Case Report

Congenital and Acquired Lutembacher's Syndrome Presenting in Two Adults

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Abstract

The coexistence of atrial septal defect (ASD) and mitral stenosis (MS) is defined as Lutembacher's syndrome (LS). LS was originally defined as the development of MS in a patient having a congenital ASD diagnosis. However, as the percutaneous interventions for MS have become widespread, the persistence of associated iatrogenic ASDs caused has given rise to a different form of the disease. LS may occur as spontaneous or iatrogenic ASD concomitant with acquired MS; this form is known as "acquired LS." This report presents two cases illustrating congenital LS and "acquired LS."

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

Introduction

Lutembacher's syndrome (LS) occurs as a rare combination of congenital secundum atrial septal defect (ASD) and an acquired mitral stenosis (MS).[1] Alternatively, LS may occur as spontaneous or iatrogenic ASD concomitant with acquired MS; this form is known as "acquired LS".[2] The literature also describes "reverse LS," which is characterized by a predominant pulmonary-to-systemic or right-to-left shunting of blood in the context of an ASD and severe tricuspid stenosis.[3] This report presents two cases illustrating congenital LS and "acquired LS."

Case Reports

Case presentation 1

A 53-year-old female was referred to our cardiology clinic with shortness of breath, orthopnea, and paroxysmal nocturnal dyspnea. The patient history showed percutaneous mitral balloon valvuloplasty (PMBV) for rheumatic MS in 1998. Physical examination revealed a systolic murmur in the mesocardiac area and a diastolic murmur in the apical area. The 12-lead electrocardiography showed atrial fibrillation with rapid ventricular response. Transthoracic echocardiography found that the left ventricular ejection fraction was preserved and revealed calcified and thickened mitral valve with giant left atrium, severe mitral regurgitation, severe eccentric tricuspid regurgitation with a pulmonary systolic pressure of 80 mmHg and a 5-mm sized, and mild left-to-right shunting across an ostium secundum ASD [Figure 1a-e]. The Qp/Qs ratio was 1.48. Mitral valve area (MVA) was 1.1 cm^2 according to the pressure half-time (PHT) method [Figure 1f], and the mean diastolic transvalvular mitral valve gradient was 13 mmHg. To minimize the risk of miscalculations due to decreased left atrial pressure and diastolic mitral pressure gradient, which is characteristic of ASD, MVA was also calculated as 0.9 cm^2 with the planimetry method. The patient having no ASD in previous echocardiography reports and having a history of PMBV was diagnosed with "acquired LS." This is thought to have arisen iatrogenically due to PMBV in 1998 and/or spontaneously due to extreme tension of the left atrium. Following refusal of cardiac surgery, the patient was discharged with warfarin, bisoprolol, digoxin, and furosemide therapy and a program of intensive follow-up. In follow-up program, warfarin dose titrated according to international normalized ratio and

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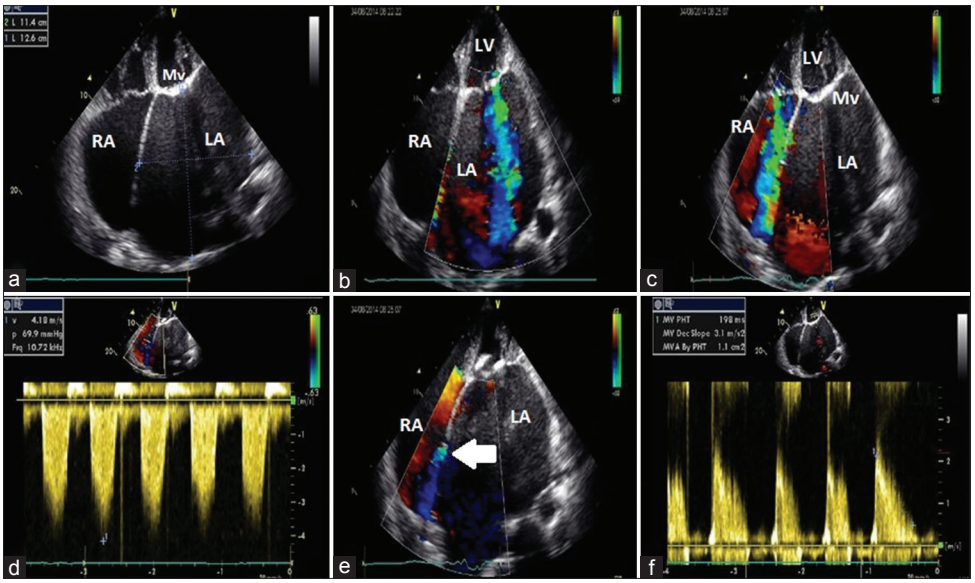

