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CASE REPORT

Left Ventricular Non-compaction Cardiomyopathy - A Case Report

Szakacs Xantus Timea, Varga Andreea*, Bocicor Andreea Elena, Tatar R, Tilea I

University of Medicine and Pharmacy Tîrgu-Mures, Romania

Background: Left non-compaction cardiomyopathy (LVNC) or "spongy myocardium" is a relatively rare primary genetic cardiomyopathy, characterized by prominent wall trabeculations and intertrabecular recesses which communicate with the ventricular cavity. It appears in isolated form or coexists with other congenital heart diseases and/or systemic abnormalities. Material and method: A 28-year-old woman was admitted with exertional dyspnoea, palpitations, non-specific chest pain and progressive fatigue on exertion. In her family history sudden cardiac-related deaths at young age are present. Cardiovascular system examination revealed tachycardia, intermittent extrabeats.The rest EKG showed sinusal tachycardia (105 bpm), negative T-waves in DII, DIII, aVF, V4-V6. Consecutive 24 hours Holter EKG monitoring revealed nonsustained ventricular tachycardia, paroxysmal atrial fibrillation, isolated ventricular extrasystoles. Echocardiography showed left ventricular systolic dysfunction (LVEF:30-35%), slight LV enlargement, normal right ventricle and small left ventricle (LV) trabeculae in the apical area. Cardiac MRI demonstrated dilated LV and the presence of the trabeculations of LV walls suggestive for non-compaction cardiomyopathy. A combined treatment for heart failure and cardiac arrhythmias was initiated with good clinical results. Patient was scheduled for an implantable cardioverter defibrillator "life-saving". Conclusions: The symptoms of heart failure and cardiac arrhythmias should be considered important in apparently healthy young patients. Besides intensive medical treatment is indicated the implantation of an ICD "life-saving" and in advanced cases heart transplantation. Even if the electrocardiographic findings are non specific for noncompaction, a complete diagnostic evaluation is important, including sophisticated imaging techniques, a screening of first-degree relatives, and an extensive clinical, and genetic appreciation by a multidisciplinary team.

Keywords: non-compaction cardiomyopathy, genetic disorder, cardiac imaging

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Introduction

Non-compaction cardiomyopathy is a rare cardiac malformation caused by a genetic disorder, characterized by prominent wall trabeculations and intertrabecular recesses that communicate with the ventricular cavity. It appears in isolated form or coexists with other congenital malformations (1). The clinical manifestations may vary; patients can be asymptomatic or can develop progressive heart failure, thromboembolic events, arrhythmias or sudden death (2). Echocardiography is the main screening method for this disease, with further use of magnetic resonance to confirm the diagnosis, especially in patients with inconclusive echocardiograms. This can also help in the differential diagnosis as well as give prognosis information. It is important to have a correct diagnosis because of the possible association with other congenital malformations and the need for long-term therapeutic management and screening of living first-degree relatives (3-5).

Case Report

We present the case of a 28-year-old woman, who was admitted in a tertiary referral center with frequent and short lasting episodes of rapid palpitations associated with shortness of breath, burning discomfort in the chest during activity and while at rest, dry cough and progressive fatigue occurring during exertion. Family medical history revealed two brothers and a first cousin with sudden death before 30 years of age and a twin sister with almost similar symptoms; first-degree relatives had not been screened. She has a personal history of viral C hepatitis since 2009 that had been treated with Interferon for 6 months without further gastroenterological follow-up. At the moment of admission physical examination showed no relevant changes: blood pressure was 130/80 mmHg, heart rate 105 bpm, respiratory rate 16 cpm. The auscultation of the heart revealed an irregular heart rhythm and no heart murmurs. Laboratory examination showed a normal blood count, the hepatitis C antibody test was positive, and the quantitative HCV RNA test by PCR detected 60,877 UI/ml (detection limit 15 UI/ml). The rest electrocardiography showed sinus rhythm, normal QRS axis and duration, negative T-waves in DII, DIII, aVF, V4-V6 leads). Consecutive Holter electrocardiography recordings (24 hours) demonstrated nonsustained ventricular tachycardia, paroxysmal atrial fibrillation and isolated ventricular extrasystoles (Figure 1).

