Jurnal
Kardiologi Indonesia
J Kardiol Indones. 2012;33:99-105
ISSN 0126/3773

Case Report

# Lutembacher's Syndrome An Echocardiographic Assessment

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Lutembacher'ssyndrome (LS), consisted of Atrial Septal Defect (ASD) and Mitral Stenosis (MS), is a very rare form of cardiac anomaly. Rene Lutembacher'sfirst described this syndromein 1916. Currently, any combination of ASD, congenital or iatrogenic and MS, acquired or congenital is referred as LS.

By using echocardiography, the hemodynamic of LS could be assesed. Pathophysiologically, the hemodynamic of ASD is relatedto the magnitude and direction of shunting across the interatrial communication.The determinants ofthe amount of shunting are the defect size and theventricles relative resistance to inflow. In MS, the restricted inflow leads to increased diastolic pressures in the left ventricle.This resulted in marked accentuation of the left-to-rightshunt.

We reported a case of a 34 years old female, first came to Harapan Kita National Cardiac Center, in 2010. She was diagnosed with LS, and was planned to have ASD closure and mitral valve repair by surgery. However, she refused to undertake the procedure.

In summary, to illustrate the interactions between ASD and MS, the presence of ASD underestimated the severity of MS; meanwhile the existence of MS magnified the left to right shunt in patients with ASD.

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(J Kardiol Indones. 2012;33:99-105)

**Keywords:** Lutembacher'ssyndrome, ASD, MS, hemodynamic.

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Laporan Kasus

# Sindrom Lutembacher Sebuah Penilaian ekhokardiografi

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Sindrom Lutembacher (SL), terdiri dari Atrial Septal Defek (ASD) dan Mitral Stenosis (MS), adalah suatu bentuk kelainan jantung yang langka. Rene Lutembacher pertama kali menjabarkan sindrom ini pada tahun 1916. Saat ini, semua kombinasi dari ASD ataupun MS, baik kongenital maupun didapat, diartikan sebagai SL.

Dengan menggunakan ekhokardiografi, hemodinamik sindrom ini dapat dinilai. Secara patofisiologis, hemodinamik dari ASD tergantung dari besarnya defek dan aliran darah yang melalui komunikasi interatrial tersebut. Penentu dari jumlah aliran yang melewati defek adalah ukuran dari defek dan resistensi relatif dari aliran yang masuk ke ventrikel kiri. Pada MS, aliran masuk ke ventrikel kiri yang terestriksi menyebabkan peningkatan tekanan diastolik di ventrikel kiri. Hal ini menyebabkan peningkatan aliran dari kiri ke kanan yang berrmakna (*left to right shunt*).

Kami melaporkan sebuah kasus, seorang wanita berusia 34 tahun yang berobat ke Pusat Jantung Nasional Harapan Kita pada 2010. Pasien didiagnosis dengan SL dan ditawarkan untuk dilakukan penutupan defek ASD dan perbaikan katup mitral secara operasi. Namun, pasien menolak prosedur tersebut.

Sebagai ringkasan, untuk menggambarkan interaksi antara ASD dan MS, kehadiran dari ASD meringankan gejala yang ditimbulkan oleh MS; sementara keberadaan MS akan semakin meningkatkan aliran *left to right* shunt.

(J Kardiol Indones. 2012;33:99-105)

**Kata kunci:** Sindrom Lutembacher, ASD, MS, hemodinamik**.**

## Case History

Patient first came to HKNCC in 2010 with chief complain of breathlessness since 2005 that occurred with mild activities. She had dyspnea d'effort (DOE), orthopnea (OP), and paroxysmal nocturnal dyspnea (PND). Her physical examinations in 2010 revealed a moderately-looking ill patient, with signs and symptoms of RV overload.

Her electrocardiogram (ECG) displayed atrial fibrillation(AF) with normal QRS rate, right ventricle hypertrophy (RVH), and completeright bundle branch block (RBBB).

Her transthoracic echocardiogram (TTE) in 2010 showed large secundum ASD, bidirectional shunt, moderate MS, severe Tricuspid Regurgitation (TR), mild mitral regurgitation (MR), and mild Pulmonary Hypertension (PH), and normal left ventricle (LV) and right ventricle (RV) function.

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Her cardiac catheterization in 2010 revealed a secundum ASD, mild MR, mild MS, PH with high-flow, low-resistance that was reactive to the $O_2$ test.