Figure 1: Transthoracic echocardiography apical 4-chamber view image showing calcified and thickened mitral valve with giant left atrium (a), severe mitral regurgitation (b), severe eccentric tricuspid regurgitation with a pulmonary systolic pressure of 80 mmHg (c and d) and mild left-to-right shunting across an ostium secundum atrial septal defect (e, arrow) and mitral valve area was 1.1 cm^2 according to the pressure half-time method (f). RA: Right atrium, La: Left atrium, LV: Left ventricle, MV: Mitral valve

bisoprolol and digoxin doses titrated according to heart rate, blood pressure, and serum digoxin level in every 3 weeks.

Case presentation 2

A 48-year-old female patient was referred to our cardiology clinic with palpitation and dyspnea. Her medical history was unremarkable. On physical examination, a diastolic murmur of 2/6° in the apical area and a systolic murmur in the mesocardiac area were auscultated. The 12-lead electrocardiography showed sinus rhythm with frequent atrial premature complexes, and *P* wave morphology was consistent with "P mitrale." Transthoracic echocardiography showed thickened, fibrotic mitral valves with a valve area of 1.6 cm^2 using the planimetry method [Figure 2a and b]. Doppler echocardiography revealed mild mitral and tricuspid regurgitation, a 4-mm sized, left-to-right shunting across an ostium secundum ASD [Figure 2c and d]. The patient was diagnosed with LS with the coexistence of MS and ASD. The Qp/Qs ratio was 1.22 and systolic pulmonary arterial pressure was 38 mmHg. The patient was discharged with metoprolol therapy and a program of echocardiographic follow-up in every 6 months.

Discussion

The coexistence of congenital ASD and acquired MS is defined as LS.[1] LS is a rare condition with a reported incidence of 0.001 in one million. Since both ASD and MS are frequently found in women, LS also occurs more commonly among women.[4] Hemodynamic interactions between ASD and MS are challenging. As in LS, one lesion can alter the clinical and hemodynamical effects of the other lesion. Clinical and hemodynamic interactions are dependent on three of the following properties: the diameter of ASD, severity of MS, and compliance of the right ventricle.[5] In a patient with MS, a large ASD provides an additional outflow point for the left atrium. The left atrium is decompressed via ASD, resulting in decreased left-atrial and pulmonary capillary pressures. Consequently, symptoms observed in isolated MS patients, such as exertional dyspnea, orthopnea, and paroxysmal dyspnea, occur in the later phases of the disease. Blood flow through the mitral valve is decreased due to an increase in the left-to-atrial shunt. The result of this effect is a decrease in the severity of mitral murmur and presystolic accentuation in patients having sinus rhythm, and the opening snap cannot be auscultated.[6] On the other hand, increased left-atrial pressure due to MS progressively increases the left-to-right shunt via ASD. In particular, in the presence of a large ASD, the right ventricular volume and pressure overload will increase over time, causing the right ventricle to dilate, leading to pulmonary hypertension and right-heart failure.[2] In summary, the coexistence of MS and ASD has a distinct course as compared with isolated MS. The existence of ASD in an MS patient can mimic the symptoms and auscultatory findings of the disease. Increased left-atrial pressure due to MS increases the left-to-atrial shunt, causing early right-heart failure and pulmonary hypertension.

