Myocardial Noncompaction

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Left ventricular noncompaction is a primary cardiomyopathy with genetic transmission in the vast majority of autosomal dominant cases. It is characterized by the presence of excessive myocardial trabecularities that generally affect the left ventricle. In diagnosing this condition, echocardiography is the gold standard, although this method involves an increased risk of overdiagnosis and underdiagnosis. There are also uncertain cases where echocardiography is inconclusive, a multimodal approach is needed, correlating echocardiographic results with those obtained by magnetic resonance imaging. The clinical picture may range from asymptomatic patients to patients with heart failure, supraventricular or ventricular arrhythmias, thromboembolic events and even sudden cardiac death. There is no specific treatment of left ventricular noncompaction, but the treatment is aimed at preventing and treating the complications of the disease. We will present the case of a young patient with left ventricular noncompactioncardiomyopathy and highlight the essential role of transthoracic echocardiography in diagnosing this rare heart disease.

Keywords: Left ventricular noncompaction, cardiomyopathy, echocardiography

Left ventricular noncompaction cardiomyopathy (LVNC) is a primary genetic cardiomyopathy that is not fully accepted as a distinct cardiomyopathy, being still considered a congenital or acquired morphological characteristic of other different cardiomyopathies [1,2]. The European Cardiology Society includes LVNC in the unclassified cardiomyopathy group because it is not clear whether this is a separate cardiomyopathy or simply a morphological feature shared by many distinct phenotypic cardiomyopathies [3]. According to the American Cardiology Society classification, LVNC is considered as a primary cardiomyopathy. LVNC is characterized by the persistence of the fetal-type spongiform structure with intense trabecular myocardium with deep intertrabecular recesses that communicate freely with the cavity of the left ventricle but not with the coronary circulation. [4,5] In the heart of healthy people, the left ventricle has up to three prominent trabecular tracts and is less trabecular than the right ventricle [6]. The pathogenic mechanism of this pathology consists in stopping the cardiomyocyte compaction process during intrauterine life. Genetic studies conducted on patients with LVNC have highlighted the genetic transmission of this disease (18% - 50% of cases are familial) [7]. Although the incidence of LVNC is low (according to literature: 0.12 / 100.000 births) [8] in recent years there has been an increase in prevalence (0.014-1.3%), which could be explained by a better diagnosis with newechocardiography techniques [9]. Myocardial noncompaction may be isolated or sometimes associated with other congenital anomalies (septal ventricular defect (VSD), atrial septal defect (ASD), arterial canal persistence). The clinical picture is similar to that of other cardiomyopathies, with patients experiencing cardiac failure, systolic and / or diastolic dysfunction, cardio-embolic events or malignant ventricular arrhythmias. Echocardiography is the primary screening tool for this pathology, complemented by nuclear magnetic resonance to confirm the diagnosis (especially when the apex is hard to visualize).

Experimental part

We present the case of a 32-years-old patient without a known cardiovascular history presenting to the emergency room accusing dyspnea at low physical exertion and restlessness, precordial pain, volume increase of the lower limbs, physical asthenia.

The clinical examination at admission revealed the general influenced state of the patient, extremity cyanosis, normal weight, normal pulmonary stethosqual, large area of heart dullness, rhythmic heart, 100 bpm, protodiastolic gallop, grade 3 systolic murmur in the mitral area , BP = 120 / 80mmHg, bilateral pretibial edema.

The patient's biological balance on admission reveals the presence of inflammatory syndrome (C reactive protein = 2.05 mg / L, fibrinogen = 260 mg / dL), D-dimers = 2.55 μg / mL, NTproBNP = 4074 ⌈g / mL, negative myocardial necrosis markers CK-MB = 2.12 ng / mL, TnI = 0.018 ng / mL).

Electrocardiography reveals the sinus rhythm, the presence of isolated ventricular extrasystoles as well as an incomplete right bundle branch block with secondary repolarization changes (fig. 1).

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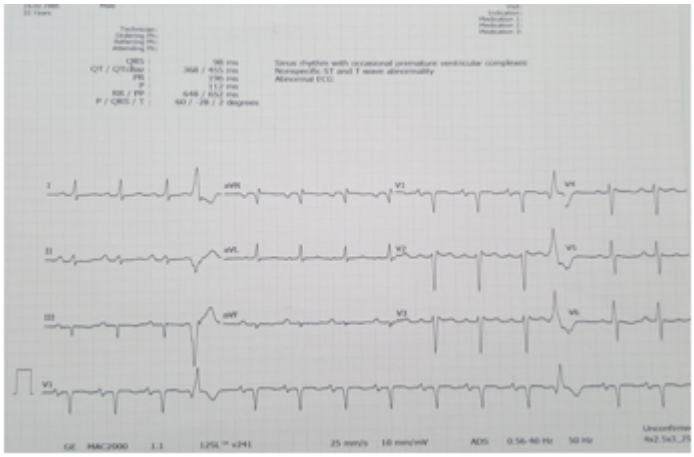

Fig. 1.Sinus rythm, 90bpm, QRS axis -10 grade, isolated ventricular extrasystoles, incomplete RBBB with secondary repolarization changes.