Management by mitral valve repair and ASD closure by surgery was offered but the patient refused to undergo the procedure.

Patient later was hospitalized in HKNCC several times with signs and symptoms of heart failure. Her latest admission was February 2012. She came with chief complaint of breathlessness since 4 days before admission.

Her physical examination exhibited a severely-looking ill patient, with BP of 100/90 mmHg, HR of 58 – 67bpm. She had anemic conjunctiva, distended JVP (5+3 cmH20), irregular first and second heart sound, Pansystolic Murmur (PSM) grade 3/6 at apex, and rhales that covered basal portion of her lung.

Her chest x-ray exhibited an enlarged heart with a cardio-thoracic ratio of 74%, normal aortic segment, enlarged pulmonary segment, and flattened cardiac waist, hard-to-distinguish cardiac apex, and congestion.

Her TTE in February 2012 demonstrated an inter atrial septal (IAS) echo gap with left to right shunt (flow ratio of 3,84), with a diameter of 3,6 cm, severe MS, mild MR e.c RHD, moderate TR, moderate PH, normal LV systolic function, global normo kinetic, and depressed RV systolic function. Figure 3 displayed her TTE.

## Discussion

Our patient was first diagnosed with LS in 2010. This rare anomaly consist of ASD and MS.[1] As a

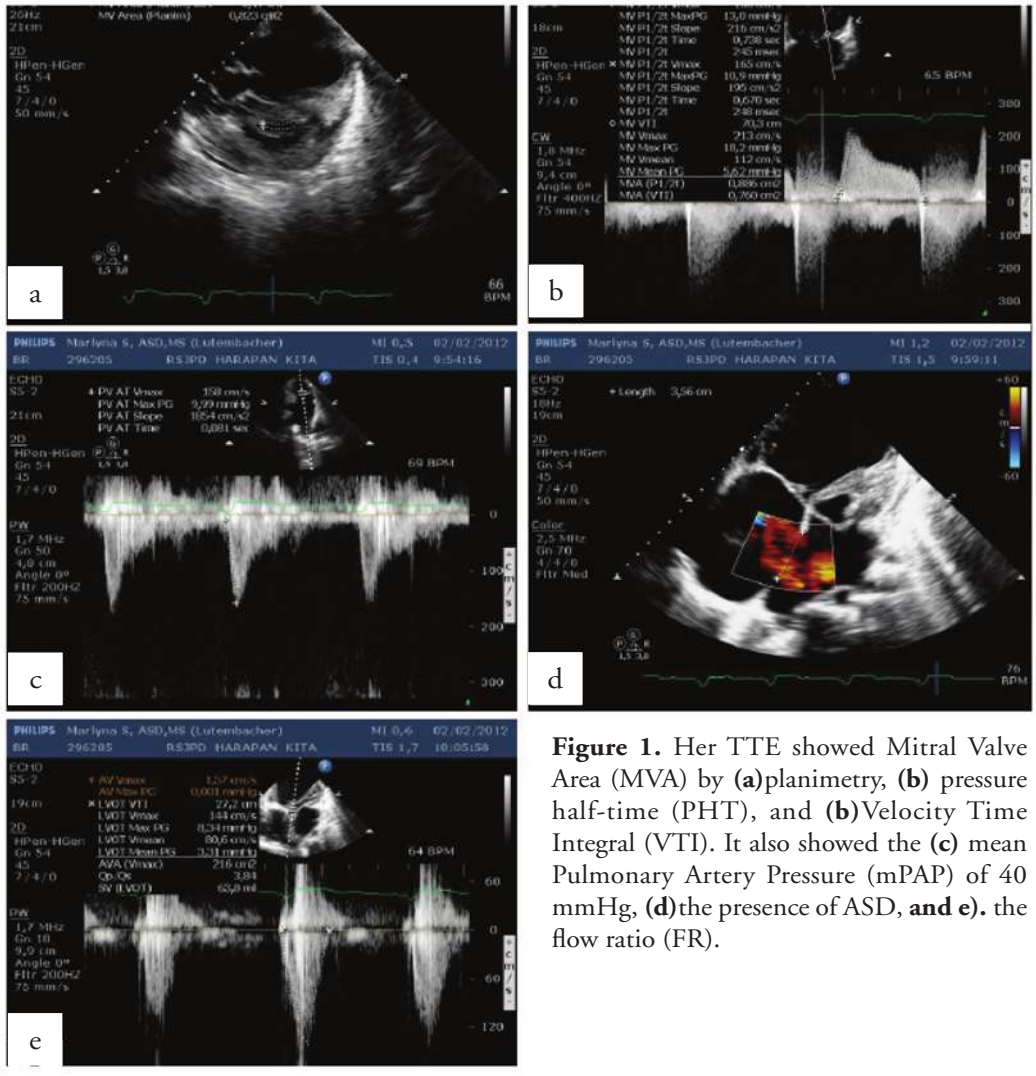


**Figure 1.** Her TTE showed Mitral Valve Area (MVA) by **(a)**planimetry, **(b)** pressure half-time (PHT), and **(b)**Velocity Time Integral (VTI). It also showed the **(c)** mean Pulmonary Artery Pressure (mPAP) of 40 mmHg, **(d)**the presence of ASD, **and e).** the flow ratio (FR).

component of LS, MScould be either congenital, as initially described, or it couldbe acquired (commonly by RHD). The true incidence rate of LS is not available yet.[2, 3] The incidenceof ASD in patients with MS was thought to be 5 out of 2.500 autopsies.[4] This syndrome could not be properly diagnosed in the past decades due to lack of echocardiographic facility.[4]

The most frequent cause of MS is chronic RHD, a complication of one or more prior episodes of acute RHD.[5] The dramatic decline in the incidence of acute rheumatic fever in most developed countries has led to a corresponding decline in the incidence and prevalence of MS. Based on HKNCC MS registry, the incidence of MS because of RHD is 76%.

Mitral stenosis may impact "proximal" cardiac chambers and blood vessels, such as the LA, pulmonary vasculature and right ventricle. For example, elevated LA pressure may lead to LA enlargement, which may predispose the patient to AF[6], as in this case. She had an enlarged LA showed by her Left Atrial Volume index (LAVi) of 76 ml/m$^2$ and AF.

