

CASE PRESENTATION

Atypical presentation of Barlow's disease – a case report

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Abstract: Barlow's disease is echocardiographically characterized by thickened and prolapsed mitral leaflets due to diffuse excess of tissue caused by myxomatous degeneration. We report the case of a 65-year-old male with lumbar pain as presenting feature of Barlow's disease. The patient was admitted for dyspnoea at rest and reported acute lumbar pain before admission. Clinical examination and EKG changes were consistent with atrial fibrillation with rapid ventricular response. The abdominal-pelvic CT scan showed suggestive aspect of splenic infarction. After admission, the patient suffered an episode of transitory aphasia compatible with a transient ischaemic attack. Echocardiography revealed prolapse of both mitral leaflets, moderate-severe mitral regurgitation, LVEF of 40% and left atrial enlargement. The transesophageal echocardiography confirmed Barlow's disease and asserted there were no thrombi in the left appendage. During hospitalization and after discharge the patient received anticoagulation therapy (high molecular weight heparin followed by oral anticoagulant), beta-blocker and angiotensin-converting-enzyme inhibitor. Cardioversion was performed after one month and the patient remained in sinus rhythm during two years follow-up. After cardioversion the LVEF and LV dimensions normalised. The therapeutic approach is mitral valve repair which was temporized due to reversed systolic dysfunction and NYHA class I symptoms. The particularity of the case consists in the presentation with lumbar pain and the reversibility of LV dysfunction to sinus rhythm restoration, proving tachycardiomyopathy.

Keywords: Barlow's disease, atrial fibrillation, mitral regurgitation, splenic infarction, heart failure, transient ischaemic attack.

Rezumat: Boala Barlow este caracterizată ecocardiografic de îngroşarea şi prolabarea foiţelor mitrale din cauza excesului de ţesut rezultat din degenerescenta mixomatoasă. Raportăm cazul unui bărbat de 65 de ani care prezintă durere lombară ca prim simptom al bolii. Pacientul a fost internat pentru dispnee de repaus şi a relatat durere lombară anterior internării. Examenul clinic şi evaluarea EKG au evidenţiat fibrilaţie atrială cu răspuns ventricular rapid. Evaluarea CT abdomino-pelvină a evidenţiat prezenţa unui infarct splenic. Pe parcursul internării pacientul a dezvoltat un episod de afazie compatibil cu un accident ischemic tranzitor. Ecocardiografia a arătat prolaps al ambelor foiţe mitrale, regurgitare mitrală moderat-severă, FEVS de 40% şi dilatare atrială stângă. Ecocardiografia transesofagiană a confirmat boala Barlow şi a evidenţiat lipsa trombilor auriculari. Pe durata spitalizării şi după externare, pacientul a primit terapie anticoagulantă (heparină cu greutate moleculară mare, urmată de anticoagulante orale), beta-blocante şi inhibitori ai enzimei de conversie. Cardioversia s-a realizat după o lună, iar pacientul a rămas în ritm sinus al timp de doi ani la reexaminare. FEVS şi dimensiunile VS s-au normalizat după cardioversie. Conduita terapeutică implică plastie de valvă mitrală, care a fost temporizată datorită reversibilităţii disfuncţiei sistolice şi a simptomelor de IC clasa NYHA I. Particularitatea acestui caz constă în prezentarea cu durere lombară şi reversibilitatea disfuncţiei ventriculare stângi după restaurarea ritmului sinus al, dovadă a tahicardiomiopatiei.

Cuvinte cheie: boala Barlow, fibrilaţie atrială, regurgitare mitrală, infarct splenic, insuficienţă cardiacă, accident ischemic tranzitor.

BACKGROUND

Degenerative mitral valve regurgitation or Barlow's disease is characterized by diffuse excess tissue caused by myxomatous degeneration^{1,2}. The mitral valve leaflets are thickened, while annular dilatation with

varying degrees of annular calcification and lengthening of the chordae tendineae may be observed. The valve is generally enlarged (especially the posterior leaflet, which can reach the same dimensions as the opposite leaflet) and multiple segments are usually affected^{3,4}. These changes generate mitral valve prolapse in the

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left atrium beyond the mitral annulus with leaflet mal-coaptation which can result in varying degrees of mitral regurgitation^{5,7}. It occurs in approximately 2-3% of the population and is equally distributed between men and women, irrespective of race⁸.

