Lutembacher Syndrome An Echocardiography Assessment: A Rare Case

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ABSTRACT

Background: Lutembacher Syndrome (LS) is a rare heart defect, characterized by the presence of Atrial Septal Defect (ASD) (congenital or acquired) and Mitral Stenosis (MS) (congenital or acquired). Case Description: A 56 years old woman came to the cardiovascular clinic at Balaraja Hospital with complaints of shortness of breath has been felt since the last 3 months. Shortness of breath has been aggravated by activity and relieved by rest. Shortness of breath is also felt when lying down, if she sleeps, she has to use 2-3 pillows or an elevated position. On physical examination there was an increase in jugular venous pressure (JVP) 5+4 cm, irregular 1st and 2nd heart sounds, and a grade 2/4 diastolic murmur at the apex and a pansystolic murmur were found. grade 3/6 in the tricuspid area. During auscultation, wide fixed splitting was also found. On examination of the extremities, edema was found in both legs. An electrocardiogram (ECG) examination revealed atrial fibrillation (AF), bigemini ventricular extrasystole (VES) and echocardiography conclusion were found Reumatic Heart Disease, MS severe, mild mitral regurgitation, severe tricuspid regurgitation, pulmonary hypertension, ASD secundum left to right shunt.

Conclusion: Lutembacher Syndrome is still a rare disease, including in Indonesia. If diagnosed early and appropriate therapy is carried out, it will provide a good prognosis, if the diagnosis is late, it will increase mortality due to heart failure, arrhythmia and embolic stroke.

Keyword: Lutembacher syndrome, rare case, heart failure, case report.
INTRODUCTION

Lutembacher Syndrome (LS) is a rare heart defect, characterized by the presence of Atrial Septal Defect (ASD) (congenital or acquired) and Mitral Stenosis (MS) (congenital or acquired). The hemodynamic effects of these two diseases are very interesting. ASD decreases left atrial pressure and mitral valve gradient whereas mitral stenosis increases left to right flow through ASD, making diagnosing this disease very difficult. Lutembacher Syndrome can occur at any age, but most often occurs in young women. ASD is estimated to have a prevalence of 56 per 100,000 live births and 7%-10% of congenital heart disease cases occur in adults. Meanwhile, congenital Mitral Stenosis is rare with a prevalence of 0.6% of the total cases of congenital heart disease cases occur in adults. Based on data published by Global Burden Of Disease 2015, approximately 33.4 million people in the whole world suffers from rheumatic heart disease. The prevalence of Lutembacher syndrome is reported to be 0.001 per million population with a predominance of women. This disease is more likely to occur in areas with a higher prevalence of rheumatic heart disease. Therefore, this disease is more frequently reported in countries where developing countries such as Southeast Asia. In addition, in developing countries 40% of patients with Lutembacher syndrome are reported to have a previous history of rheumatic fever.

Therefore, we report the case of a 56 year old woman who came for treatment to the Balaraja Hospital with complaints of shortness of breath accompanied by a pounding chest. Echocardiography examination revealed MS and ASD.

CASE PRESENTATION

A 56 year old woman came to the cardiovascular clinic at Balaraja Hospital with complaints of shortness of breath that came and went. Shortness of breath has been felt since the last 3 months. Before 3 months,
the patient had no complaints of shortness of breath and activities as usual. In the last 3 months, shortness of breath has been aggravated by activity and relieved by rest. Shortness of breath is also felt when lying down, if you sleep you have to use 2-3 pillows or an elevated position. Shortness of breath is also accompanied by chest pounding and swelling in both legs. The patient also complained of feeling full in the stomach and coughing up phlegm for 2 weeks, the phlegm was white and foamy. The patient had no history of hypertension, diabetes mellitus or previous lung disease.

On examination of vital signs, the consciousness status was componens and appeared moderately ill, blood pressure 115/57 mmHg, pulse 66 times/minute, respiratory rate 24 times/minute, temperature 36.8 degrees Celsius, and oxygen saturation 96% room air. On physical examination there was an increase in jugular venous pressure (JVP) 5+4 cm, on chest examination vesicular breath sounds were found, there were no soft wet rhonchi, irregular 1st and 2nd heart sounds, and a grade 2/4 diastolic murmur at the apex and a pansystolic murmur were found. grade 3/6 in the tricuspid area. During auscultation, wide fixed splitting was also found. On examination of the extremities, edema was found in both legs.

