Lutembacher's Syndrome and Persistent Left Superior Vena Cava in a Young Female

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ABSTRACT

We report the case of a 28-year-old woman with ostium secundum atrial septal defect, mitral stenosis and a persistent left superior vena cava, who developed heart failure at age 17 and was admitted at different institutions for shortness of breath. The patient had one complicated pregnancy at age 25 in which she presented with worsening heart failure. She was able to deliver her baby via spontaneous vaginal delivery under epidural anesthesia, but had to stay admitted for an additional 3 weeks.

Two-dimensional echocardiogram with doppler studies showed an ostium secundum atrial septal defect, severe rheumatic mitral stenosis (Mitral Valve Area = 0.60 cm^2 by planimetry and 0.90 cm^2 by pressure half-time), severe tricuspid regurgitation, mild aortic regurgitation, pulmonary regurgitation with severe pulmonary hypertension, and a dilated coronary sinus. During bubble contrast study, micro-bubbles from the agitated saline appeared initially in the coronary sinus before appearing in the right sided chambers, suggesting the presence of a persistent left superior vena cava. Cardiac magnetic resonance imaging confirmed the initial echocardiographic findings. Cardiac catheterization and venography confirmed the presence of a persistent left superior vena cava which was draining into a markedly dilated coronary sinus, a severe mitral stenosis (MVA=0.86 cm^2), an atrial septal defect with a left-to-right shunt (Qp:Qs = 2:1), and severe pulmonary hypertension. The combination of an atrial septal defect and mitral stenosis is rare, with an incidence ranging from 0.6 to 4% among those with congenital disease. The incidence of the combination of the three lesions – ASD, mitral stenosis and persistent left superior vena cava – is unknown. Since the pulmonary vascular disease was still reversible based on the hemodynamic studies, the plan was to have the patient undergo mitral valve replacement, tricuspid valve annuloplasty, and patch closure of the atrial septal defect. Before any surgical procedure could be done however, the patient succumbed to heart failure.

Key Words: congenital heart disease, atrial septal defect, rheumatic heart disease, mitral stenosis, echocardiography, contrast echocardiography, cardiac magnetic resonance imaging, heart failure, cardiology, persistent left superior vena cava

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Case Report

A 28 year old female with shortness of breath since eleven years ago presented at the outpatient clinic with worsening heart failure symptoms. At age 17, she was seen by a physician who advised her to undergo a 2D echocardiogram. She took various unrecalled medications which allowed her to perform household chores without feeling short of breath. Seven years after the initial consult, she became pregnant and began to experience bipedal edema and shortness of breath when performing ordinary activities. In the interim between her first clinical consult and her pregnancy, she visited various clinics and hospitals, complaining of dyspnea on exertion, orthopnea and edema. Due to an acute deterioration in her functional status, she was admitted to the intensive care unit of a tertiary hospital, where she delivered via spontaneous vaginal delivery without obstetric complications. She had to stay an additional three weeks to allow her to recover from the ordeal and she was discharged with a prescription for diuretics and digoxin. She was unable to have a 2D echocardiogram done due to financial reasons.

The initial 2D echocardiogram, done almost a year after her pregnancy, revealed an ostium secundum atrial septal defect with a left to right shunt and severe rheumatic mitral stenosis. In the next three years, she was admitted at our institution several times for acute decompensated heart failure, often precipitated by a respiratory tract infection.

In a recent physical examination, she was found to have an elevated jugular venous pressure and rales on both basal lung fields on auscultation. In addition, she was found to have bipedal edema, a right ventricular heave, a grade 3/6 holosystolic murmur heard at the left parasternal border which increases in intensity on inspiration, and a grade 2/6 diastolic rumble at the cardiac apex. The patient's liver edge was palpable 2 cm below the right subcostal margin.

Right axis deviation and right ventricular hypertrophy were noted in her electrocardiogram. A repeat transthoracic 2D echocardiogram revealed a dilated coronary sinus and a thickened mitral valve with an anterior leaflet which demonstrated diastolic doming and a fixed and upright posterior leaflet (see Figure 1A). On M-mode, there was flattening of the EF slope and loss of antiparallel motion (see Figure 1B), consistent with mitral stenosis. The mitral valve area was computed to be 0.60 cm^2 by planimetry and 0.90

cm^2 by pressure half-time. An ostium secundum type atrial septal defect was also seen, with a predominantly left-to-right shunt (see Figures 2A and 2B). The other echocardiographic findings were severe tricuspid regurgitation, mild aortic regurgitation, and pulmonary regurgitation with severe pulmonary hypertension.

