Prikaz slučaja / Case report

Lutembacherov sindrom: prikaz slučaja

Lutembacher's syndrome: a case report

Stanko Biočić, Diana Rudan*, Josip Vincelj
Klinička bolnica Dubrava, Zagreb, Hrvatska
Clinical Hospital Dubrava, Zagreb, Croatia

SAŽETAK: Lutembacherov sindrom je kombinacija kongenitalnog atrijskog septalnog defekta (ASD) i stečene mitralne stenoze (MS). Prvi put ga je opisao francuski liječnik Lutembacher 1916. godine. Ovaj sindrom je vrlo rijedak i smatra se da mu je incidencija 0.001/10,00000. Hemodinamski efekti ovog sindroma posljedica su relativnog utjecaja težine MS i veličine ASD.

Prikazujemo 54-godišnju pacijenticu kojoj je bio indiciran transtorakalni ehokardiografski pregled (TTE) zbog evaluacije prijašnjeg nalaza srednje teške MS u sklopu progresije zaduhe unatrag mjesec dana. Prije dvije godine prilikom hospitalizacije uslijed srčanog popuštanja, primjenom TTE registrirana je teška MS uz urednu sistoličku funkciju lijeve klijetke. Sada se primjenom TTE i transezofagijske ehokardiografije registrira srednje teška MS te ASD koji nije bio opisan u prijašnjim ultazvučnim nalazima pa je postavljena dijagnoza Lutembacherova sindroma.

Ovaj prikaz slučaja pokazuje kako je ehokardiografija nezaobilazna dijagnostička metoda u otkrivanju mnogih kardioloških entiteta uključujući i ovaj rijetko prisutan sindrom.

KLJUČNE RIJEČI: Lutembacherov sindrom, atrijski septalni defekt, mitralna stenoza, ehokardiografija.

SUMMARY: Lutembacher's syndrome refers to a congenital atrial septal defect (ASD) complicated by aquired mitral stenosis (MS). It was first described by Lutembacher, a French physician, in 1916. This syndrome is a very rare disease, it is found that the incidence of Lutembacher's syndrom is 0.001/10,00000. The hemodynamic effects of this syndrome are a result of the interplay between the relative effects of the ASD and MS.

We present a 54-year-old female referred to hospital for echocardiographic evaluation of previously diagnosed mild MS. She reported progression of dyspnea over the last month. Two years ago, she was admitted to another hospital because of heart failure and transthoracic echocardiography (TTE) revealed moderate MS with perserved left ventricular systolic function. We performed TTE and transesophageal echocardiogram and found severe MS with ASD that was previously unrecognised. The diagnosis of Lutembacher's syndrome was established.

This case demonstrates the presence of this rare disease in our population, but we would also like to stress the importance of the role of echocardiography in identifying many clinical syndromes including this one.

KEYWORDS: Lutembacher"s syndrome, atrial septal defect, mitral stenosis, echocardiography.

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Prikaz slučaja

Pedesetčetverogodišnja pacijentica s anamnezom preboljele reumatske groznice u djetinjstvu i arterijskom hipertenzijom u nekoliko je navrata hospitalizirana zbog zaduhe u sklopu srčanog zatajivanja. Koronarografijom, učinjenom 4 godine ranije, nije dokazana koronarna bolest srca.

Sada je transtorakalna ehokardiografija (TTE) indicirana zbog pogoršanja zaduhe unazad mjesec dana. Pacijentica je afebrilna, nepravilnih otkucaja srca frekvencije oko 70/min te izmjerenog arterijskog tlaka 100/70 mmHg. Bolesnica je bila tahipnoična, ortopnoična i cijanotična. Auskultatorno se registrira holosistolički šum i dijastoličko bubnjanje u području apeksa. Na okrajinama su prisutni bilateralni edemi s palpabilnim perifernim pulzacijama.

Elektrokardiografski se bilježi atrijska fibrilacija s frekvencijom 70/min. Liječena je beta-blokatorom, diuretikom i oralnom antikoagulantnom terapijom.

