

LUTEMBACHER'S SYNDROME: A RARE COMBINATION OF CONGENITAL AND ACQUIRED HEART DISEASE – A CASE REPORT AND REVIEW OF LITERATURE

Uguru SU, Raphael SC, Idoko PO, Onuh JA, Amusa GA
Cardiology Unit, Department of Internal Medicine, Jos
University Teaching Hospital, P.M.B. 2076, Jos, Nigeria.

Correspondence:

Samuel U. Uguru

E-mail: ucheuguru@yahoo.com

ABSTRACT

Lutembacher's syndrome is defined as the rare combination of congenital atrial septal defect and acquired mitral stenosis. The haemodynamic effects of this syndrome are a result of the interplay between the relative effects of the atrial septal defect and mitral stenosis. Mitral stenosis augments the left to right shunt through the atrial septal defect. The definition of Lutembacher's syndrome has undergone many changes. The earliest description in medical literature was found in a letter written by anatomist Johann Friedrich Meckel to Albrecht von Haller in 1750. In 1916, Lutembacher described his first case of this syndrome, involving a 61-year-old woman, and he attributed the mitral valvular lesion to congenital mitral stenosis. Because the mitral stenosis was, in fact, rheumatic in aetiology, the syndrome was defined eventually as a combination of congenital atrial septal defect and acquired, almost always rheumatic, mitral stenosis.

Keywords: *Lutembacher's syndrome, congenital heart disease, valvular heart disease, atrial septal defect, mitral stenosis*

CASE REPORT

A 52-year-old woman presented to the Cardiology Clinic with a 2-month history of progressive breathlessness, recurrent cough (productive of mucoid sputum) and bilateral leg swelling. She also had an associated history of orthopnoea and paroxysmal nocturnal dyspnoea. On examination, she had bilateral pitting pedal oedema up to the knees and bibasal crepitations. Pulse rate was 88 beats per minute (irregularly irregular), blood pressure was 100/74 mmHg and her jugular venous pressure was raised. Her apex beat was displaced to the 6th left intercostal space, lateral to the anterior axillary line. Heart sounds were 1st and 2nd only (with the second heart sound having a loud pulmonary component). She also had grade 2/6 mitral and tricuspid regurgitation murmurs. Her liver was palpable 8 cm below the right costal margin and ascites was demonstrable by shifting dullness.

Electrocardiography (ECG) done showed atrial fibrillation with her chest x-ray showing cardiomegaly, upper lobe diversion and a full pulmonary conus. Echocardiography done showed sclerosis of the anterior mitral valve leaflet with hockey-stick appearance on B-mode. M-mode echocardiography revealed the classic Mexican hat appearance of the mitral valve consistent with mitral

stenosis. The left atrium was mildly dilated (46 mm). The right atrium and right ventricle severely dilated with severe tricuspid regurgitation and evidence of severe pulmonary hypertension. An atrial septal defect was also observed which measured 20mm. She received diuretics, digoxin, aspirin and losartan and currently stable and on follow-up at the Cardiology Clinic.



Figure 1: Two-dimensional echocardiography (apical four-chamber view) showing the atrial septal defect and mild dilation of the left atrium. Severe right ventricular and atrial dilation is also observed.

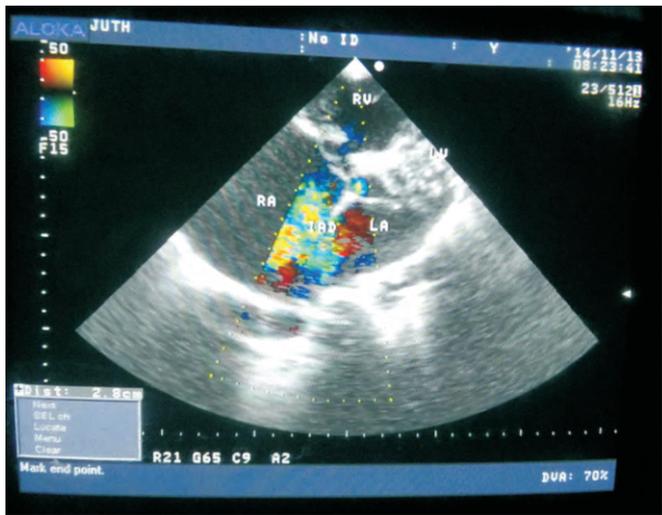


Figure 2: Two-dimensional echocardiography with colour Doppler showing flow turbulence across the atrial septal defect (IAD = inter-atrial defect).

