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CASE REPORT

Familiar form of ASD with Mitral Stenosis – An Uncommon Presentation of Lutembacher's Syndrome

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Abstract:

Lutembacher's syndrome refers to combination of congenital Atrial Septal Defect with acquired mitral stenosis. Lutembacher's syndrome is a vety rare disease and in the past, it has been either over diagnosed or misdiagnosed. Here, we will discuss the case of a lady who presented with chief complaints of palpitation and dyspnea and after detailed examination and investigations; she was diagnosed as a case of "Lutembacher's syndrome".

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Introduction:

Lutembacher's syndrome is defined as the rare combination of congenital ASD and acquired mitral stenosis, it is a complex heart diseases and its incidence is very rare.[1] 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).[2] In a typical case with LS, the ASD is usually more than 15 mm in size. The incidence of Lutembacher's syndrome is 0.001/10,00000.[3] The incidence of congenital ASD in patients with mitral stenosis is 0.6-0.7%. The syndrome can present at any age but usually more common in young adults. There is a predilection for females because ASD and rheumatic MS are both more prevalent in females.[4] In one US study, the combination was found in 5 of 25,000 autopsies. This rare form of disease can be familial and may remain asymptomatic until late in life and diagnosis is often missed leading to fatal outcome. Strong clinical suspicion is important for timely diagnosis to prevent the unwanted complication from this rare complex disease especially in developing countries like Bangladesh where prevalence of rheumatic heart disease is still high. Here, we will present a case of a middle aged women who was diagnosed to be a case of Lutembacher's syndrome (LS) by transthoracic, transesophageal echocardiography and cardiac catheterization and managed accordingly. Interestingly her younger daughter and grandson was also a case of ASD (Secundum)

Case Report:

A 50 year old lady presented with shortness of breath and palpitation for 2 months. Shortness of breath and palpitation was paroxysmal in onset, usually occurred during exertion, subsided by taking rest and was associated with orthopnea, ankle swelling and pain in right upper abdomen.

On query, There were no history of accompanying chest pain, cough, hemoptysis, fever or symptoms of upper respiratory tract infections, any long term medications for any premorbid condition. No past histoty of fever, joint pains with throat infection, rashes or abnormal body movements.

She was married since the age of 20 years, had three children and all three deliveries were uneventful. Her younger daughter and grandson were also suffering from palpitation and exertional dyspnea. Younger daughter and

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grandson had no history of accompanying chest pain, cough, hemoptysis, fever, symptoms of upper respiratory tract infections, joint pains, throat infection, rashes, abnormal body movements, or any long term medications for any premorbid condition.

On general examination, she was mild anemic, ankle edema ++, afebrile with regular pulse of 116 beats/min, JVP not raised, no clubbing, no cyanosis. Blood pressure was 114/80 mm of Hg, with respiratory rate of 16 breaths/min,On cardiac examination, diffuse apical impulse was found in 6th ICS just lateral to mid clavicular line and there was left parasternal heave. On auscultation, HSl is present, mostly loud; and a low pitched, localized mid-diastolic murmur in the mitral area, a pansystolic murmur (gr.3/6) was heard at tricuspid area, and a ejection systolic murmur (2/6) in left upper sternal edge, with wide and fixed splitting of HS2. Respiratory system examination revealed, bilaterally equal normal vesicular breath sounds and no added sounds. Abdominal examination revealed tender hepatomegaly and ascites.

Investigations

ECG : Right axis deviation, RBBB. Rate 100 bpm, Sinus Rhythm. (Fig:1)

Transthorasic Echocardiography: RA, RV dilated; Moderate MS; Mild MR; Moderate TR; Severe pulmonary hypertension (PASP 70mmhg); Good LV systolic function.CFM shows, flow from LA to RA through ASD. (Fig:2)

CXR PA View: Cardiomegaly with RV type apex, enlarged pulmonary arteries & presence of pulmonaty plethora. (Fig:3)

Transesophageal echocardiography:

Multiple ASD (Secundum), largest one is 21mm, just above the aortic rim. No LA thrombus, but spontaneous echo contrast is present, LA appendage is clear. (Fig:4)

USG whole Abdomen:

Hepatomegaly, Dilatecl IVC and hepatic veins, Cardiac Catheterization: Catheter trajectojy shows, passage of catheter from IRA to LA indicating presence of ASI),

Venogram revealed: No PLSVC.