A bubble contrast study was done to further investigate the dilated coronary sinus. On left sided injection with agitated saline, micro-bubbles appeared initially in the coronary sinus before appearing in the right sided chambers, suggesting the presence of a persistent left superior vena cava.

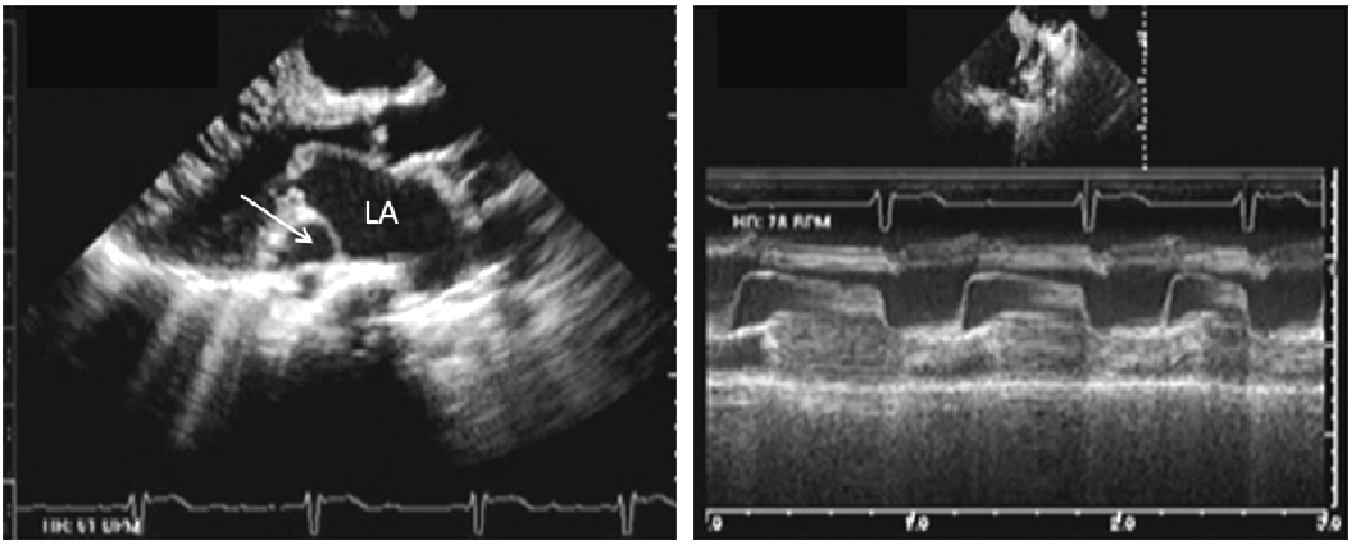

Figure 1A. Parasternal long axis reveals diastolic doming motion of the anterior mitral valve leaflet and dilated coronary sinus (arrow). During left sided injection of bubble contrast, microbubbles first appeared in the dilated coronary sinus before appearing in the right ventricle, suggesting the presence of left sided superior vena cava.
Figure 1B. M-mode demonstrates flattening of the EF slope and loss of antiparallel motion. LA, left atrium.