Chest X-ray revealed an enlarged cardiac shadow. Echocardiography findings: diffuse left ventricular hypokinesis with an ejection fraction of 30%, slight left ventricle enlargement, normal right ventricle and small left ventricle trabeculae in the apical area (Figure 2).

* Correspondence to: Andreea Varga
E-mail: dr.andreeavarga@gmail.com

A cardiac magnetic resonance imaging exam was performed which confirmed the presence of trabeculae of the lateral, anterior and the apical part of left ventricle walls suggestive of left ventricle non-compaction (Figure 3).

Medical treatment was focused on the three major possible complications of the disease: heart failure, arrhythmias and systemic embolic events. The patient received treatment with beta-blockers, diuretic, ACE inhibitor,

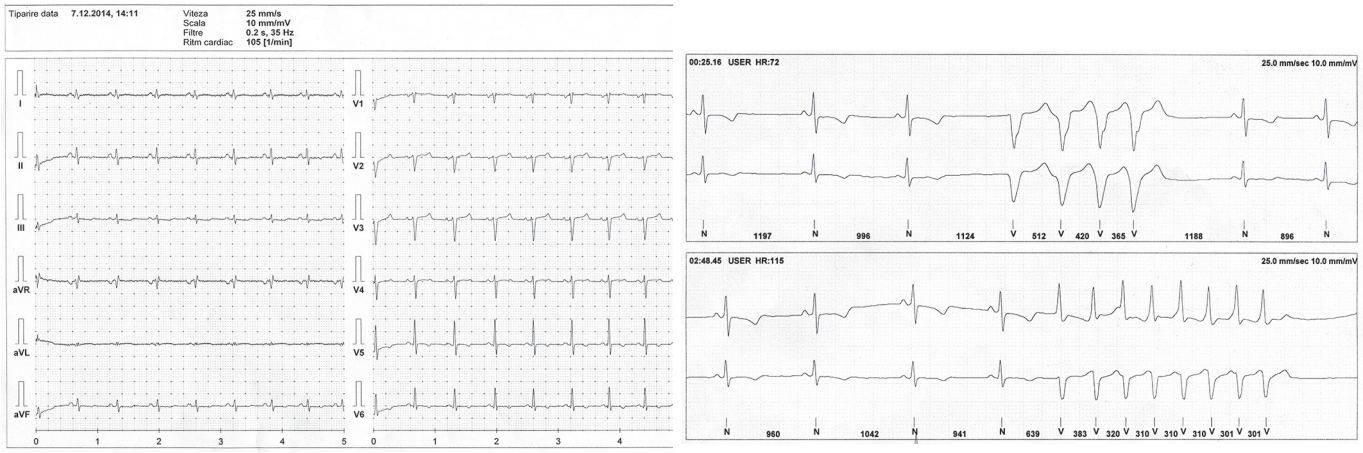

Fig. 1. Rest EKG at admission (left) and ventricular tachycardia during hospitalization (24 hours Holter recording – right)