Final Diagnosis: large ASD secundum with moderate MS with mild MR with moderate TR with severe pulomonaty hypertension.

Plan: Surgical intewention So the lady was cliagnosed as LS and was transferred to cardiac surgety department for surgical intetvention.

Transthoracic Echocardiography of the patient's daughter revealed: Multiple ASD (Secundum), the largest one is about 15 mm. CXR of the patients daughter showed Cardiomegaly with RV type apex.

Transthorasic echocardiography of the patient's grandson reveled, single ASD (Secundum) of 8mm size. CXR of the patient's grandson was apparently normal.

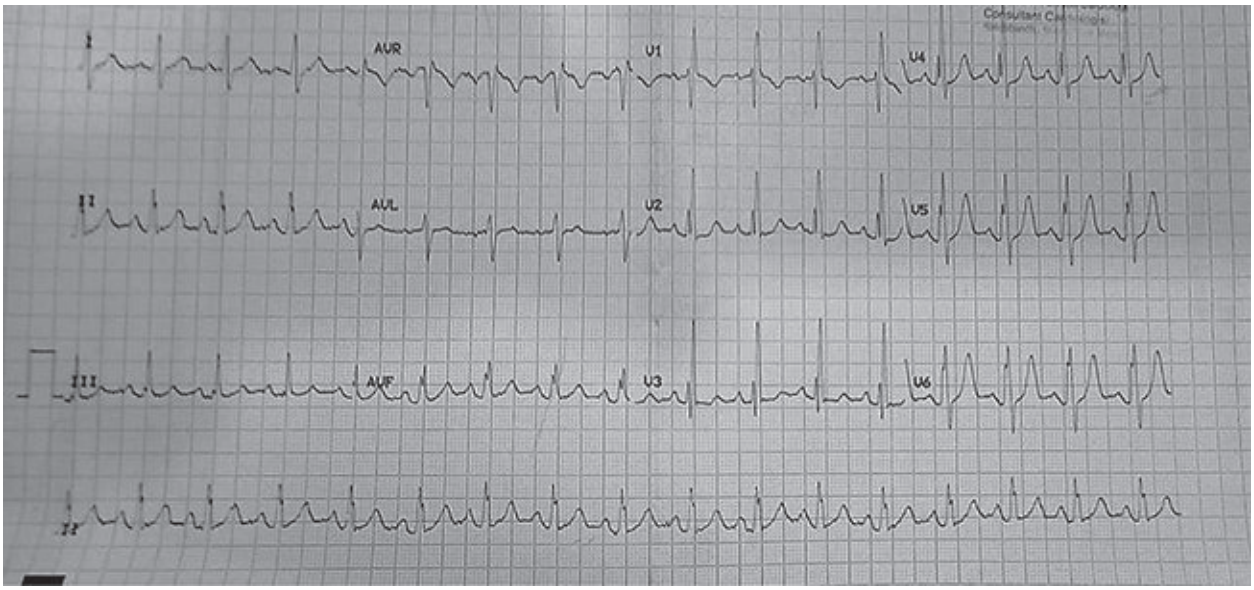

Figure 1: *ECG*

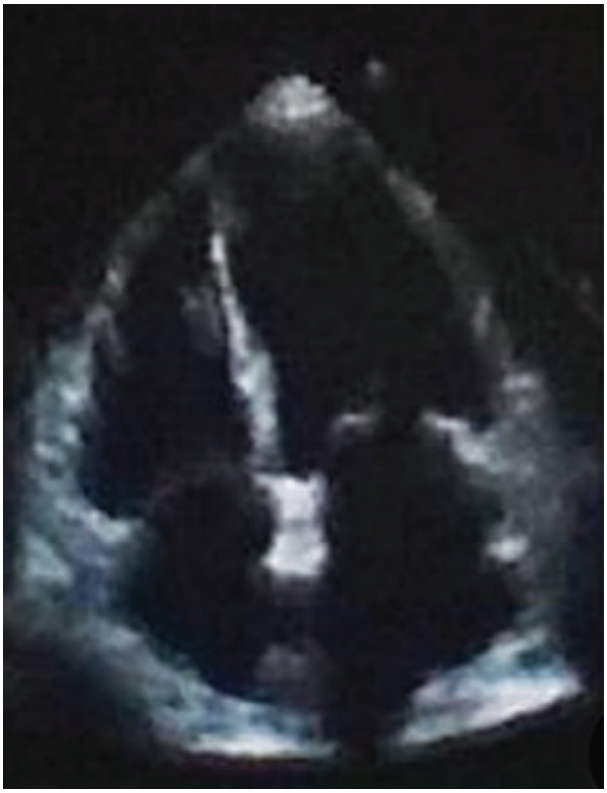

Figure 2: *TTE*

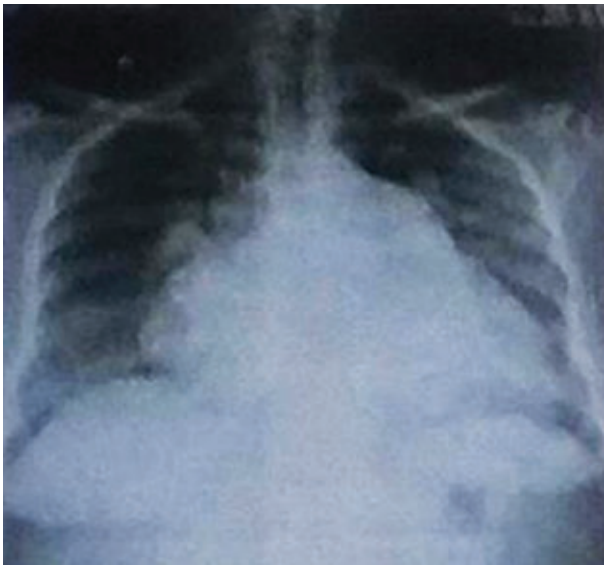

Figure 3: *CXR, PA View*

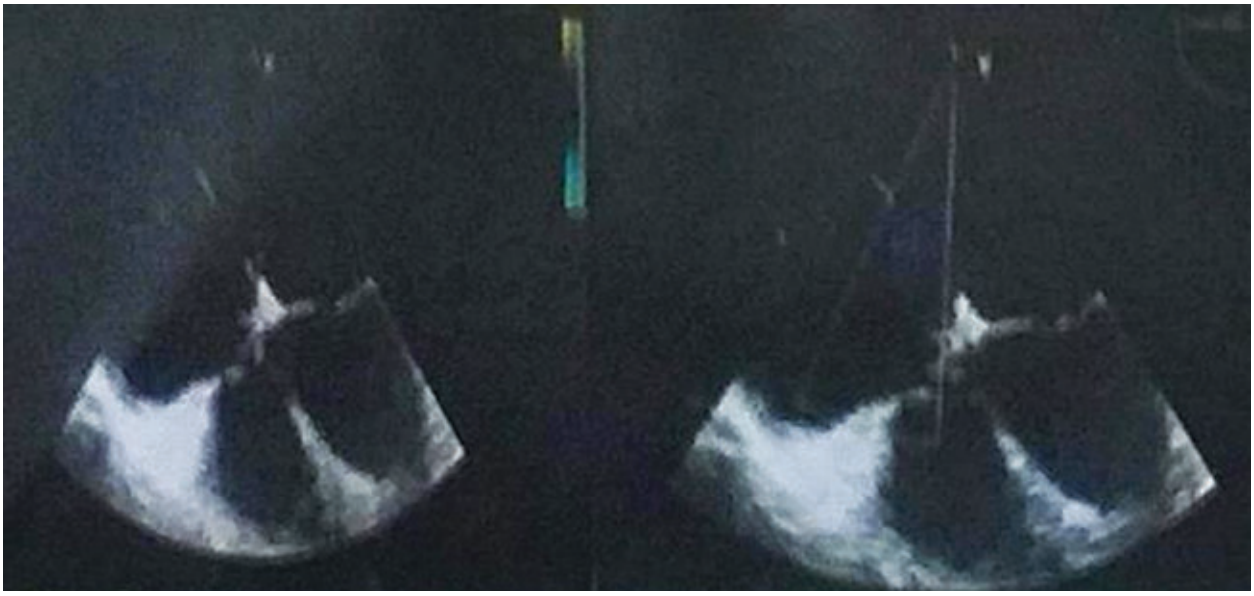

Figure 4: *TEE*

Discussion:

Lutembacher is credited with the first comprehensive account of atrial septal defect with mitral stenosis in 1916. ASD of lutembacher's syndrome is said to be large ASD. Defects defined as small when 3-6mm, moderate 6-12mm, or large >12 mm.[5] Opinions differed with regards to the type of mitral lesions to be included in this syndrome. In association with non-primum atrial septal defects, the MV can be affected in a variable manner. The lesion produced can be stenotic,

regurgitent or both, and the etiology can be rheumatic or nonrheumatic. As in our case, there was both stenosis and regurgitation of mitral valve, which is common and mitral pathology is rheumatic in origin. Each of these combinations has different prognostic and management implications. There is now a tendency to broaden the definition, by including not only MR, but also iatrogenic defects created for balloon mitral valvotomy as well as congenital defects with an intact septum.