Supporting examination, Diagnosis and treatment

An electrocardiogram (ECG) examination revealed atrial fibrillation (AF), bigemini ventricular extrasystole (VES). Echocardiography showed dilated RA, RV, LA, concentric remodeling, normal LV systolic function with LVEF 83%, D shape LV, diastolic could not be assessed due to atrial fibrillation, mitral valve calcification due to RHD, MS severe, MR mild, TR severe, high probability of PH, reduce RV contractility, IAS gap 2.5cm, ASD secundum left to right shunt. Conclusion echocardiography RHD, MS severe, MR mild, TR severe, pulmonary hypertension, ASD secundum left to right shunt. Diagnosis of Lutembacher Syndrome, Pulmonary Hypertension, AF NVR + Multifocal PVC. The treatment given
was furosemide 1 x 40mg, spironolactone 1 x 50mg, bisoprolol 1 x 2.5mg, lisinopril 1 x 2.5mg, warfarin 1 x 2mg.

Figure 1. Electrocardiogram (ECG)

Figure 2. 2D Echocardiography Depicting a Stenosed Mitral Valve with Dilated Right Ventricle in Left Parasternal Long Axis View.
Figure 3. Parasternal Short Axis Echocardiogram Showing LV D Shape due to Increased Pressure in the Right Ventricle

Figure 4. Apical Four Chamber Echocardiogram showing Dilated Right Heart Chambers with Atrial Septal Defect
Corvisart first described the association of MS with ASD in 1811. René Lutembacher, a French doctor born in 1884, first described the combination of rheumatic MS and ASD (usually the patent foramen ovale type) and the data were first published in 1916.8

Here we report a very rare case, a 56 year old female patient who was newly diagnosed with congenital heart disease, namely ASD accompanied by Mitral stenosis since 3 months SMRS. The main complaints were shortness of breath when doing activities and lying down, palpitations, swelling of both legs, productive cough for 2 weeks and discomfort in the stomach. This complaint is due to reduced systemic blood flow caused by MS and the presence of a shunt from left to right through the ASD during diastole, thereby reducing blood flow to the left ventricle.4 The presence of a blood shunt through the ASD prevents pulmonary congestion in the early stages of the disease. atrial dilatation causing atrial palpitations in this patient. Palpitations are caused

DISCUSSION

Figure 5. Apical Four Chamber Echocardiogram showing Severe TR (Tricuspid Regurgitation)
by atrial fibrillation. Patients with non-restrictive ASD complain of pulmonary congestion appearing later than those with restrictive ASD and moderate to severe MS. On physical examination, the manifestation of ASD accompanied by MS is very difficult. In MS there is a diastolic murmur at the apex, opening snap, hardened 1st heart sound and RV tapping which may be difficult to find due to LA decompression through the ASD, whereas in ASD there is a wide fixed splitting and a systolic murmur at the pulmonary valve. In our case there was a grade 2/4 diastolic murmur at the apex and a grade 3/6 pansystolic murmur in the tricuspid area. During auscultation, wide fixed splitting was also found.

On ECG examination, AF NVR and Multifocal PVC were found. Atrial fibrillation is caused by dilatation of the left and right atria which receive a lot of blood due to ASD shunting. Bigemini VES with LBBB morphology is thought to occur due to RV dilatation.

Echocardiography was the modality of choice for diagnosis and evaluation in our patients. Echocardiography examination found dilated RA, RV, LA, normal LV systolic function with LVEF 83%, D shape LV, diastolic cannot be assessed due to atrial fibrillation, mitral valve calcification due to RHD, MS severe, MR mild, TR severe, high probability of PH, reduce RV contractility, IAS gap 2.5cm, ASD secundum left to right shunt.

The prognosis for patients with Lutembacher syndrome is better if detected early and pulmonary hypertension has not occurred. Initial treatment can use diuretics to reduce symptoms of right heart failure and pulmonary congestion if present. Beta blockers and CCBs can be used for rate control therapy for atrial fibrillation. Also, antibiotics can be given for prophylactic therapy for infective endocarditis.

Definitive therapy for Lutembacher syndrome is open heart surgery (OMV/MVR with surgical repair for ASD). Surgical therapy is only for large ASDs that cannot be closed with a device or MS that cannot be corrected with a BMV. Percutaneous transcatheter in BMV form for MS correction and
subsequent planning of ASD closure is preferred. If at the time of ASD closure, the MS is not corrected, it will cause pulmonary edema post-surgery.\(^4\)\(^,\)\(^1\)\(^1\)

**CONCLUSION**

Lutembacher Syndrome is still a rare disease, including in Indonesia. If diagnosed early and appropriate therapy is carried out, it will provide a good prognosis and reduce mortality and morbidity. If it is diagnosed late and there is already pulmonary hypertension, it will give a poor prognosis which will cause symptoms of pulmonary hypertension. Mortality will increase due to heart failure, heart rhythm disturbances and embolic stroke.

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