Case report

A 54-year-old female with a past medical history of reumatic fever as a child and hypertension was previously hospitalized on several occasions for severe shortness of breath due to heart failure. Left side cardiac catheterisation that was done 4 years ago showed coronary arteries without stenosis.

She was referred to transthoracic echocardiography (TTE) for evaluation of progression of shortness of breath over the last month. On physical examination she was afebrile. Her blood pressure was 100/70 mmHg and pulse was irregular at a rate of 70 beats per minute. The patient was tachypneic, orthopneic and cyanotic. A grade II/IV holosystolic murmur and a diastolic rumble were heard at the cardiac apex. The examination of lower extremities revealed palpable pulses and ankle edema bilaterally.

Electrocardiogram showed atrial fibrilation and chest X-ray revealed signs of pulmonary congestion. She was treated with beta-blockers, diuretics and oral anticoagulant therapy.

Na TTE registrira se zadebljana mitralna valvula s fibrosklerotičnim prednjim zaliskom (**Slika 1**) te posljedičnom teškom mitralnom stenozom (MS) s mitralnom areom od 0,8 cm^2 (**Slika 2**) uz blagu mitralnu insuficijenciju. Maksimalni transvalvularni gradijent u dijastoli je 5 mmHg. Također se transezofagijskom ehokrdografijom (TEE) bilježi atrijski septalni defekt (ASD) tipa ostium secundum promjera 0,5x0,6 cm s areom u 3D prikazu od 0,2-0,3 cm^2 (**Slika 3**), a doplerom se potvrdi lijevo-desni spoj na nivou atrija (**Slika 4**). Postoji i umjerena plućna hipertenzija s maksimalnim tlakom u plućnoj arteriji od 50 mmHg procjenjena temljem doplerskog zapisa mlaza trikuspidne regurgitacije. Sistolička funkcija lijevog i desnog srca su u granicama normale.

Obzirom na nalaz kombinirane reumatske MS i ASD postavlja se dijagnoza Lutembacherovog sindroma.

Transthoracic echocardiography showed thickened mitral valve with fibrosclerotic changed anterior mitral leaflet (**Figure 1**). As a consenquence, there was severe mitral stenosis (MS) with mitral valve area og 0.8 cm^2 and mild mitral regurgitation (**Figure 2**). The diastolic pressure gradient was 5 mmHg. However, an ostium secundum atrial septal defect (ASD) was noted by transeophageal echocardiography (TEE), having diameter of 0.5x0.6 mm, and area of 0.2-0.3 cm^2 (**Figure 3**). Doppler echocardiography showed left to right interatrial shunt (**Figure 4**). Mild pulmonary hypertension with maximal pressure gradient in pulmonary artery of 50mmHg was estimated by using Doppler recording of the tricuspidal regurgitation jet. Left and right ventricular systolic function was normal.

On the basis of the combined finding of rheumatic mitral valve stenosis and a ASD the diagnosis of Lutembacher's syndrome was made.