Elevated LA pressure is also transmitted back to the pulmonary vasculature. Although initially reversible, these elevated pressures may become largely fixed as the pulmonary vasculature remodels, and permanent pulmonary hypertension can arise.[6] This patient also had pulmonary hypertension, with a mean Pulmonary Artery Pressure (mPAP) of 40 mmHg at rest.

As the MV orifice becomes increasingly stenotic, higher-pressure gradients are necessary to "move" blood from the LA to the LV. These gradients can be measured and used as a means to estimate the hemodynamic significance of MS.[6] Her mitral valve characteristics resembled MS in RHD, such as the calcified mitral valve leaflet, prolonged EF slope on the Parasternal Long Axis (PLAX) M-Mode view, LA dilatation, AF, Mitral Regurgitation (MR), and signs of increased of right-sided pressure and volume overload secondary to pulmonary hypertension. Those features were consistent to the literature as stated by Solomon et al.[6]

There are many different ways to assess the severity of MS[6, 7].The American Society of Echocardiography recommendations for assessing the severity of MS are categorized into two groups, specific and supportive findings, as showed in **Table 1**.[7] Each of the measurement has its own advantages and pitfalls.

Pressure gradients across the MV are determined by placing the continuous-wave (CW) Doppler probe across the MV orifice. The Doppler investigation should be performed parallel to the direction of blood flow, in order to avoid underestimating the gradient[6]. **Figure 1.a) and b)**revealed patient's MS severity. However, there are discrepancy between the severity assesed by planimetry and mean gradient. She had an MVA by planimetry of 0,8 cm$^2$ which indicated the presence of severe MS. However, the MV mean gradient was only 6 mmHg which designated as moderate MS. This discrepancy, nevertheless, was also reported in the literature. Solomon et al.[6] wrote that the basis of using mean gradient as a measurement of MS severity was that the tighter the stenosis is, the higher the pressure it needs to move the blood from LA to LV. In the existence of an ASD, though, the interatrial shunt acted as a pressure relief for the LA. Hence, the pressure in LA decreased and also the mean gradient.[6] **Table 2** summarized the pitfalls that are associated with assessing the severity of MS based on mean gradient.