[6] The secundum atrial septal defect accounts for 10% of congenital heart disease. Familial occurrence is rare and may present as an isolated lesion or with association. The rate of transmission is 40-100%, suggestive of autosomal dominant inheritance. The hemodynamic effects of lutembacher's syndrome (LS) are a result of the interplay between the relative effects of ASD and mitral stenosis. In its initial description, the ASD was typically large in Lutembacher syndrome, thus providing another route for blood flow. The direction of blood flow is determined largely by the compliance of left and right ventricles. Normally, the right ventricle is more compliant than the left ventricle. As a result, in the presence of mitral stenosis, blood flows to the right atrium through the ASD instead of going backward into the pulmonary veins, thus avoiding pulmonary congestion. This happens at the cost of progressive dilatation and, ultimately, failure of the right ventricle and reduced blood flow to the left ventricle. Development of Eisenmenger syndrome or irreversible pulmonary vascular disease is very uncommon in the presence of large ASD and high left atrial pressure, because of mitral stenosis.[7] It is difficult to diagnosis in bed side. Mitral stenosis, at the bedside, is recognized by the combination of a loud first sound, normally split second sound, an opening snap, and a loud mid diastolic murmur with presystolic accentuation. An associated atrial septal defect decompresses the left atrium, resulting in the absence of presystolic accentuation of the diastolic murmur. Then flow across the tricuspid valve, which is relatively large because of the mitral obstruction, results in a loud mid diastolic murmur. The mitral diastolic murmur becomes inseparable from the tricuspid flow murmur and both together are heard as a loud mid diastolic murmur from the left sternal edge to the apex.