Case Report

Lutembacher Syndrome: A Case Report
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Abstract
Lutembacher Syndrome (LS) is a rare cardiac abnormality characterized by congenital atrial septal defect (ASD) complicated by development of acquired mitral stenosis (MS). Clinical features and hemodynamic effects of LS depend on the balance of effects of the MS and the ASD. Echocardiography remains the gold standard for diagnosis and evaluation of LS. The condition is usually treated surgically; however, both abnormalities are amenable to percutaneous transcatheter therapy.

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Introduction
Lutembacher syndrome is a combination of a congenital atrial defect and acquired mitral stenosis. Mitral stenosis can be either congenital, as initially described, or acquired in origin, most commonly due to rheumatic mitral valve disease. It is more common in females than males. The natural history and hemodynamic features of patients with LS may vary and depend on the size of ASD, severity of mitral stenosis, pulmonary vascular resistance and the compliance of right ventricle. This rare form of disease may remain asymptomatic until late in life and diagnosis often missed leading to fatal outcome. Strong clinical suspicion is important for timely diagnosis to prevent the untoward 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 man who came to outdoor patient department, diagnosed to be a case of Lutembacher’s syndrome by echocardiography and managed conservatively.

Case report
The patient was a 58 year old male who presented to outdoor patient department with complaints of shortness of breath and palpitation of two months duration.

An echocardiographic evaluation was done and revealed an ostium secundum atrial septal defect with large left to right shunt with moderate mitral stenosis of rheumatic etiology. He had right atrial, left atrial as well as right ventricular enlargement. Size of ASD was 24x18 mm. His mitral valve area (MVA) was 1.1 cm² which was calculated by planimetry. There was trivial mitral regurgitation. Mild tricuspid regurgitation was present with apeak pressure gradient of 50 mmHg. Left ventricular systolic function was normal with an ejection fraction of 60%. Mitral valve leaflets were thickened, doming of anterior mitral leaflet with good pliability and posterior mitral leaflet immobile. Postero-medial commissure was free and antero-lateral commissure was fused with moderate subvalvular changes and focal calcification on both leaflets.

Discussion
In 1916, Rene Lutembacher, a French Physician described his first case of this syndrome in a 61 year old woman who had been pregnant 7 times before. It describes the combination of an ASD and mitral stenosis (MS, congenital or acquired). Lutembacher believed that the MS was congenital, but current consensus supports the opposite view, that is, the stenosis is acquired. It is almost always labeled and presumed to be rheumatic, especially in developing countries like Bangladesh with a high rheumatic case load. The ASD may either be congenital (most commonly ostium

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secundum) or iatrogenic secondary to trans-septal puncture during mitral valvuloplasty for acquired MS. ASD causes a systemic to pulmonary shunting of blood causing progressive pulmonary vascular disease and pulmonary arterial hypertension (PAH) and eventually Eisenmenger syndrome. The rare scenario of reverse LS describes development of a predominant right-to-left shunt in the context of ASD and severe tricuspid stenosis.

In patients with ASD, up to 4% have MS while the incidence of ASD in patients with MS is 0.6-0.7%. Nevertheless, some data suggests that the incidence of LS is 0.001 per million populations, while the proportion of iatrogenic LS stands at 11-12%. This rare form of disease may remain asymptomatic until late in life as in our case. This syndrome can present at any age. Although our patient was male, the disease is more common in females because ostium secundum atrial septal defect and rheumatic mitral stenosis are both more prevalent in females. There are limited epidemiological data in medical literature on this rare syndrome.

The natural history of Lutembacher’s syndrome, in general, depends on the size of the inter-atrial communication and severity of mitral valvular pathology. Patients with smaller defects and significant valvular lesions would be symptomatic mainly due to the valvular dysfunction. On the other hand in patients with larger defects, the natural history (at least in the early stages) resembles that of a pre-tricuspid shunt despite the presence of even severe valvular pathology. The MV dysfunction in these instances would increase the risk of arterial PH. With both rheumatic and nonrheumatic stenosis, the region of postero-medial comissure with adjoining leaflets and sub-valvular apparatus show a more severe pathology.

In patients with Lutembacher syndrome, the natural history of MS is favorably influenced by ASD, which decompresses the left atrium, reduces the mitral gradient, and improves symptoms associated with MS. The long-term natural history of ASD is unfavourably influenced by MS, which augments the left-to-right shunt predisposes to atrial fibrillation and right ventricular failure. The presence of MS, especially when accompanied by mitral regurgitation, increases susceptibility to infective endocarditis, in contrast to the low incidence of infective endocarditis in uncomplicated ASD. The interplay of ASD and MS are seen in Table 1.