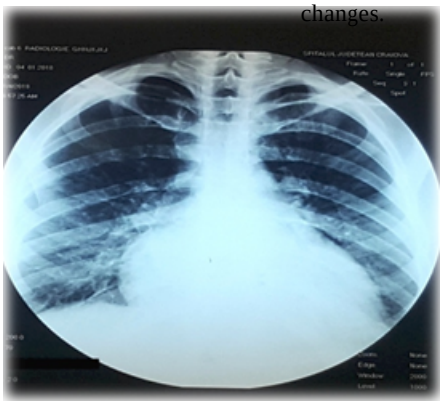

Fig. 2. Cardio-pulmonary radiography, antero-posterior incidence: large cardi-thoracic index, increased pulmonary hills with increased hilliobasal circulation

The cord-pulmonary radiography has been noted for an enlarged heart with a cardiothoracic index of 0.9, increased pulmonary hills with increased hilliobasal circulation (fig. 2).

Echocardiography revealed tetra-cameral dilatation (left ventricular end-diastolic volume of 226 mL, end-systolic left ventricle volume of 98 mL, left atrium 54 mm, right ventricle 46 mm, right atrium 50 mm), moderate mitral insufficiency, moderate tricuspid insufficiency, severe left ventricle dysfunction (15% left ventricular ejection fraction), and particularly in the lateral and posterior wall of the left ventricle, there is an exuberant trabecular apparatus with inter-triple recesses with Doppler color - suggestive of cardiomyopathy through myocardial noncompaction. Also, the presence of two layers was observed: one compact and thin epicardium and one thick endocardial, with permanent trabeculae and very deep recesses (fig. 3-8). All this raised suspicion of LVNC.

The HolterEKG revealed sinus rhythm throughout the monitoring period with frequent ventricular extrasystoles with 2 episodes of NSVT (3 consecutive ventricular extrasystoles) (fig. 9).

Coronarography was performed to exclude the ischemic etiology of dilated cardiomyopathy, highlighting permeable coronary arteries. Ventriculography revealed clear trabeculae located in the apical and medial segments of the lateral and inferior wall of LV.

Corroborating clinical and para-clinical data, the diagnosis was of:

1.DILATED CARDIOMIOPATHY BY LVNONCOMPACTION (congenital illness).

2. MODERATE MITRAL REGURGITATION BY MITRAL ANNULAR DILATATION.

3.MODERATE SECONDARY TRICUSPIDREGURGITATION.

4. NYHA FUNCTIONAL CLASS IV CHRONIC HEART FAILURE.

In this context, our recommendation was for conservative therapy. The patient followed treatment with unfractionated heparin, conversion enzyme inhibitor, beta-blocker,amiodarone, anesthetic and antialdosteronic diuretics. In view of the high risk of embolic events due to the presence of blood stasis from intertrabecular recesses,

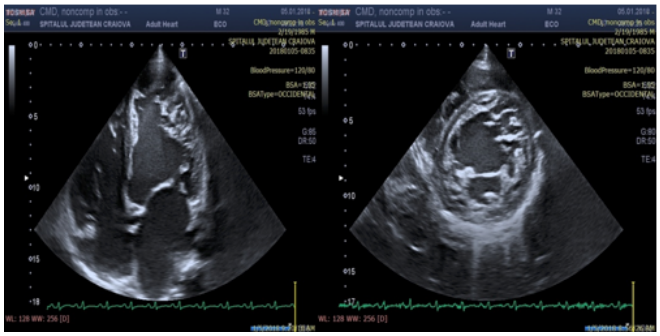

Fig. 3. Apical section 4 chambers: Trabecular apparatus on the posterior and lateral wall of the left ventricle - suggestive aspect for cardiomyopathy by compaction.

Fig. 4. Parasternal short axis incidence: presence of recesses that communicate freely with the left ventricle cavity.

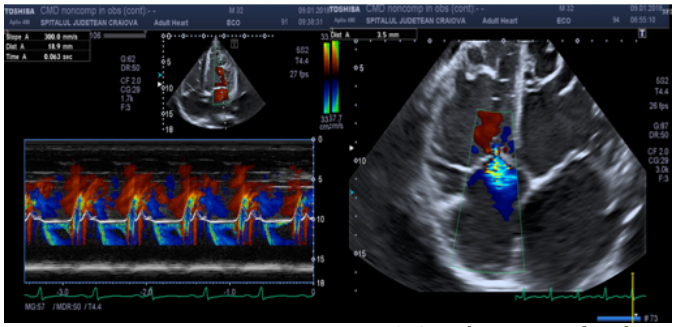

Fig. 5. Apical section 4 chambers (Color Doppler and Module M in the mitral valve): moderate mitral regurgitation

Fig. 6. Apical section 4 chambers (Color Doppler): moderate tricuspid regurgitation, vena contracta = 3.5mm