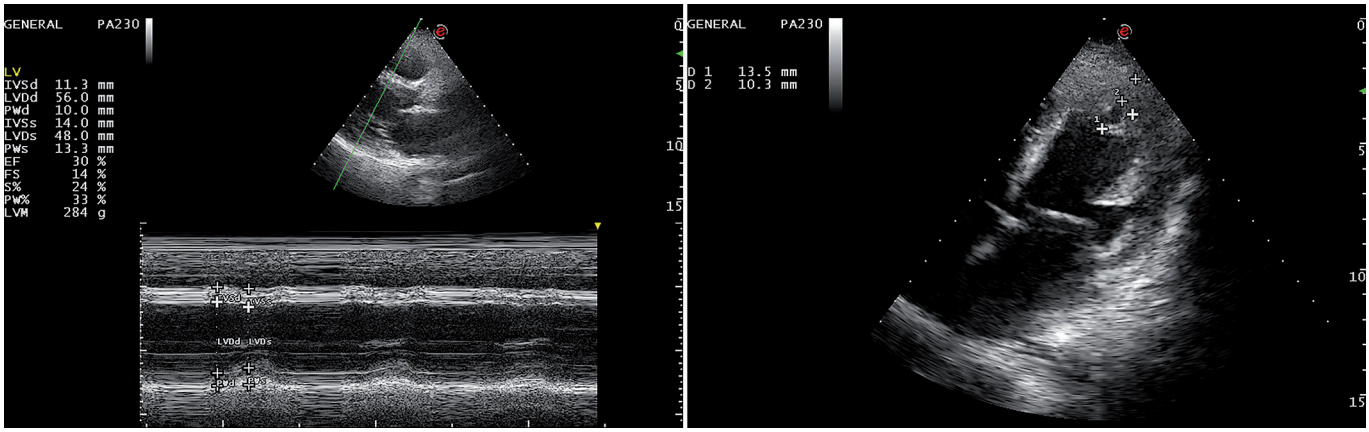

Fig. 2. Transthoracic echocardiogram with slightly dilated left ventricle, ejection fraction = 30%. Unconvincing findings for left ventricle non-compaction.

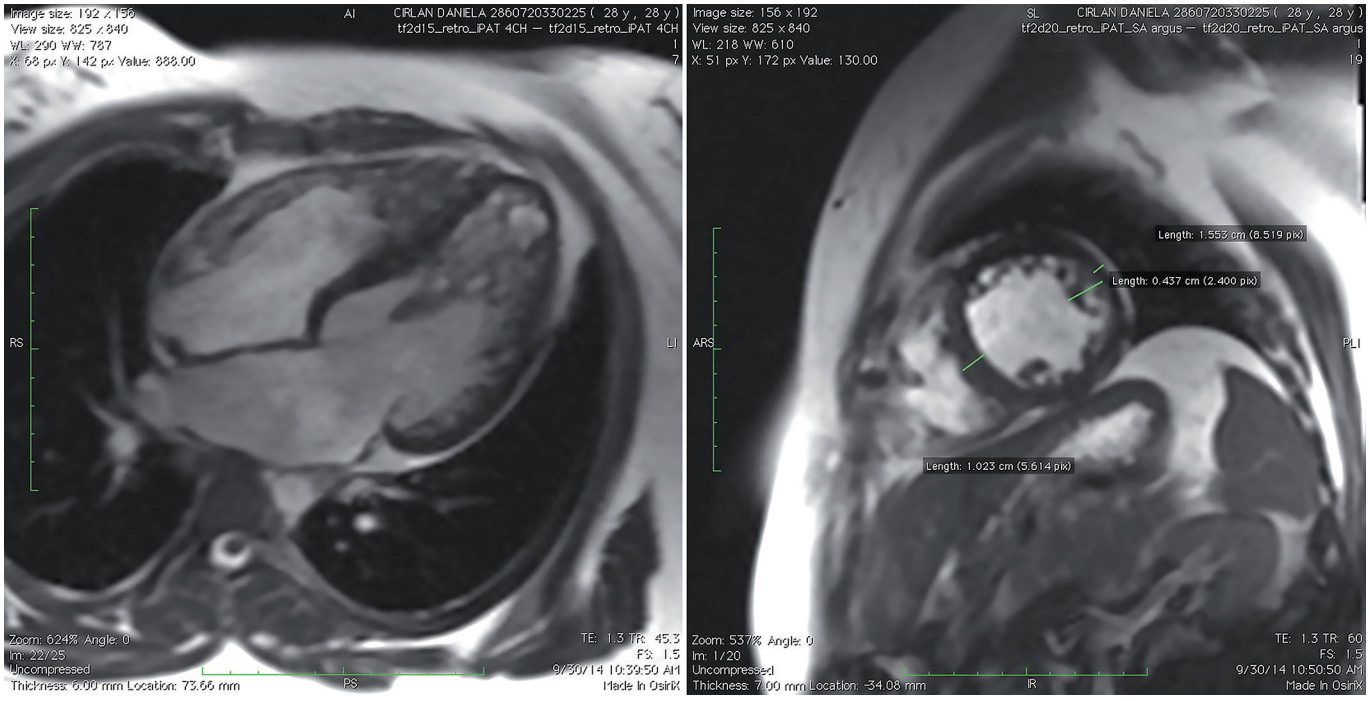

Fig. 3. Cardiac MRI exam showing left ventricular noncompaction in four chamber (left) and short-axis (right) views.

antiarrhythmics and oral anticoagulation, with good clinical results (increased tolerance on physical exertion and affirmative no palpitations) at the cardiology check-up one month later. Even if the Holter electrocardiography recording (24h) showed no ventricular tachycardia after a one-month treatment, the patient was scheduled for a "life-saving" implantable cardioverter defibrillator because of the high risk of sudden cardiac death.