Her MS severity was also assessed by using PHT. The PHT is the time for the pressure gradient across the mitral valve to decrease by half.[6] The valve is

**Table 1**. Sums up the American Society of Echocardiography recommendations for classification of mitral stenosis severity.

| | Mild | Moderate | Severe |
|---|---|---|---|
| Specific findings | | | |
| Valve area (cm$^2$) | >1.5 | 1.0-1.5 | <1.0 |
| Supportive finding | | | |
| Mean gradient (mmHg)* | <5 | 5-10 | >10 |
| Pulmonary artery presusure (mmHg) | <30 | 30-50 | >50 |

*At heart rates between 60 and 80 born and in sinus rhythm
taken from ref no. 6

**Table 2**. Pitfalls of Using Mean Gradient to Assess the Severity of MS.

- Flow-rate dependent. e.g.. affected by volume depletion or anemia
- Low cardiac output states and bradycardia may lead to low mean pressure gradient calculations in the presence of several mitral stenosis
- Heart rate dependent, e.g., affected by exercise
- Atrial fibrillation: need to average over 5 to 10 cardiac cycles
- Doppler beam alignment dependent, especially with eccentric jets

taken from ref no. 6

measured by using the equation, MVA = 220/PHT. In the existence of ASD, nevertheless, the pressure in the LA already decreased. Hence, the measurement was underestimated. Patient had an MVA as determined by PHT of 0,9 $cm^2$. It may not be as much of a difference with the MVA that was measured by planimetry (0,8 $cm^2$). Nonetheless, had the difference was between 0,9 $cm^2$ and 1,1 $cm^2$, the management would be totally different. **Table 3**displayed the PHT pitfalls. Furthermore, patient was in AF, so it would be needed to average the PHT 5 – 10 cardiac cycles.[6]

Besides MS, patient had ASD. The type of ASD that she had is secundum ASD. Anderson et.al[8] stated that these defect are within the confines of the oval fossa. **Figure 2** displayed the defect, while **Figure 3** and **Figure 1.d)** exhibited the TTE based on the literature and the case, respectively.

The hemodynamic of ASD is relatedto the magnitude and direction of shunting of bloodacross the interatrial communication.[8] There is usually asubstantial left-to-right shunt, resulting in a high ratio ofpulmonary to systemic flow. The primary determinants ofthe amount of shunting are the size of the defect and therelative resistance to inflow, or the compliance, of the ventricles.[8] In this case, patient was presented with a large ASD with the diameter of 36 mm and the presence of severe MS.

In addition, abnormal fillingcharacteristics of the left ventricle can also influence themagnitude of the atrial shunt. Filling of the left ventricleis impaired by MS,

