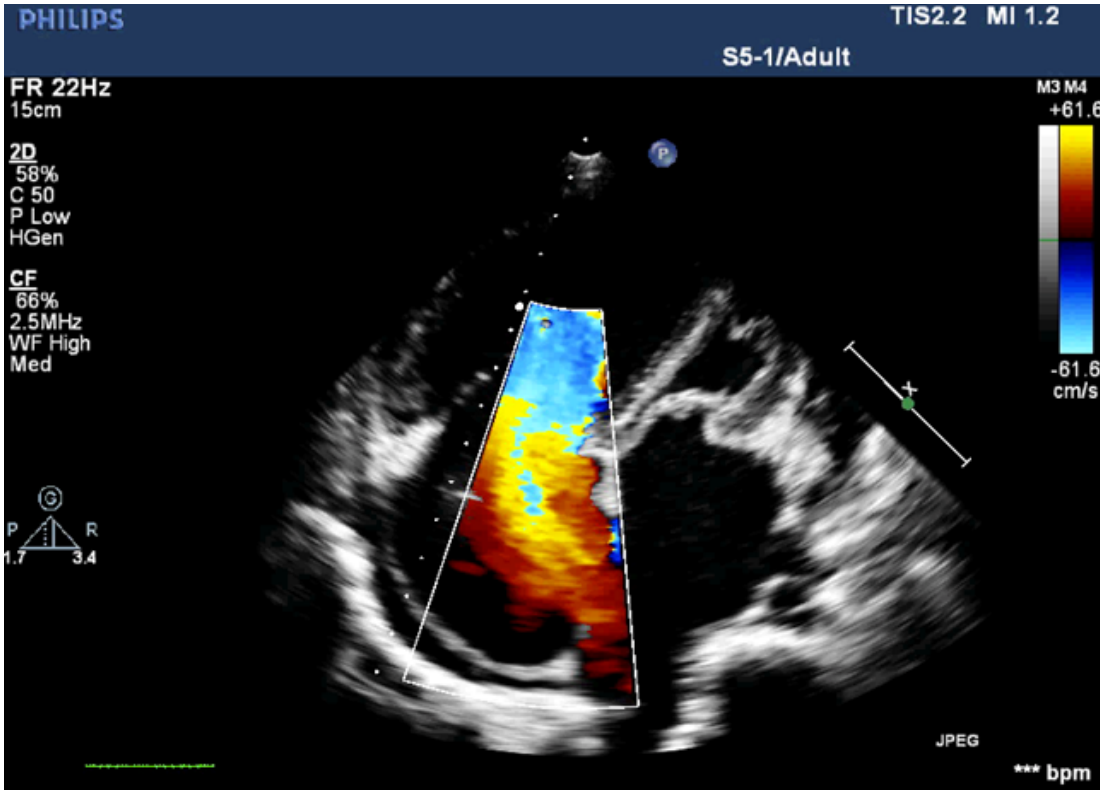

Figure 1: Transthoracic echocardiogram, apical-4-chamber view, showing a large atrial septal defect with left to right shunt. Also note the thickened mitral leaflets. Mild pericardial effusion is also seen.