LS was originally defined as the development of MS in a patient having an ASD diagnosis. However, as the percutaneous interventions for MS have become widespread, the persistence of associated iatrogenic defects caused has given rise to a different form of the disease. This condition, "acquired LS," may result from two different scenarios. In the first scenario, in isolated MS patients, an "iatrogenic ASD" may occur due to PMBV, which is performed through atrial septal puncture by

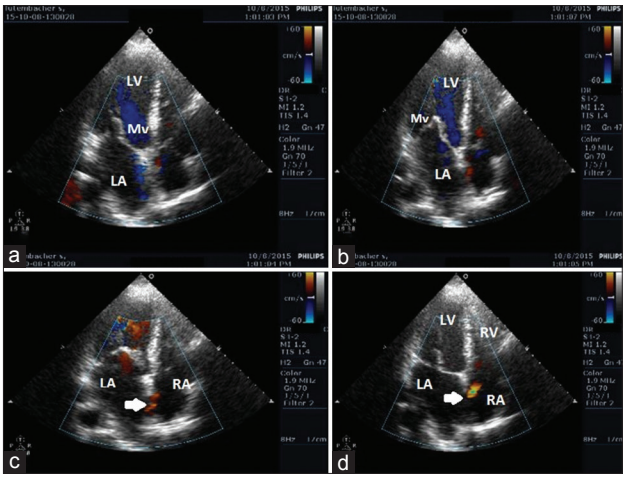

Figure 2: Transthoracic echocardiography apical 4-chamber view image showing thickened, fibrotic, and stenotic mitral valves (a and b), Doppler echocardiography revealed left-to-right shunting across an ostium secundum atrial septal defect (c and d, arrow). RA: Right atrium, LA: Left atrium, RV: Right ventricle, LV: Left ventricle, MV: Mitral valve

the transseptal approach.[2] In a trial by Yoshida *et al*., within the first 24 h of the procedure, 50% of the patients had ASD in transthoracic echocardiography, and 87% of the patients had ASD in transesophageal echocardiography.[7] Iatrogenic ASD, which occurs after PMBV, usually has a diameter of 0,0.5–1 cm. Defects that are >1 cm rarely occur and are associated with large left-to-right shunts. Iatrogenic defects that are <0.7 cm have a tendency to close spontaneously within 24 h.[8] The second scenario for "acquired LS" is spontaneous secundum type ASD development due to progressive left-atrial enlargement and pressure overload in the presence of severe MS.[2]

The parameters used to assess the severity of MS are as follows: MVA, mitral valve mean diastolic gradient, and pulmonary arterial pressure. The most commonly used echocardiographic methods for calculating MVA are PHT, planimetric MVA calculation, and continuity equation.[9] In LS patients, the existence of ASD directs blood flow from the left-to-right atrium, causing diminished blood flow through the mitral valve in the diastole. Diminution of blood flow decreases PHT, causing an underestimation of the severity of MS. For this reason, PHT is inappropriate and not recommended for calculating MVA in LS patients. The appropriate choices for calculating MVA are planimetry and continuity equation. However, in patients with atrial fibrillation, MVA cannot be calculated via continuity equation.[10]

LS has a good prognosis if it is diagnosed early. In patients who are diagnosed before the development of right-heart failure or pulmonary hypertension, survival is excellent following mitral valve replacement and ASD closure. However, prognosis will be poor if pulmonary hypertension and right-heart failure exist at the time of diagnosis.

Conclusion

The purpose of this report is to review the clinical characteristics, echocardiographic diagnosis, and prognosis of LS. LS is a rare diagnosis for most adult cardiologists. This report seeks to help clinicians to better grasp the clinical and echocardiographic characteristics of "acquired LS" presenting in older patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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