INTRODUCTION

Non-compaction of the myocardium (left ventricular non-compaction - LVNC) is a cardiomyopathy characterized by the occurrence of numerous excessive trabeculations and intertrabecular recesses which communicate with the left ventricular (LV) cavity. In normal human hearts, the left ventricle has up to 3 prominent trabeculations and is less trabeculated than the right ventricle. The clinical presentation of this entity includes depressed systolic and diastolic function, systemic embolism and tachyarrhythmias. The mechanisms that lead to LVNC are unclear. It was suggested that the basic morphogenetic anomaly may be an arrest of normal compaction of myocardial fibers in the intratertine development, resulting in two different myocardial layers (one compacted and one non-compacted, trabeculated).

Studies in the genetics of LVNC have strongly suggested that the disease has an inheritance pattern (18% to 50% of cases are familial). The incidence is noted to be low (from literature review: 0.12/100,000 births). Nevertheless, there was a raise of prevalence of LVNC in the last years, which could be explained by better diagnostic due to more performant echocardiographic machines and new techniques.

Clinical presentation is similar to other cardiomyopathies, and includes systolic and diastolic dysfunction of the LV, tachyarrhythmias and systemic embolism. The main symptom described at first presentation is dyspnea, due to low cardiac output. Tachyarrhythmias in Wolf-Parkinson-White syndrome, ventricular tachycardias, atrio-ventricular blocks, bundle branch blocks and even sudden cardiac death have also been reported.

Transthoracic echocardiography (TTE) is the method of choice for the diagnosis of LVNC. Two-dimensional TTE, three-dimensional TTE, Doppler color method, and contrast echocardiography are all used for the diagnosis and screening of patients with LVNC.

In LVNC, the LV is thickened and has two distinct layers: one compacted, thin, towards the epicardium, and the other thicker, noncompacted, with deep trabeculations towards the endocardium, which communicate directly with the LV cavity. On
two dimensional TTE, measurements must be done in parasternal short axis view in endsystole, when the myocardium is the thickest. For positive diagnosis, the ratio between non-compacted and compacted myocardium must be above 2 (Jenni Criteria, see Table 1).

Table 1. Jenni Criteria (1999).

<table>
<thead>
<tr>
<th>Criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Absence of any other coexisting cardiac structural abnormality</td>
</tr>
<tr>
<td>Numerous, excessively prominent trabeculations and deep intertrabecular recesses</td>
</tr>
<tr>
<td>Views: parasternal short axis, and apical</td>
</tr>
<tr>
<td>Focus on a 2-layer structure</td>
</tr>
<tr>
<td>Measured in end-systole</td>
</tr>
<tr>
<td>Ratio of thick noncompacted layer to thin compacted ≥ 2</td>
</tr>
<tr>
<td>Perfused intertrabecular recesses supplied by intraventricular blood on color Doppler analysis</td>
</tr>
</tbody>
</table>

The apical and midventricular segments of lateral and inferior LV wall are affected in more than 80% of patients. Three-dimensional TTE brings us supplementary data, the trabeculations and recesses are better visualized and the distinction between compacted and noncompacted myocardium is easily demarcated.

Contrast echocardiography is being used successfully in obese patients, in patients with lung disease and poor acoustic window. This method enhances endocardial border delineation and makes the differential diagnosis with prominent normal myocardial trabeculation, hypertrophic cardiomyopathy, dilated cardiomyopathy and left ventricular apical thrombus much easier.

Cardiac magnetic resonance imaging (MRI) is also used in patients with poor acoustic window and applies the same diagnostic criteria as the echocardiography.

Genetic tests for identification of specific genes are reserved to research purpose or family screening.

There is no specific treatment for LVNC at this moment. All patients should receive treatment for heart failure: beta blocker, angiotensine converting enzyme inhibitor, diuretic. Heart transplant is the only therapeutic solution in final stages. Aspirin should also be recommended, due to the risk of thromboembolic events. Patients with chronic atrial fibrillation should be orally anticoagulated.

Treatment for specific arrhythmias consists of antiarrhythmic drugs and intracardiac defibrillator (ICD) for selected patients.

Patients with LVNC have a controversial prognosis. Some studies associate this disease with high mortality due to heart failure and sudden cardiac death. Others present a better prognosis. That could be explained by different stages of disease at the moment of diagnosis, by the severity of heart failure and the improvement achieved with invasive and noninvasive treatment methods.

**CASE REPORT**

We present the case of a 46 years old male, with no previous cardiovascular pathology. For the last 6 months he complained of dyspnea on mild physical exertion and fatigue. On physical examination he showed slightly elevated blood pressure values (BP = 150/90 mmHg); the rest of the examination was normal. Resting electrocardiogram revealed sinus rhythm, HR = 80 b/min, left bundle branch block. TTE was performed and showed a dilated LV with an end diastolic volume (LVEDV) of 216 ml and slightly impaired systolic function (EF = 40%), type 1 diastolic dysfunction, trabecular aspect of the apex and lateral wall. All of these raised the suspicion of LVNC and contrast transthoracic echocardiography was performed using SonoVue contrast agent. Two ml of this agent were injected into a peripheral vein and LV endocardium was accurately delineated. LV apical and medial walls were bold, with deep recesses that communicated freely in color Doppler with the LV cavity. (Figures 1-3)

**Figure 1.** TTE, apical 4 chambers view. Deep trabeculations can be observed at LV apex and lateral wall.

LV layers ratio was calculated from the parasternal short axis view at papillary muscle level, and the non-compacted/compacted myocardium was about 2. (Fig. 4) The left atrium and right cavities were of normal size.

Angiocoronarography was performed to exclude ischemic etiology of the dilated cardiomyopathy and revealed normal coronary arteries. Ventriculography showed clear trabeculations located at the apical and medial segments of LV lateral and inferior walls.
In this context, our recommendation was for conservative therapy. Treatment with beta blockers, angiotensin converting enzyme inhibitor, diuretic and antiplatelet agents was started, with mild improvement in symptoms and normalization of blood pressure.

We tried to make an echocardiographic screening of his family (patient has two male children, with no specific medical problems), but the patient failed to return for follow-ups.

CONCLUSION

In conclusion, complete echocardiographic evaluation in this patient allowed the noninvasive diagnosis of LVNC. Particular echocardiographic appearance demonstrated with standard echocardiography and confirmed with contrast echocardiography completed the diagnosis correctly and accurately for this rare entity of cardiomyopathy.

REFERENCES