CASE PRESENTATION

M.I., a 65-year-old male, was admitted to hospital for one week history of dyspnoea at minimal effort and at rest. The patient reported intense left lumbar pain accompanied by diaphoresis occurring three weeks before admission. The diagnosis at that time was renal colic – and he received NSAIDs and antispasmodics. He also reported a transient episode of intermittent claudication with the duration of 2-3 hours.

Clinical exam revealed slightly diminished murmur and pulmonary rales bilaterally in the bases, rapid irregular heart sounds with a heart rate of 150/minute and mid-systolic click followed by end-systolic murmur at the apex, while peripheral pulses were palpable bilaterally. Blood pressure (BP) at arrival was 140 over 75 mmHg. An examination of his renal system was unremarkable with bilaterally absent Giordano maneuver.

The EKG changes were consistent with atrial fibrillation with rapid ventricular rate of response (Figure 1). Standard blood test results were within normal limits, except for mild leukocytosis. Urine tests were normal and the urine cultures came negative.

Echocardiography – performed after obtaining heart rate control – showed prolapse of both mitral valve leaflets with moderate-severe mitral regurgitation (eccentric jet reaching the posterior wall of the left atrium), severe left atrial enlargement (46/59/50 mm), left atrium volume of 91 ml/sqm and atrial fibrillation.

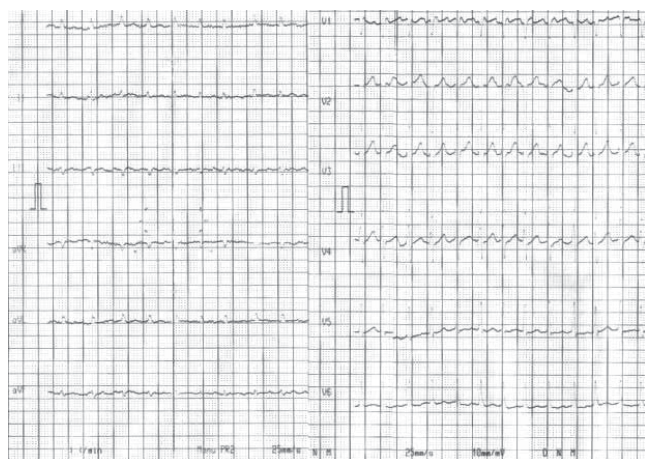


Figure 1. Electrocardiogram - atrial fibrillation with rapid rate of ventricular response; heart rate of 150/minute.

on. There was also ultrasound evidence of moderate systolic dysfunction (left ventricular ejection fraction (LVEF) of 40%, left ventricular end-diastolic volume (LVEDV) of 160 ml, left ventricular end-systolic volume (LVESV) of 93 ml and left ventricular end-systolic diameter (LVESD) of 55 mm) due to left ventricle (LV) diffuse kinetics modifications (Figure 2 and Figure 3).

An abdominal-pelvic CT scan found a hypodense region (49/21mm) in the inferior splenic pole suggestive of infarction and minor bilateral pleural effusion. There were no calculi in the urinary tract (Figure 4).