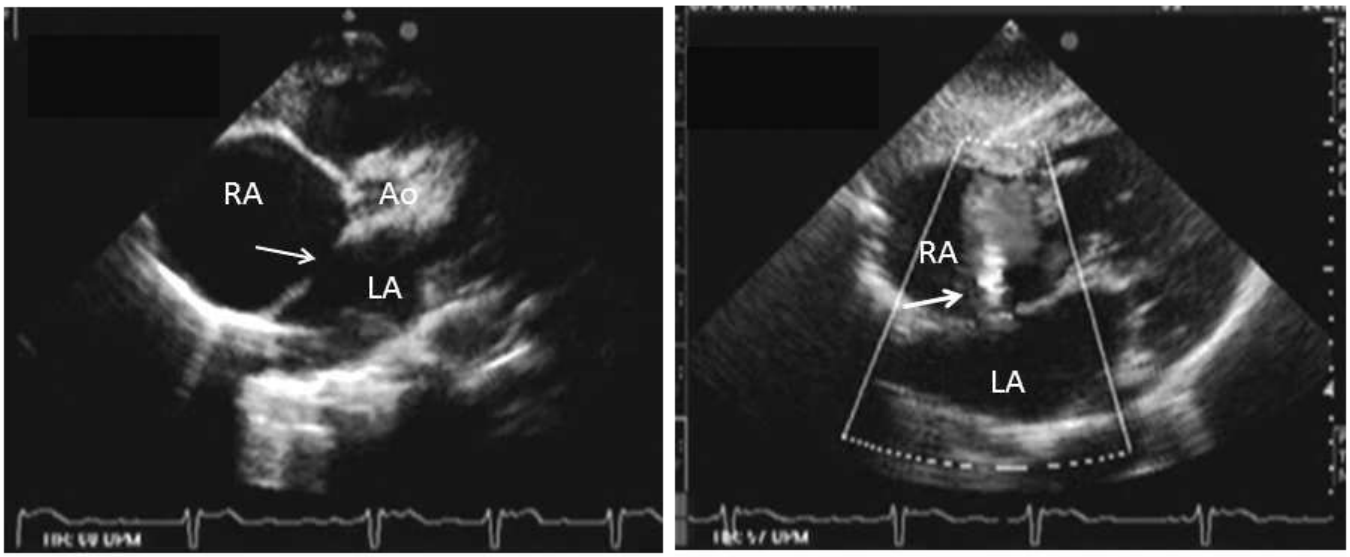

Figure 2A. Short axis view at the level of the aortic valve shows the ostium secundum atrial septal defect (arrow).
Figure 2B. Doppler examination in the subcostal view reveals the left to right flow across the atrial septal defect (arrow). RA, right atrium. Ao, Aortic valve. LA, left atrium.

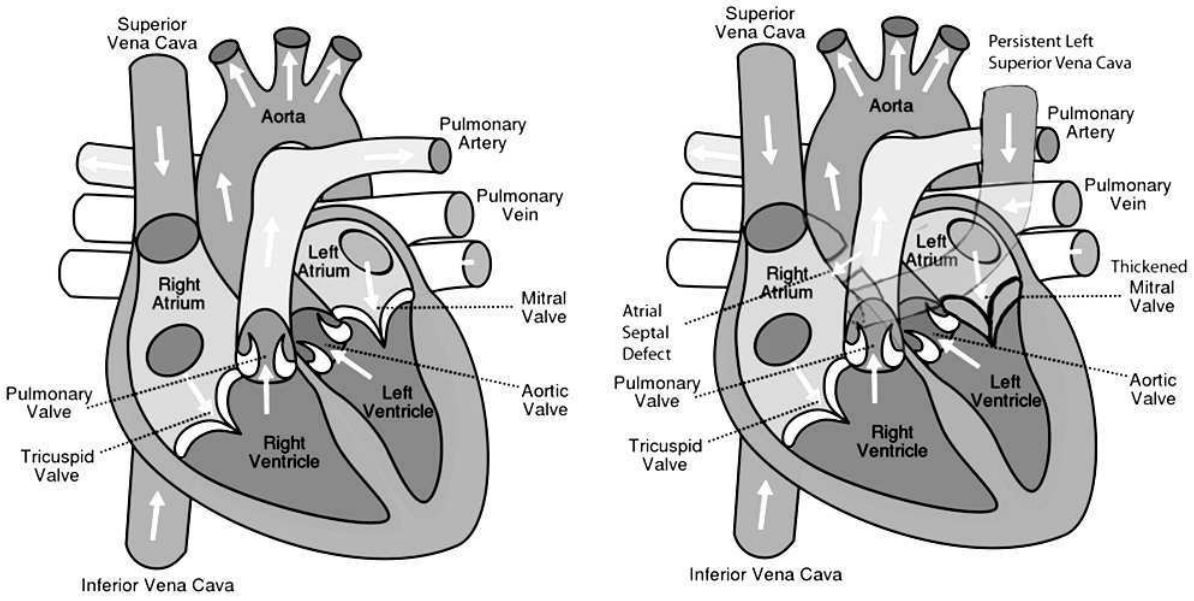

Figure 3. Diagrammatic representation of a normal heart (left) and a heart with an atrial septal defect, mitral stenosis and a persistent left superior vena cava draining into the right atrium (right). Chamber enlargement of the left atrium, right atrium and right ventricle were not reflected in the diagram. The figure was based on a diagram of the heart (http://commons.wikimedia.org/wiki/File:Diagram_of_the_human_heart.svg) by Wikimedia user Wacaplet (http://en.wikipedia.org/wiki/User:Wapcaplet). Alterations done by the author. This figure is licensed under the Creative Commons Attribution ShareAlike 3.0 License (http://creativecommons.org/licenses/by-sa/3.0/).