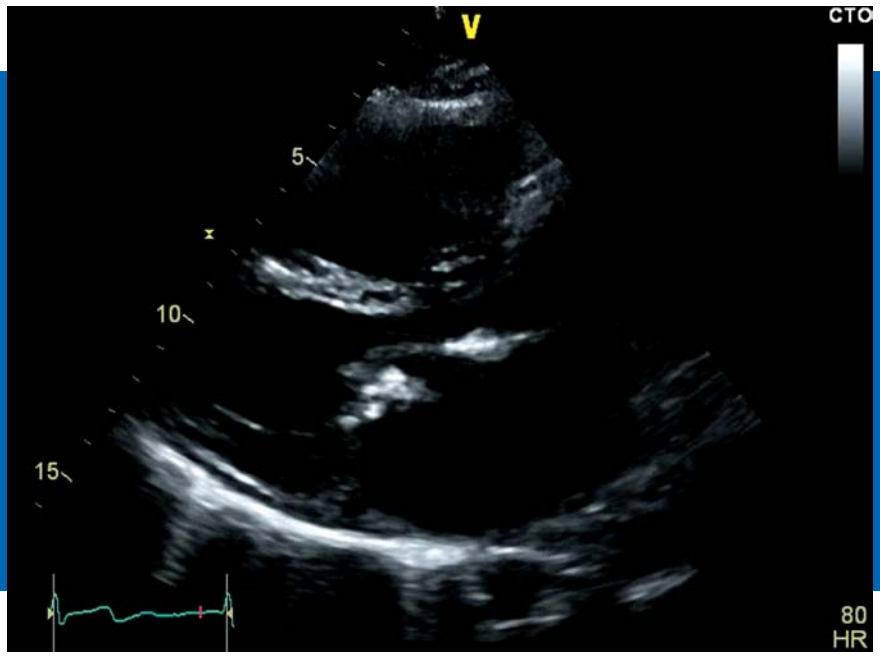

Figure 1. *Two-dimensional transthoracic echocardiography presenting fibrosclerotic mitral valve; long parasternal view.*

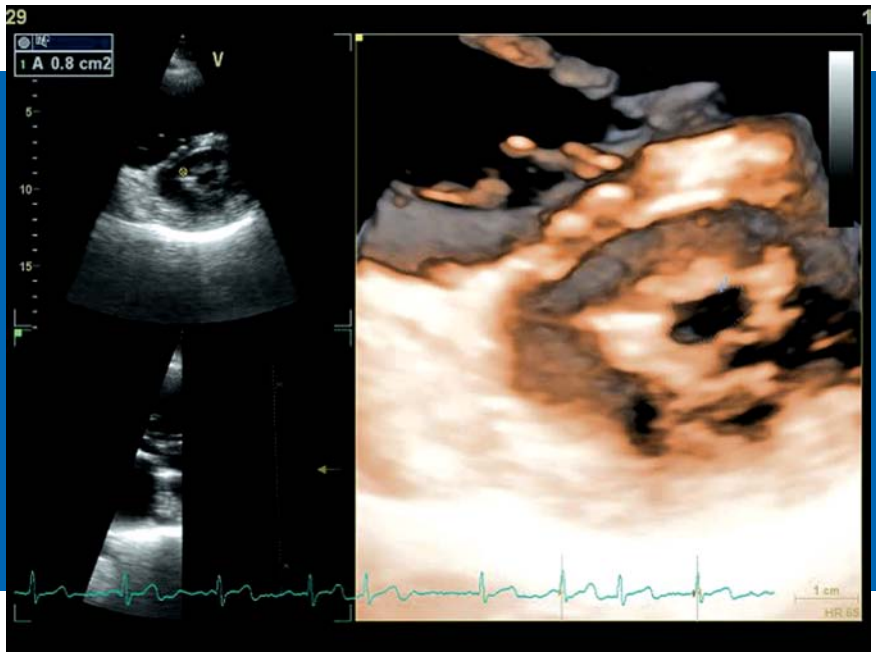

Figure 2. *Stenotic mitral valve orificium obtained by three dimensional transthoracic echocardiography.*