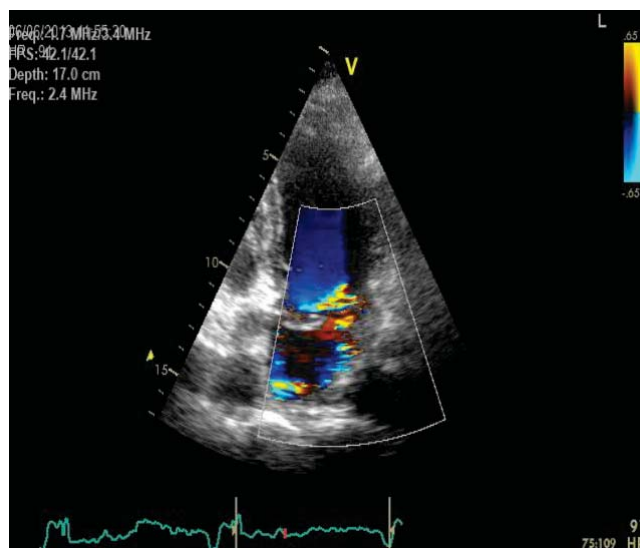


Figure 2. Transthoracic echocardiography apical four chamber view – moderate severe mitral regurgitation, severe left atrial enlargement.

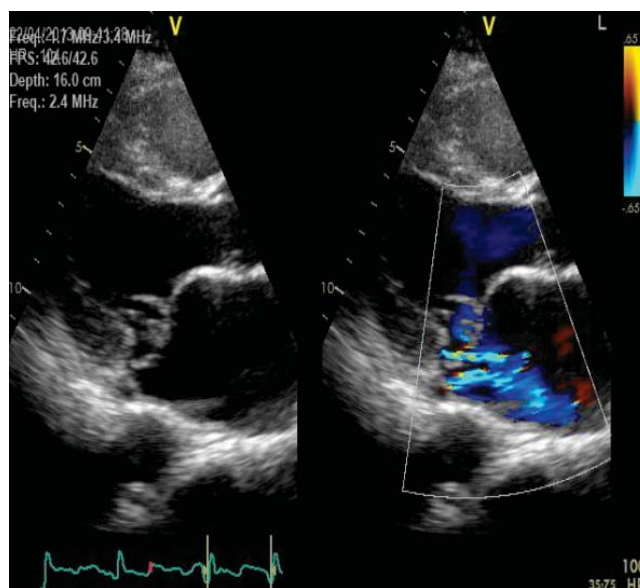


Figure 3. Transthoracic echocardiography parasternal long axis view – myxomatous degeneration and prolapse of both anterior and posterior mitral leaflets, moderate severe mitral regurgitation.

The patient was started on anticoagulation therapy with i.v. sodic heparin followed by oral anticoagulants to maintain international normalized ratio (INR) 2-3. He also received metoprolol succinate titrated as to achieve a heart rate at rest of about 80 bpm and ramipril as basic treatment for his heart failure.

Under treatment with oral anticoagulants, at an INR of 2.3, he suffered an episode of aphasia. The neurologic exam diagnosed a left Sylvian stroke and recommended urgent CT that asserted the absence of intracerebral hemorrhage (Figure 5). The aphasia resolved spontaneously and the patient was continued on heparin and then switched to oral anticoagulants at an INR between 3 and 4.



Figure 4. Abdominal pelvic CT scan – red arrow: hypodense region – splenic infarction, green arrows: minor bilateral pleural effusion.

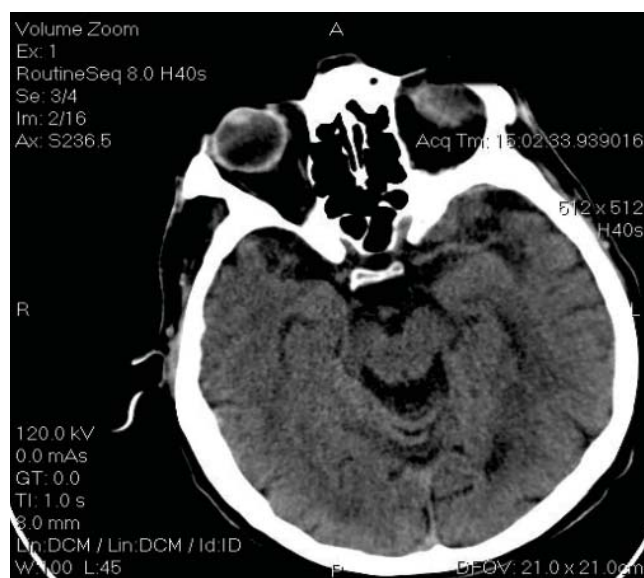


Figure 5. Cerebral CT scan - absence of intracerebral hemorrhage.