<table>
<thead>
<tr>
<th>Table 1: Haemodynamic and clinical expression of ASD and MS</th>
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<tr>
<td>Effects of ASD on MS</td>
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<tr>
<td>Increase in left to right shunt</td>
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<tr>
<td>Pulmonary hypertension occurs earlier</td>
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<tr>
<td>Increased incidence of right ventricle failure</td>
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<tr>
<td>Increased incidence of bacterial endocarditis</td>
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<td>Increased incidence of atrial fibrillation</td>
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ASD: atrial septal defect; MS: mitral stenosis.

Clinical features are usually due to ASD and variations in symptoms and signs are dependent on the size of ASD. Commonly, patients present with fatigue, exercise intolerance and palpitations. Fatigue and exercise in tolerance are due to the reduced systemic blood flow which is a result of the MS and the systemic to pulmonary shunting of blood across the ASD in diastole, there by reducing blood flow into left ventricle. The MS and increased left atrial pressure from augmented left-to-right shunt of blood lead to atrial dilatation. This predisposes patients to atrial arrhythmias commonly atrial fibrillation (AF) and most patients present with palpitations.

In patients with non-restrictive ASD, the features of pulmonary congestion present late in the course of the disease. With moderate to severe MS, patients would present mainly with features of right ventricle (RV) overload and right heart failure (RHF). Patients with restrictive ASDs and moderate to severe MS, present much earlier and usually with features of pulmonary congestion from MS. Due to the non-specific features at the early stage of the disease, patients almost always present to hospital in advanced states.

Doppler echocardiography is the gold standard technique to establish the diagnosis of LS. It is non-invasive, available in several clinical settings, and accurate for diagnosing both ASD and MS. 3D echo and Transeosophageal echocardiography (TEE) further helpful in excluding co-existent cardiopathologies. At varying stages of LS, the 2-dimensional (2D) transthoracic echocardiography (TTE) determines the following: Left atrial enlargement, enlargement of right side cavities, ASD, pulmonary artery enlargement and mitral valve stenosis. Planimetry by Doppler echo remains the best method for assessing MVA.
Medical therapy involves symptomatic relief and prophylaxis for subacute bacterial endocarditis (SBE). Symptomatic relief of RHF: diuretics such as furosemide are generally used to ameliorate symptoms of RHF. Atrial arrhythmias: cardiac glycosides, beta blockers and calcium channel blockers may be used for rate control while drugs like amiodarone, besides rate control, also help in achieving and maintaining a normal sinus rhythm. SBE prophylaxis: LS patients who have undergone repair with prosthetic device usually need antibiotic prophylaxis for the first six months following the procedure.

Till 1990 s open heart surgery had been the preferred method of treatment of patients with LS involving ASD closure and mitral commissurotomy or valve replacement. Recently, progress in interventional cardiology has significantly changed the treatment of LS with trans-catheter therapies (in eligible patients) with impressive success rates. Whilst several techniques have been proposed for trans-catheter therapy in LS, the mos twidely used today is the Inoue balloon for percutaneous balloon mitral valvuloplasty (PBMV) for mitral valvuloplasty and the Amplatzer septal occluder for percutaneous closure of ASD.

A number of situations in which percutaneous therapy can be conducted include: ASD with Qp/Qs ratio >1.5 with inadequate rims, symptomatic moderate to severe MS with valve morphology favorable for PBMV, and any degree of pulmonary hypertension, with the exclusion of patients with Eisenmenger syndrome (irreversible pulmonary hypertension). However, the following clinical situations currently are contraindications to percutaneous therapy in LS: presence of left atrial thrombus, absence of adequate rims around the septal defect and presence of anomalous pulmonary drainage, grade 3 MR or higher, bicommissural calcification, and finally lack of expertise. Combined percutaneous therapy is documented to reduce morbidity and mortality following open heart surgery, decrease psychological trauma from thoracotomy scar and finally significantly reduces length of hospital stay following surgery.

Prognosis is influenced by several factors: pulmonary vascular resistance, right ventricle (RV) compliance, size of ASD and MS severity. But the occurrence of secondary pulmonary hypertension and congestive heart failure is commonly associated with poor outcome.

Conclusion
Lutembacher Syndrome (LS) is a rare clinical condition. Echocardiography assessment is the current diagnostic modality of choice. The prognosis is good before the onset of pulmonary hypertension and right heart failure. It is well established that early diagnosis and timely surgical treatment has good prognosis, while feasibility and safety of percutaneous treatment is demonstrated in few case reports. Outcome is poor with high mortality in the absence of appropriate and timely intervention in low income settings.

References