[8] Color Doppler Echocardiography is the best tool for diagnosis. Transthoracic echocardiography establishes the diagnosis of LS. It is helpful in identifying the type and size of ASD and degree of MS. ASD in LS should have a diameter of more than 15mm. Mitral valve area is best calculated by planimetry, PISA and continuity equation in these cases. Pressure half time is unreliable as it gives false low value due to the simultaneous flow across ASD. ASD is also best assessed by subcostal window and thus avoids echo dropouts in apical 4-chamber view. The characteristic Doppler flow pattern across the atrial septum shows continuous or late systolic and holodiastolic left to right flow produced by the high gradient in the left atrium caused by MS.[9] These cases are better managed by early diagnosis and surgical treatment and are associated with good outcome. However the presentation amongst our rural population is usually late. The prognosis tends to worsen with the onset of pulmonaty hypertension and heart failure. The size of ASD is also crucial before the therapeutic intervention. An ASD of more than 38 mm are usually ineligible for percutaneous therapy but rather open heart surgery. Many corrective surgery options are available now. Percutaneous transcatheter therapy has become the most widely accepted therapy using balloon mitral valvuloplasty for MS (the Inoue balloon being most widely used) and the Amplatzer atrial septal occluder for closure of an ASD. Percutaneous correction is preferred to surgical correction in view of decreased morbidity and faster recovery. In advanced cases, the mortality is increased due to heart failure, cardiac arrhythmia (most commonly atrial fibrillation), and thromboembolic cerebrovascular disease. The classical LS can be corrected satisfactorily whereas the acquired LS usually need early surgical intervention as they are more prone to deteriorate.[9] This is contrary to our case, where LS was not amenable to surgery despite of being classical LS.