The transesophageal echocardiography (TEE) showed a suggestive aspect for Barlow's disease: thickening of the leaflets with diffuse tissue excess, multi-segment, generalised prolapse of different grades of all scallops of both mitral leaflets, exceeding the mitral annulus plan in systole (Figure 6). The left atrium was severely dilated with moderate spontaneous contrast, but without thrombi in the left appendage– after one week of sodic heparin therapy (Figure 7). The mitral regurgitation was considered moderate-severe – PISA radius of 8.3 mm, effective regurgitant orifice area (EROA) of 0.23 cm², regurgitant volume (RV) of 31 ml (probably underestimated due to the eccentric regurgitation) (Figure 8, Figure 9). Doppler ultrasound revealed normal carotid and femoral arteries.

The final diagnosis is Barlow's disease with moderate-severe mitral regurgitation complicated with mild left ventricular systolic dysfunction and left atrial dilatation, atrial fibrillation, splenic and cerebral embolisms.



Figure 6. Transesophageal echocardiography, bicommisural view –multi-segmentar prolapse of mitral valve.

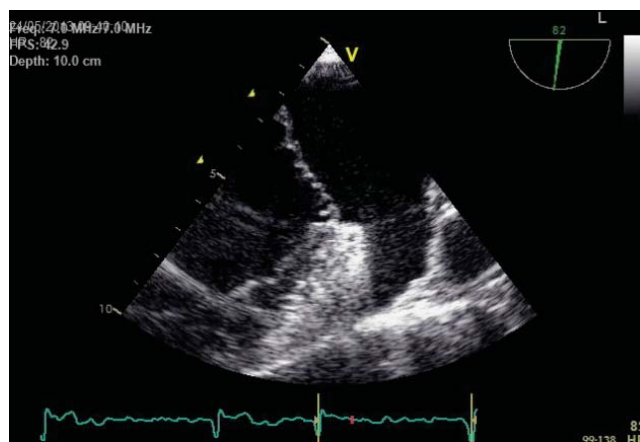


Figure 7. Transesophageal echocardiography focusing on the left appendage: no spontaneous contrast or thrombi.

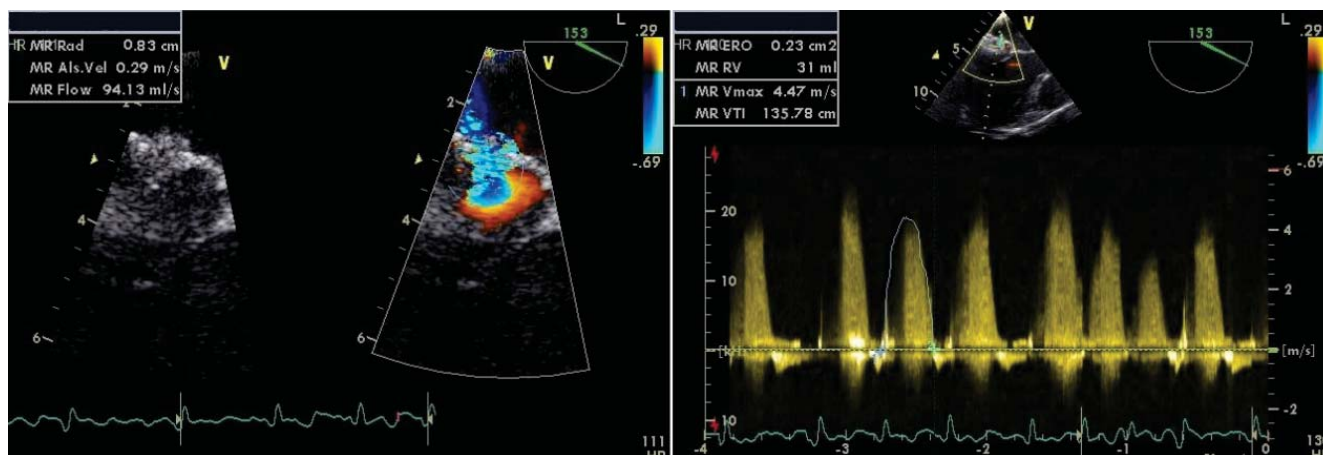


Figure 8. Transesophageal echocardiography for measuring quantitative parameters for MR – 8 A and 8 B moderate severe mitral regurgitation PISA radius=8.3 mm, EROA 0.23 cm², RV 31 ml.