A diagrammatic representation of a normal heart and a heart with the three lesions – namely, an atrial septal defect, rheumatic mitral valve stenosis, and a persistent left superior vena cava – is shown in Figure 3.

Transesophageal echocardiography and cardiac magnetic resonance imaging confirmed the initial echocardiographic findings (see Figure 4). The cardiac catheterization findings provided confirmatory evidence of the anatomical and hemodynamic abnormalities, as well as information valuable for prognostication. The measured significant O_2 step up at the level of the right atrium was consistent with the presence of an atrial septal defect. There was a significant pressure gradient between the left ventricle and left atrium (left atrial pressure as measured by PCWP) because of the mitral stenosis. The cardiac output, computed through Fick's method, was low (CO=1.66 L/min). The ratio of pulmonic to systemic blood flow (Qp:Qs) measured by cardiac catheterization was 2:1. The mitral valve area was computed to be 0.86 cm^2. Venography confirmed the presence of a left superior vena cava which drained into the markedly dilated coronary sinus (see Figure 5). The pulmonary-to-systemic vascular resistance ratio (Rp/Rs) was computed to be 0.40 but this improved to 0.11 with oxygen. The pulmonary vascular disease was deemed reversible, thus the patient, after being briefed of the operative risks, was advised to undergo mitral valve replacement, tricuspid valve annuloplasty, and patch closure of the atrial septal defect. She however succumbed to complications of heart failure before any surgical procedure was done.

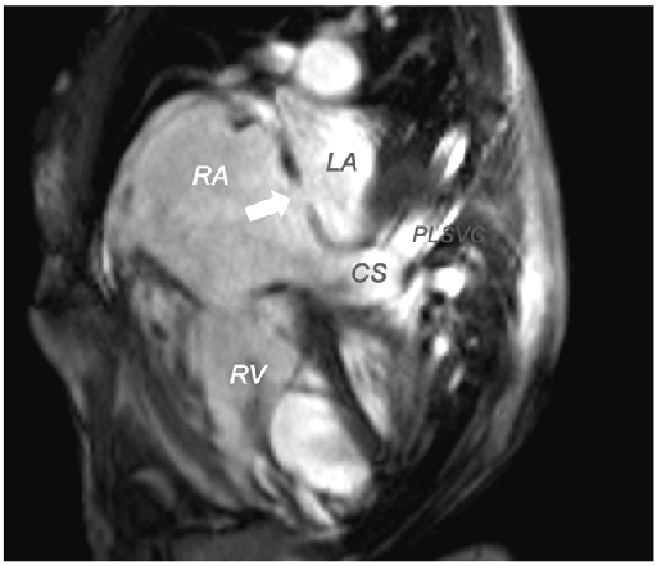

Figure 4. Magnetic Resonance Imaging showing the Atrial Septal Defect (arrow) and the persistent left superior vena cava draining into the dilated coronary sinus. Atrial Septal Defect (arrow); RA, right atrium; LA, left atrium; CS, coronary sinus; PLSVC, persistent left superio vena cava; RV, right ventricle

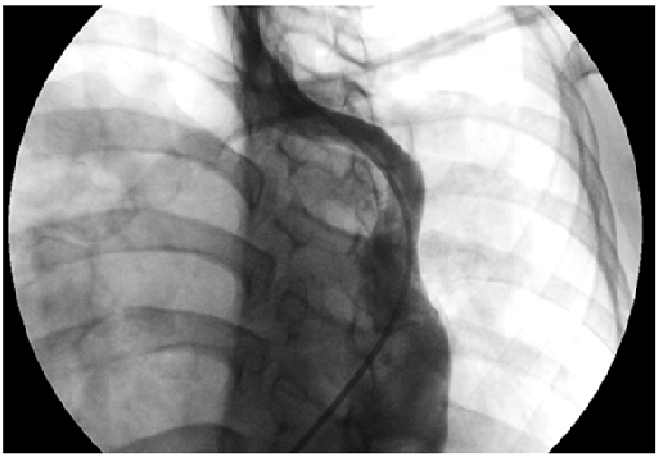

Figure 5. Cardiac catheterization confirmed the presence of a persistent left SVC and provided additional information regarding the other lesions.