Conclusion:

Lutembacher's syndrome is refers to combination of congenital atonal septal defeat with rheumatic MS (rheumatic). In patients with MS who lack of typical clinical findings, the possibility of LS should be kept under consideration. Echocardiography is a helpful tool in diagnosing this condition. Early diagnosis helps a patient by undergoing only percutaneous correction of the condition as prognosis become quite worse in the advanced cases. With most of these conclusions drawn essentially from this case report, we propose prospective multicenter registries to evaluate different treatment procedures and its long term outcome in patients with IS.

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Conflicts of interest: The authors have none to declare.

Data & Materials: Available from the corresponding author, on reasonable request.

Reference

1. Bashi, V. V., Ravikumar, E., Jairaj, P. S., Krishnaswami, S., & John, S. Coexistent mitral valve disease with left-to-right shunt at the atrial level: Clinical profile, hemodynamics, and surgical considerations in 67 consecutive patients. *American Heart Journal*, 1987;*114*(6), 1406–14. doi:10.1016/0002-8703(87)90544-8
2. Vaideeswar, P., & Marathe, S. (2017). Lutembacher's syndrome: Is the mitral pathology always rheumatic? Indian Heart Journal, 69(1), 20–23. doi:10.1016/j.ihj.2016.07.003
3. Kabir, M. M., Rahman, M. L., Badruzzaman, M., Islam, M. S., & Mokhlesuzzaman. (2017). Lutembacher's Syndrome - A Case Report. KYAMC Journal, 5(1), 484–87. doi:10.3329/kyamcj.v5i1.32322
4. Kulkarni, S. S., Sakaria, A. K., Mahajan, S. K., & Shah, K. B. Lutembacher's syndrome. Journal of Cardiovascular Disease Research, 2012;3(2), 179–81. doi:10.4103/09753583.95381
5. McMahon, C. J. (2002). Natural history of growth of secundum atrial septal defects and implications for transcatheter closure. British Heart Journal, 87(3), 256–59. doi:10.1136/heart.87.3.256
6. Kale, S. B., Punithakumar, R., & Senthilkumar, R.. Sinus venosus atrial septal defect with severe mitral stenosis: a rare variant of Lutembacher's syndrome. Indian Journal of Thoracic and Cardiovascular Surgery/†: Official Organ, Association of Thoracic and Cardiovascular Surgeons of India, 2018;34(4):516–18. doi:10.1007/s12055-018-0677-4
7. Sandhya, S., Kulkarni, A. K., Sanket, K., & Kuldeep, B. Lutembacher's syndrome. J Cardiovasc Dis Res, 2012;3(2):179–81. doi:10.4103/0975-3583.95381
8. Tandon, R., Manchanda, S. C., & Roy, S. B. Mitral stenosis with left- to-right shunt at atrial level. A diagnostic challenge. Br Heart J, 1971;33:773–81. doi: 10.1136/hrt.33.5.773
9. Priyeshkant, P., Singh, K., & Zafar, H. (2016). A rare case of lutembacher syndrome in a young female: a case report from a rural population of Western Uttar Pradesh, India. India. International Journal of Research in Medical Sciences. Kant P et al. Int J Res Med Sci, 4(2), 662–665. doi: 10.1136/hrt.33.5.773.