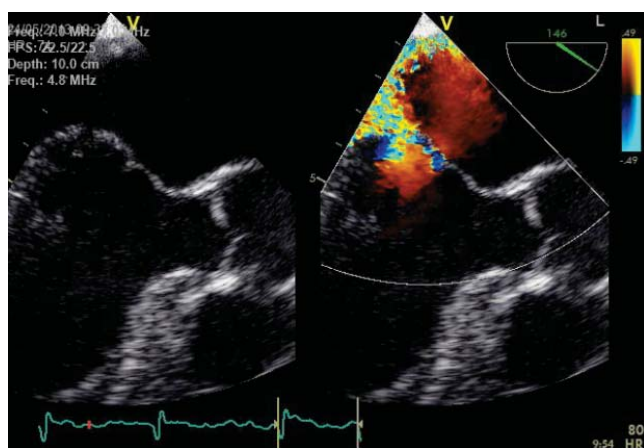


Figure 9. Transesophageal echocardiography, long axis view with colour Doppler - important mitral regurgitation with large eccentric jet and Coanda effect.

During the hospitalization, under treatment with anticoagulation therapy (high molecular weight heparin followed by oral anticoagulant), beta blocker and angiotensin converting enzyme inhibitor the patient

had a favorable evolution: eupnoea, heart rate control (90 bpm) and BP of 100/60 mmHg, efficient anticoagulation and complete remission of the neurological symptoms. The patient was discharged with NYHA class I symptoms.

After discharge, he received ramipril 5 mg od, metoprolol succinate 100 mg od and acenocumarol to maintain an of INR 3-4 in order to achieve a better prevention for embolisms, since the transient ischaemic attack (TIA) occurred at an INR of 2.3. After one month of oral anticoagulation, a new TEE was performed to ensure the safety of the cardioversion. The investigation confirmed lack of thrombi in the left atrium and appendage and cardioversion was performed successfully. No antiarrhythmic was added to metoprolol since it was the first documented atrial fibrillation episode in this patient. INR was kept to 3-4 another month after cardioversion and then it was lowered to 2-3 indefinitely. LVEF normalized at one month after cardioversion (Figure 10), while the quantitative para-



Figure 10. Transthoracic echocardiography – improved systolic function LVEF=65%, LVEDV 156 ml, LVESV 55 ml, LVESD=37 mm.

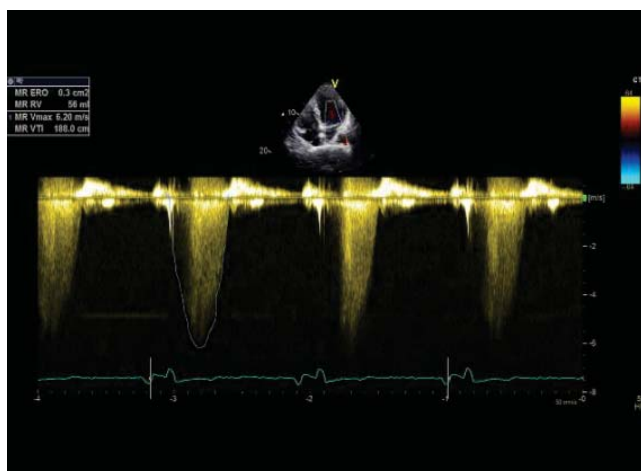


Figure 11. Transthoracic echocardiography apical four chamber view for measuring quantitative parameters for mitral regurgitation – I I A and I I B moderate severe mitral regurgitation PISA radius=10 mm, EROA 0.3 cm², RV 56 ml.

meters for mitral regurgitation modified – PISA radius of 10 mm, EROA 0.3 of cm², RV of 56 ml (Figure 11). During two years follow-up (clinical exam, echocardiography and ECG Holter every six months), the patient remained in sinus rhythm and presented one episode of paroxysmal atrial fibrillation in extreme stress conditions (a burglar trying to enter his house).