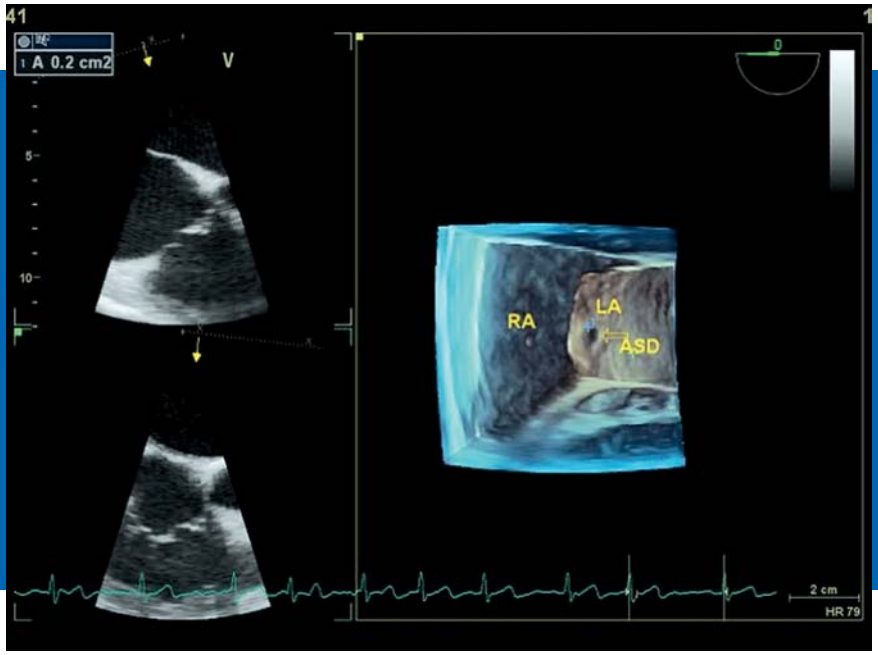

Figure 3. *Three-dimensional transesophageal echocardiography showing atrial septal defect; view from the left atrium.*

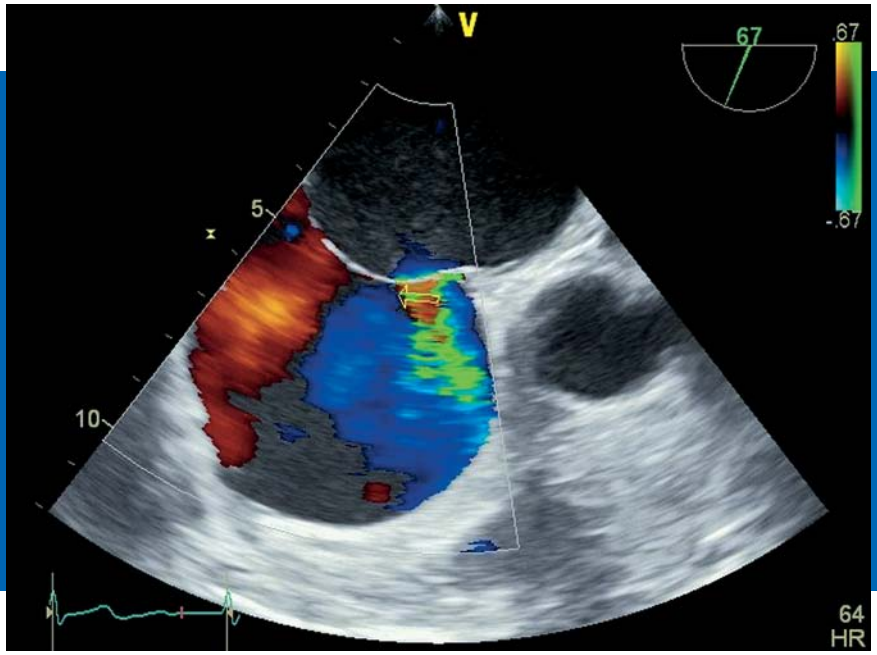

Figure 4. *Two-dimensional transesophageal echocardiography presenting left-right atrial shunt using color Doppler.*

Diskusija

Godine 1916. Lutembacher je prvi opisao kombinaciju kongenitalnog ASD i stečene MS[1]. Incidencija ovog sindroma je vrlo niska i uglavnom se javlja u žena. Incidencija MS u bolesnika s ASD je 4%, a incidencija ASD u bolesnika s MS je 0,6-0,7%[2]. Hemodinamika i sami tijek ovog sindroma ovisi o veličini ASD, težini MS, plućnoj vaskularnoj rezistenciji i prilagodljivom odgovoru desne klijetke. U slučaju teške MS i malog ASD bolesnik se uglavnom klinički prezentira slikom MS. U slučaju kada je ASD veći, kliničkom slikom dominiraju simptomi ASD. Mitralna stenoza povećava lijevo-desni shunt, dok ASD vrši dekompresiju tlačnog opterećenja lijeve pretklijetke. Treba naglasiti da smjer shunta uglavnom ovisi o odgovoru i prilagodljivosti lijeve odnosno desne klijetke. Obično desna klijetka ima bolju mogućnost prilagodbe, stoga u prisustvu MS, krv ide u desnu pretklijetku umjesto u plućne vene te je tako izbjegnuta plućna kongestija. Krajnji rezultat je progresivna dilatacija i zatajivanje desne klijetke