Discussions and Bibliographic Review

The first reported case of isolated left ventricular non-compaction was in 1990 by Chin and colleagues (6). Isolated left ventricular non-compaction is a genetically heterogeneous congenital disorder assumed to occur as an arrest of the compaction process in early embryogenesis. It may appear in an isolated form or associated with other cardiac or non-cardiac anomalies. It is characterized by existence of trabeculation and deep recesses communication with the ventricular cavity due to non-compaction (7).

Clinically, the patients can present with heart failure, cardiac arrhythmias and thromboembolic events. Most cases are characterized by such symptoms as heart failure accompanied by dyspnoea during incrementally lowered exertion as well as orthopnoea and oedema in the lower extremities. There is a greater susceptibility to ventricular and atrial arrhythmias in isolated left ventricular non-compaction patients, while ventricular clotting is facilitated by myocardial trabeculation (7-9).

It is to be noted that while genetic testing is not advised, family members should be screened echocardiographically (10).

There have been a number of research efforts aimed at reviewing the isolated left ventricular non-compaction diagnostic criteria that rely on echocardiography and cardiac magnetic resonance imaging (11,12). Echocardiographic classification could be a factor in establishing an incorrect diagnosis, because of the inter-variability of the operators. Typical echocardiographic findings for non-compaction are: 1) prominent trabeculations and deep intertrabecular recesses; 2) two-layered structure of the endocardium; 3) colour Doppler imaging shows blood flow between these deep recesses, contiguous with the ventricular cavity (13).

Patients with inconclusive echocardiography results or an "obscured" apex benefit greatly from cardiac magnetic resonance imaging. Its high sensitivity and specific nature allows a better identification of non-compacted regions. The apex and the lateral heart walls are the locations where isolated left ventricular non-compaction related changes are the most apparent. In this case, cardiac magnetic resonance imaging revealed non-compaction myocardial alterations, confirming our diagnosis (4,11,13).

There is no targeted treatment for isolated left ventricular non-compaction; instead, it is managed based on its clinical manifestations. Yearly Holter monitoring should be undertaken to detect atrial and ventricular arrhythmias presenting with no symptoms. High-risk patients with a history of atrial fibrillation, thromboembolism and a left ventricular ejection fraction lower than 40% are recommended to be treated with chronic oral anticoagulant therapy (14). Isolated left ventricular non-compaction patients in end-stage heart failure whose standard heart failure treatment proved to be ineffective may constitute suitable candidates for a heart transplant (15), with the reservation that there are insufficient statistical data to fully support this recommendation.

High mortality and morbidity rates have been found in adults with isolated left ventricular non-compaction. If the disease is discovered in early stages, when symptoms are few or non-existent, patients will have a more favourable outcome. On the other hand, if thromboembolic events, arrhythmias and advancing heart failure are present, the prognosis becomes unfavourable (5,6,16).

Conclusions

This form of cardiomyopathy is a rare illness accompanied by major cardiac risk such as: decreased function of the left ventricle; formation of endocavitar thrombi with systemic embolization; ventricular arrhythmias, occasionally sudden cardiac death. The symptoms of heart failure and cardiac arrhythmias should be considered important in apparently healthy young patients. Besides intensive medical treatment, implantation of a "life-saving" implantable cardioverter defibrillator, and, in advanced cases, heart transplantation is indicated. In the present case, the cardiac transplant was postponed, given the presence of active hepatitis C and early-stage heart failure (NYHA functional class II). The diagnosis is based on the specific echocardiographic modifications of myocardial non-compaction, but a complete and correct diagnosis would require sophisticated imaging techniques such as cardiac magnetic resonance imaging. Screening of first-degree relatives by echocardiography and an extensive clinical and genetic appreciation would also be useful and important in detecting the illness in its early stages.

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Conflicts of interest

The authors declare no conflict of interest.

Authors contributions

All authors analysed and interpreted the patient medical recordings on the clinical presentation, and participated in equal parts in the investigation of the patient. They were responsible for the orientation of this clinical case. They were major contributors in writing and revising the manuscript. All authors read and approved the final manuscript.

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