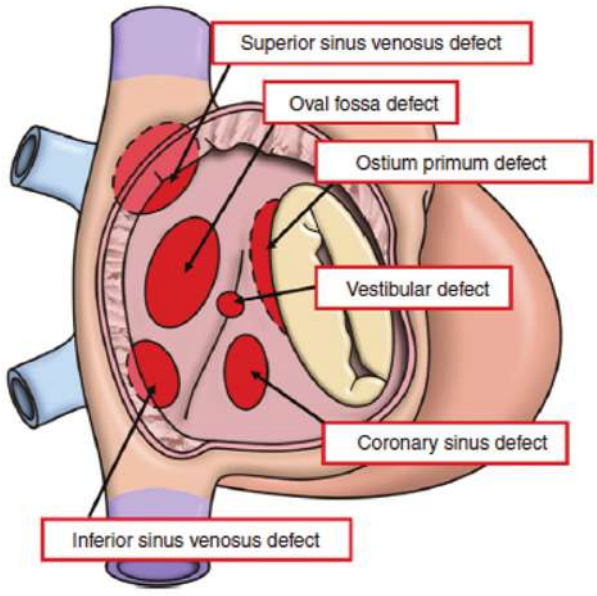


**Figure 2**. Types of ASD. Patient had secundum ASD (oval fossa defect). Taken from Ref. no.8

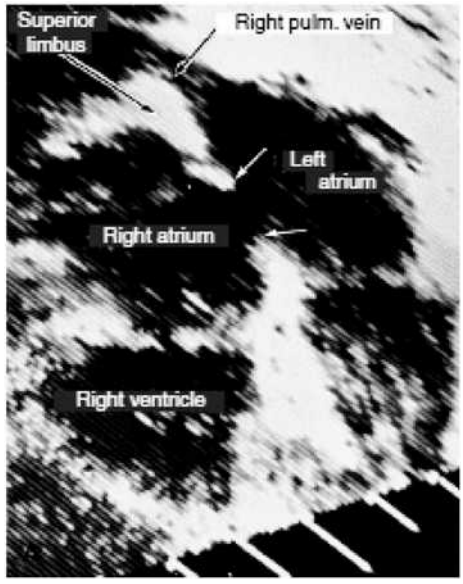


**Figure 3**. TTE of Secundum ASD, Subxyphoid View. Taken from Ref. no. 8

**Table 3**. Pitfalls of Using Mean Gradient to Assess the Severity of MS.

- Flow-rate dependent. e.g.. affected by volume depletion or anemia
- Heart rate dependent, e.g., affected by exercise
- Atrial fibrillation: need to average over 5 to 10 cardiac cycles
- Doppler beam alignment dependent, especially with eccentric jets
- Measure $P_{1/2}$ from slope with longer duration whenever deceleration slopes differ
- $P_{1/2}$ can be prolonged (i.e., increased) in nonmitral stenotic states, e.g., diastolic dysfunction, but low E peak velocities may accompany the letter
- $P_{1/2}$ method is unreliable in patient with severe aortic regurgitation or immediately post-ballon valvuloplasty
- Atrial septal defects, immediate post-mitral valvotomy, and a noncompliant left atrium shorten $P_{1/2}$. This lead to overestimation of MVA
- Changes that prolong $P_{1/2}$, e.g., a chronically dilated and overcompliant left atrium leads to underestimation of MVA

MVA, mitral valve area; $P_{1/2}$, pressure half-time
taken from ref no. 6

leads to increased diastolic pressures in the left ventricle. This may result in marked accentuation of the left-to-rightshunt across the atrial defect. [8,9,10] Furthermore, The interatrial shunt decompressesthe left atrium but increases the pulmonaryblood flow.[9, 10] So, in this case, mean gradient could not be used to calculate the severity of MS because the LA pressure is already decreased from the ASD. Furthermore, her echocardiogram proved that the trans mitral pressure gradient was only 6 mmHg, contradicted to the severity of MS which was displayed by the planimetry (0,8 $cm^2$).

Additionally, the presence of an MS in ASD augmented the left to right shunt, showed by the high mPAP (40 mmHg). The augmentation in the left to right shunt makes the flow to the PA much higher than the systemic flow. And, by using echocardiogram, the flow ratio (FR) could be measured[6]. Patient had a flow ratio of 3,8, which was consistent to the catheterization data in 2010.

Therefore, the hemodynamic effects of LS depend on two things; the severity of MS and the size of the ASD.[11] In Lutembacher syndrome, MS augments the left-torightshunt through the ASD, while the non-restrictiveASD decompresses the left atrium, reducing thediastolic mitral pressure gradient.[11] In addition, the PHT method consistently overestimated the mitralvalve area, which was evident in this case, albeit by a little. The extent of overestimation is greater inpatients with a larger atrial shunt. The hemodynamicpressure half-time is independent of the mitral valvearea, chamber compliance, and peak transmitralgradient. The hemodynamic pressure half-time isdependent on the magnitude of the atrial shunt, indicatingthat the Doppler pressure half-time method is aninaccurate measure of the mitral valve area whenever anatrial shunt coexists with MS[11].So, the best way to analyze the severity of MS is by using planimetry.[11]

In the presence of ASD, hemodynamic direction of blood flow is determined by the compliance of LV and RV. Normally, RV is more compliant than LV. In the LS, due to the presence of MS, blood flows to the right atrium through ASD without giving a backward pressure into the pulmonaryveins. So, the pulmonary congestion does not take place as it occurs in case of pure MS.[4]

Nonetheless, the magnification of the left toright shunt further makes RV overload and, eventually, RV failure.[10] This was manifested in patient's deteriorating RV function, which was indicated by the decreased Tricuspid Annular Plane Systolic Excursion (TAPSE) from 2,4 in 2010 to 1,4 in 2012.

LS is usually treated surgically.[2] By surgical procedure this condition is usually treated withmitral valve repair or replacement and with concomitant closure of the ASD.

## Summary

In summary, to put the interactions between ASD and MS into perspective, the presence of ASD underestimated the severity of MS; meanwhile the existence of MS further magnified the left to right shunt in patients with ASD. This was manifested in our patient. The severity of MS was underestimated by using mean gradient and PHT measurement, and the magnification in the left to right shunt deteriorated patient's RV function.

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