Discussion

The earliest reference to a case of atrial septal defect with concomitant mitral stenosis was written in May 5, 1750 by the anatomist Johann Friedrich Meckel in a letter to his mentor Albrecht von Haller.[1] The syndrome was described in 1916 by Rene Lutembacher, but the French cardiologist attributed the mitral valve abnormality found in his 61 year old female subject to congenital mitral stenosis. Later, it would be discovered that the mitral anomaly was a secondary lesion, often rheumatic in origin, and this lead to the contemporary definition of Lutembacher syndrome as a combination of congenital atrial septal defect and acquired mitral stenosis.

Lutembacher syndrome is more prevalent in females, but this is not surprising since both rheumatic mitral stenosis and atrial septal defect are seen predominantly in women. The combination of the two lesions is rare, the incidence of mitral stenosis in patients with ASD being 4% and ASD in patients with mitral stenosis, 0.6-0.7%.[2] A history of rheumatic fever is often absent. Characteristic symptoms of pulmonary venous congestion, such as orthopnea, paroxysmal nocturnal dyspnea, and pulmonary edema, are seen less frequently in individuals with Lutembacher syndrome compared to those with uncomplicated mitral stenosis. Fatigue, however, is more common due to the low cardiac output.

The clinical characteristics of Lutembacher syndrome arise from the interaction of the two cardiac lesions. The presence of mitral stenosis augments the left-to-right shunting through the atrial septal defect. The existence of ASD, in turn, decompresses the left atrium, leading to a decrease in the diastolic mitral pressure gradient. The interatrial communication thus has an ameliorating effect on the clinical course of mitral stenosis. This effect is evident even in the earliest case reports: Lutembacher's original patient, for example, lived to be 61 years old and was able to endure seven pregnancies.[3]

Also as a result of these hemodynamic changes, the classic findings in mitral stenosis – namely, a loud S1, an opening snap and an apical diastolic murmur with presystolic accentuation – are attenuated. A fixed, widely split S2 occurs because increased left-to-right shunting through the atrial septal defect results in late closure of the pulmonic valve and decreased left ventricular and aortic flow leads to early closure of the aortic valve.

As an added consequence to the altered hemodynamics in Lutembacher syndrome, the degree of mitral stenosis may be underestimated if determination is done through pressure half-time.[2,4] In Lutembacher syndrome, mitral valve area is best estimated echocardiographically through planimetry.

Definite mortality and morbidity data are not available but are thought to be dependent on the severity of the dominant lesion (atrial septal defect or mitral stenosis) and the degree of pulmonary hypertension. For example, when mitral stenosis occurs together with a small or restrictive atrial septal defect, the clinical presentation and course approximates that of an isolated mitral stenosis.[5] Significant mitral lesions combined with large, nonrestrictive atrial septal defects, on the other hand, can lead to significant left-to-right shunting, subsequently resulting in severe pulmonary hypertension and increased pulmonary vascular resistance.[6] Generally, the prognosis is good because of the ameliorating effect of the atrial shunt on the mitral valve lesion. In fact, survival to the eighth decade has been reported.[7]

Persistent left superior vena cava (PLSVC), considered the most common thoracic venous anomaly, is a rare vascular abnormality with a prevalence of 0.3% among healthy individuals and 4.4% in those with congenital heart disease.[8] The defect is the result of the left anterior cardinal vein's failure to involute.

The PLSVC is typically suspected because of an incidental finding of a dilated coronary sinus on transthoracic echocardiography. Rarely, the PLSVC drains directly into the right or left atrium, but usually it drains into the right atrium through a dilated coronary sinus.[9,10] Diagnosis via transthoracic echocardiography can be made through injection of agitated saline into the left brachial vein. If a PLSVC is present, bubble contrast will opacify the dilated coronary sinus before the right-sided chambers. This congenital anomaly usually is of little hemodynamic consequence and is considered clinically significant only in cases where cardiac catheterization or instrumentation is being considered.

The indications for intervention are the following: 1) a Qp/Qs ratio of more than 1.5:1, 2)moderate to severe mitral stenosis, and 3) the presence of reversible pulmonary

hypertension. Although experience is limited, transcatheter treatment of Lutembacher syndrome – with balloon commissurotomy and the use of an Amplatzer ASD occluder – is now considered a viable non-surgical option.[2,6,11,12] In this case, this option was not entertained due to the financial limitations of the patient. Of great concern was the patient's severe pulmonary hypertension but this was demonstrated to be reversible through hemodynamic studies.

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