Discussion

In 1916, Lutembacher first described a combination of congenital ASD and acquired MS[1]. The incidence of this condition is very rare and has a predilection for females. The incidence of MS in patients with ASD is 4%, and ASD in patients with MS is 0-6-0.7%[2]. The hemodynamic features and natural history of patients with this syndrome depend on the size of ASD, severity of MS, pulmonary vascular resistence and the compliance of right ventricle. When MS is severe and ASD is small, it usually presents clinically as pure MS. On the contrary, when the ASD is large the signs and symptoms of ASD dominate. MS augments the left to right interatrial shunt, while ASD serves to decompress the left atrium. However, it should be stressed that the direction of blood flow is determined largely by the compliance of left and right ventricles. Normally, the right ventricle is more compliant than the left ventricle. In the presence of MS, blood flows to the right atrium through the ASD instead of going backward

te redukcija toka krvi u lijevu klijetku. Pojava Eisenmengerov sindroma u Lutembacherovu sindromu je izuzetno rijetka zbog prisustva velikog interatrijskog septalnog defekta i visokog tlaka u lijevom atriju zbog MS.

Klinička sumnja na postojanje ovog sindroma trebala bi biti prisutna u bolesnika s anamnezom reumatske vrućice koji imaju ASD i srčano popuštanje.

Uloga ehokardiografije (2D TTE uz obojani Doppler i konvencionalni kontrast, a osobito TTE i TEE 3D prikaz) u dijagnozi ovog sindroma je krucijalna i moguće jedina dijagnostička metoda potrebna prije intervencijske ili kiruške korekcije[3,4]. Prije je kiruška korekcija oba defekta bila jedina opcija u liječenju, ali danas, razvojem medicine i novim intervencijskim mogućnostima, oba ova defekta moguće je korigirati perkutanim transkateterskim pristupom[5-7].

Zaključno, Lutembacherov sindrom je rijetka, kompleksna kongenitalna srčana bolest. Rana dijagnoza te rano invazivno ili operativno liječenje imaju za ove bolesnike dobru prognozu, za razliku od bolesnika kojima je dijagnoza kasno postavljena i koji su razvili srčano popuštanje. Stoga naglašavamo važnost ehokardiografije u ranom otkrivanju ovog rijetkog entiteta, kako bi se pravodobno moglo terapijski djelovati i time poboljšati kvaliteta i duljina trajanja života ovih bolesnika.

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*Address for correspondence: Klinička bolnica Dubrava, Avenija Gojka Šuška 6, HR-10000 Zagreb, Croatia.

Phone: +385-1-2902-444

E-mail: drudan3@yahoo.com

into the pulmonary veins, thus avoiding pulmonary congestion. The final result is the progressive dilatation and failure of the right ventricle and reduced blood flow to the left ventricle. Eisenmenger syndrome is very uncommon in the presence of large ASD and high left atrial pressure because of MS.

Clinical suspicion of Lutembacher's syndrome should be raised by history of rheumatic heart disease, heart failure and ASD.

The role of echocardiography (2D TTE with color Doppler echocardiography, using conventional contrast technique and 3D TTE and TEE) in identifying this syndrome is well documented and it is suggested that this may be the only diagnostic technique needed before interventional or surgical correction[3,4]. Surgical correction has been previously the treatment of choice. However, nowadays, both MS and ASD, are amendable to percutaneous transcatheter intervention[5-7].

In conclusion, Lutembacher's syndrome is rare, complex, congenital heart disease. Early diagnosis and invasive or operative treatment has a good prognostic value but late diagnosis and development of heart failure bears bad prognosis.

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