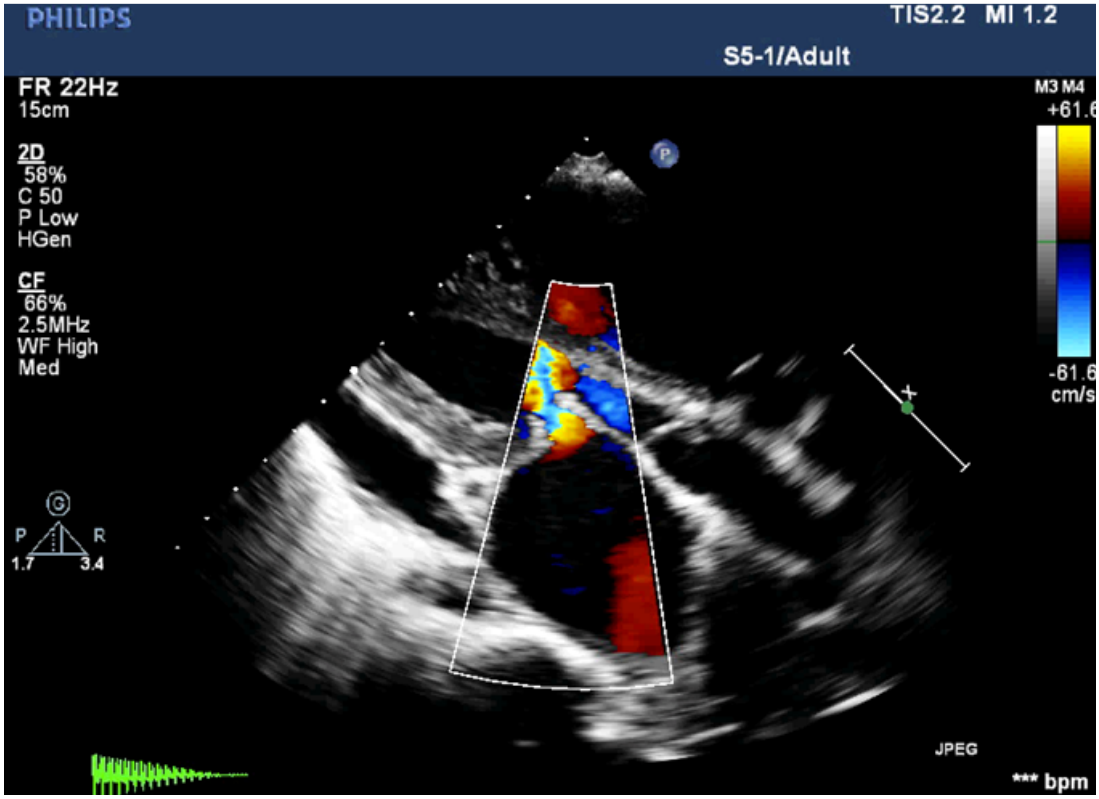

Figure 2: Transthoracic echocardiogram, Parasternal-long-axis view, showing a dilated left atrium with a turbulent jet across the mitral valve in diastole. Mitral leaflets are thickened and show restricted opening in diastole suggestive of mitral stenosis.

26 mm with left to right shunt and grossly dilated right and left atria and right ventricle. Moderate tricuspid regurgitation was present with a pressure gradient of 50 mm of Hg. Mitral valve leaflets were thickened with restricted movements and fusion of adjacent chordae. Thus the diagnosis of Lutembacher syndrome was confirmed. Patient denied any history suggestive of rheumatic fever. The patient was treated with oxygen inhalation, diuretics, beta-blockers, antibiotics and bronchodilators. Patient was advised cardiac catheterization followed by percutaneous transluminal mitral valve commissurotomy and ASD closure, but attendents refused due to financial constraints.

Discussion

Incidence: In patients with atrial septal defect, the incidence rate of mitral stenosis has been estimated at 4%, while the estimated incidence rate of atrial septal defect in patients of mitral stenosis is 0.6% to 0.7% [5]. The syndrome can present in any age but usually more common in young adults. There is predilection for females because ASD and rheumatic MS are both more prevalent in females [6]. The incidence rate of coexisting MS depends on the geographic prevalence of rheumatic fever. In the developing countries, history of rheumatic fever has been reported in 40% of patients with Lutembacher syndrome [7].

Pathophysiology: The natural history and hemodynamic features of patients with Lutembacher Syndrome may vary and depend on mitral stenosis severity, ASD size, pulmonary vascular resistance and the RV compliance. The most important clinical consequence of a nonrestrictive atrial septal defect in Lutembacher syndrome is its ameliorating effect on the symptoms of mitral stenosis as most of the blood entering the left atrium will be shunted across ASD leading to decompression of the left atrium thus ameliorating the signs and symptoms of pulmonary congestion [3]. Thus, orthopnea, paroxysmal nocturnal dyspnea, hemoptysis and pulmonary edema are infrequent and replaced by fatigue from reduced left ventricular filling and low cardiac output [4]. When atrial septal defect restrictive and mitral stenosis is severe the symptoms and clinical course resemble isolated mitral stenosis of equivalent severity [4]. Susceptibility to infective endocarditis increases in Lutembacher syndrome compared to ASD alone because of increased turbulent flow across the mitral valve.

Clinical Presentation: Patients may remain asymptomatic for years and symptoms mainly depend on the predominance and severity of mitral stenosis and size of ASD [4]. In patients with non restrictive ASD symptoms of pulmonary congestion due to isolated MS do not appear until late in the course of disease but once the pulmonary hypertension and right heart failure supervene like in our patient, the symptoms of progressive ankle edema, ascites and right upper quadrant pain develop [8]. There is elevated JVP in the absence of right ventricular failure and elevated jugular 'a' wave in the absence of pulmonary hypertension [4]. In non-restrictive ASD with MS prominent parasternal heave is present in comparison to restrictive ASD as MS augments left to right shunt. The under filled left ventricle cannot be palpated. The ausculatory findings of MS are attenuated as in the non-restrictive ASD, the left to right atrium decompression across ASD causes decreased flow across the stenotic mitral valve [3]. Secondly the volume overloaded right ventricle forms the apex, the signs of left ventricular inflow obstruction are less audible. When the atrial septal defect is restrictive, the electrocardiogram resembles that of mitral stenosis [4, 8]. However, in restrictive ASD, the signs and symptoms of pulmonary congestion develop depending on the severity of MS. When the atrial septal defect is nonrestrictive, the electrocardiogram resembles that of an atrial septal defect [3]. On chest X-ray, restrictive atrial septal defect results in an x-ray that resembles mitral stenosis with pulmonary venous congestion and left atrial enlargement [9]. When the atrial septal defect is nonrestrictive, the x-ray shows increased pulmonary arterial blood flow but little or no pulmonary venous congestion because the left atrium is decompressed. Echocardiography is used to establish the diagnosis of Lutembacher syndrome, identify the size and location of the ASD and determine the severity of MS [10].

Treatment: Management includes symptomatic treatment in the form of diuretics for pulmonary congestion or right heart failure, digoxin or beta-blockers for heart rate control. Definitive treatment is in the form of either percutaneous or surgical procedures [11].

Conclusion

Lutembacher syndrome is an unusual clinical entity of congenital ASD in combination with congenital/acquired MS (most commonly rheumatic). The presence of ASD alters the haemodynamics and thus the clinical course of Lutembacher syndrome as compared to isolated MS. It may not be evident on clinical examination alone and for this reason it is best confirmed by echocardiography. Early diagnosis and transcatheter or surgical management can reduce morbidity and mortality.

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