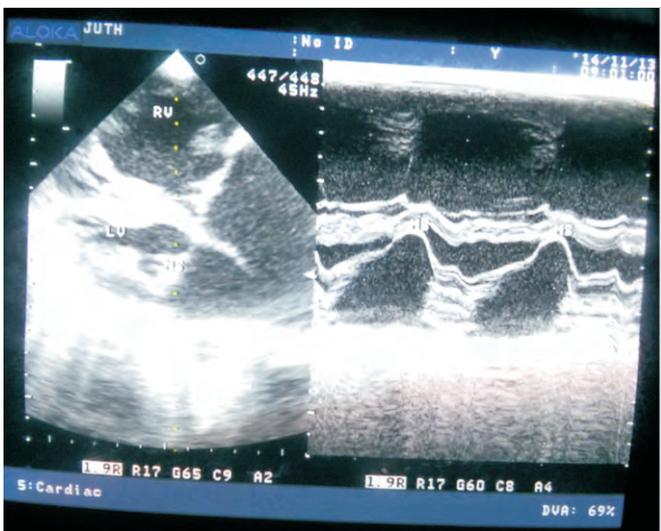


Figure 3: Two-dimensional (parasternal long axis view)/M-mode echocardiography showing sclerosis of the anterior mitral valve leaflet (left) and the classic Mexican hat appearance of mitral stenosis (right).



Figure 4: Two-dimensional echocardiography (parasternal short axis view) showing moderate mitral stenosis (mitral valve orifice area = 1.43 cm²).

DISCUSSION

Most cardiovascular conditions are either acquired or congenital in origin, but in rare instances, a combination of both is found. Lutembacher's syndrome (LS) refers to the uncommon combination of an acquired mitral stenosis (MS) and a congenital atrial septal defect (ASD) (secundum type). The interatrial septum develops from two sources: septum primum and septum secundum. If there is a defect in the formation of septum primum, it leads to the formation of a primum ASD and if the defect occurs during formation of septum secundum, it is called a secundum ASD. Other forms of LS described in the literature include iatrogenic LS and reverse LS (discussed later).

In 1916, René Lutembacher, a French Physician described his first case of this syndrome in a 61-year-old woman who had been pregnant 7 times before. LS was described as a rare combination of congenital ASD and acquired mitral stenosis. The incidence of this condition is very rare 0.001/1,000,000 according to one study published in the American Heart Journal in 1997. The condition also has a female preponderance and symptoms present at any age. An earlier case report in the literature in 1880 was of 74-year-old woman who had endured 11 pregnancies. Survival to advanced age has been reported. In one instance in an 81-year-old who experienced no symptoms related to heart disease until she reached 75 years of age. The foregoing highlights the ameliorating role of the ASD in the setting of a coexistent MS.

These favourable reports, however, should not obscure the fact that the long-term natural history of ASD is unfavourably influenced by MS, which augments the left-to-right shunt and 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 ASD in LS generally should have a diameter >1.5 cm. This later causes a systemic to pulmonary shunting of blood causing progressive pulmonary vascular disease and pulmonary arterial hypertension and eventually Eisenmenger's 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. The

haemodynamic effects of LS are usually due to the relative interplay of the effects of the MS and the ASD, and the direction of blood flow depends greatly on the compliance of the right and left ventricles. Thus, pulmonary vascular resistance, compliance of the right ventricle, size of ASD and severity of MS are recognized factors which influence the natural history and haemodynamic features in patients with this syndrome.

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.

Initially, in LS, high left atrial pressure due to MS was thought to stretch open the patent foramen ovale, causing left-to-right shunt and providing another outlet for the left atrium. Now ASD in this syndrome, like MS, is recognized as being either congenital or acquired, as discussed earlier.

LS may present at any time in the life of a patient. In low-income countries with a high incidence of rheumatic heart disease, it is seen most commonly among symptomatic young adults. In developing countries, a history of rheumatic fever is identified in up to 40% of patients with LS. Depending on the stage of the haemodynamic changes at the time of diagnosis, patients may present with fatigue and exercise intolerance or frank symptoms of heart failure as seen in the index case. Features of pulmonary congestion may also be present especially in patients with reverse LS and those with small ASD.

Cardiomegaly and/or pulmonary congestion may be seen on chest radiography while ECG may show atrial fibrillation, bundle branch block and/or right ventricular hypertrophy. However 2-dimensional echocardiography with colour flow and Doppler is the diagnostic modality of choice. Echocardiography allows identification of right atrial dilation and presence, size and type of ASD. Planimetry (preferred for MS severity as opposed to the pressure half-time method) defines the mitral valve orifice area and colour flow and Doppler demonstrate the shunt across the ASD, mitral valve stenotic gradient and presence of mitral and tricuspid regurgitation.

CONCLUSION

Early diagnosis and surgical treatment of this syndrome is associated with good outcome, but with

the onset of pulmonary hypertension and heart failure, the prognosis is usually poor. If identified early, the patient may benefit from corrective surgery, with several described surgical techniques. However, more recently, percutaneous trans-catheter 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 as there is decreased morbidity compared to open-heart surgery. There is also faster recovery with decreased length of hospital stay. When patients present in advanced states (with severe pulmonary hypertension and congestive heart failure) like in our case, and are considered ineligible for surgery, the treatment plan is to optimize medical therapy with adequate control of heart failure and with rheumatic fever prophylaxis.

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