DISCUSSION

For this patient, the presenting feature was lumbar pain which proved to be consecutive to a splenic infarction. In addition, he had complained of a transiently occurring intermittent claudication. No clinical or echo Doppler signs of peripheral artery disease were found at admission and both manifestations were considered to be of cardio-embolic origin, taking into consideration the patient's heart condition (mitral valve prolapse and atrial fibrillation) as well as the normal aorta and peripheral arteries. Another particularity of the case was the TIA occurring under correct anticoagulation during the patient's admission. Whether this was caused by the embolism from the faulty surfaced mitral valve leaflets^{9,10} or by thromboembolism from the left atrium has been debatable. In the end, we consider the TIA to be thromboembolic, even if the TEE showed absence of thrombi, since the TEE was performed after the TIA had occurred. In our opinion the presence of rapidly occurring multiple embolisms and the excellent response on the long time under anticoagulant therapy makes embolism with valve material unlikely.

Medical treatment indications are for symptomatic patients to relieve symptomatology and prevent

complications. They include beta-blockers for rate control and oral anticoagulation with INR value at 2-3 for recurrent embolisms. Surgical therapy targets exclusively patients with mitral valve prolapse and mitral regurgitation and follows the European Society of Cardiology guidelines (2012) – symptomatic patients with a LVEF over 30% and LVESD under 55 mm and asymptomatic patients with LV dysfunction (LVESD ≥ 45 mm and/or LVEF $\leq 60\%$) have a class I level of indication for mitral repair¹¹. This technique is preferred to mitral valve replacement because the native valve is kept and the patient is spared of the risks of chronic anticoagulation¹¹⁻¹⁴. In this case, although initially our patient respected the criteria for mitral valve repair recommendation – symptomatic heart failure, LVEF of 40% and LVESD of 55 mm, due to the recent TIA we decided to initiate conservative treatment with beta blockers and angiotensin converting enzyme inhibitors for rate control and heart failure, anticoagulation with sodic heparin followed by oral anticoagulants for embolism prevention, while a cardioversion was planned at one month. The patient's condition improved under treatment and the he was discharged with NYHA class I symptomatology. After cardioversion, the patient's LVEF and LV dimensions normalized at one month. This feature of LVEF normalization after cardioversion – and not after rate control - indicates a component of arrhythmia-induced cardiomyopathy, a potentially reversible condition after the treatment of the arrhythmia^{15,16}. The patient's surgical indication had changed, as the LV volumes and function became normal. Theoretically, with atrial fibrillation mitral valve surgical repair is a Class IIa indication. After discussing this option with the patient, we decided to continue conservatively with a close follow-up. The patient was NYHA I, with normal LV volumes and function and he remained in sinus rhythm during two year follow-up.

CONCLUSIONS

The first symptoms, described as a renal colic and intermittent claudication were misleading. The CT scan revealed a splenic infarction, and echo Doppler for peripheral arteries was normal. In correlation with the patient's heart condition, it was suggestive for cardio-embolisms. A correct diagnosis algorithm led to the final conclusion of Barlow's disease with moderate severe mitral regurgitation, mild LV dysfunction, atrial fibrillation and multiple systemic embolisms (splenic, cerebral and lower limbs). The therapeutic approach of the disease is mitral valve repair which was tempo-

rized due to reversed systolic dysfunction and NYHA class I symptoms. The particularity of the case consists in the atypical clinical presentation and the presence of tachycardiomyopathy as the cause of LV dysfunction in a case of Barlow disease with moderate - severe mitral regurgitation.

Conflict of interest: none declared.

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