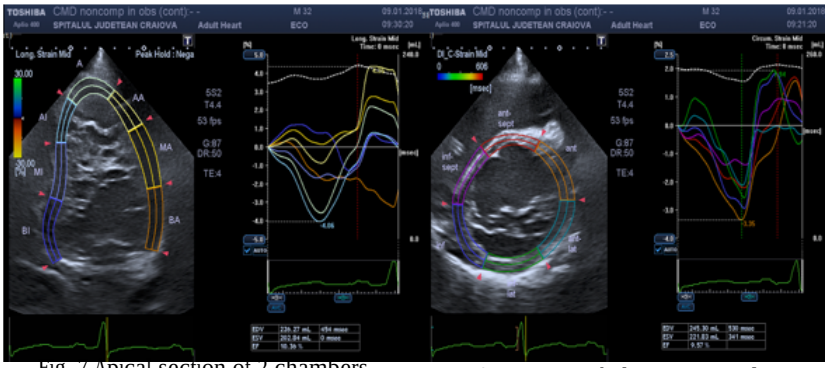

Fig. 7.Apical section of 2 chambers (speckle-tracking method): severe systolic dysfunction with LVEF 10% and GLS -4

Fig. 8. Parasternal short axis incidence (speckle-tracking method): severe systolic dysfunction with 10% FEVS and GLS -3.36

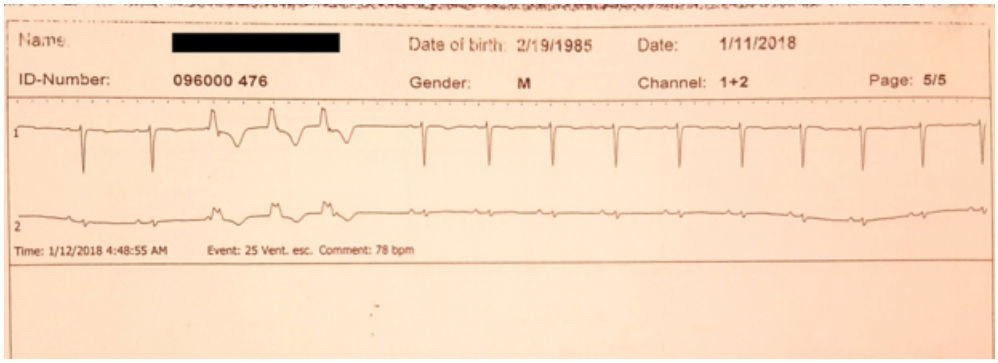

Fig. 9. ECG Holter: sinus rhythm throughout the monitoring period with frequent ventricular extrasystoles with 2 episodes of NSVT (consecutive 3VEx).

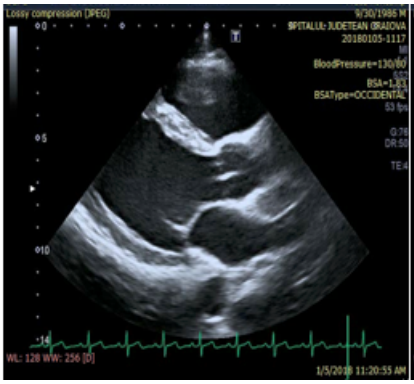

Fig. 10. Parasternal long axis incidence: normal aspect.

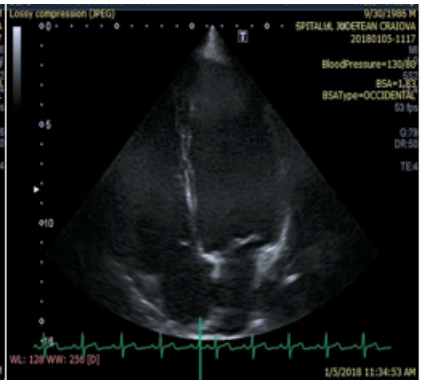

Fig. 11.Apical section 4 chambers: normal aspect

it was considered appropriate to initiate oral anti-coagulation. Evolution was favorable, switching to a lower functional class of heart failure and improved systolic function, with a re-evaluation recommendation every 6 months (echocardiography and Holter rhythm monitoring / 24 h).

Considering the possible genetic transmission of this pathology, the patient's family screening was performed. Patient's brother control echocardiography revealed a normal echocardiographic aspect without positive criteria for the diagnosis of myocardial noncompactation (fig. 10, 11). However, one year (possibly magnetic resonance) echocardiographic re-assessment and ECG Holter monitoring / 24 h for the detection of malignant ventricular arrhythmias remains necessary.

Results and discussions

Myocardial noncompaction was first described in 1932 following the autopsy of a newborn with aortic atresia andcoronary-ventricular fistula [10]. In 44% of cases, the disease is genetically transmitted [11], the genes most commonly involved in the disease being: the a-distrobrevine gene (cytoskeletal protein in the dystrophin-associated complex) that occurs in forms associated with congenital diseases and the G 4.5 gene on chromosome X, involved in the isolated form.

The clinical picture is variable, from asymptomatic patients to patients with heart failure phenomena, systolic and / or diastolic dysfunction, embolic events (from the blood stagnated at therecesses) or malignant ventricular arrhythmias (consequence of myocardial disruption).

The positive diagnosis of myocardial noncompaction is based on echocardiographic criteria, subsequently confirmed by MRI exploration where echocardiography is not conclusive [12,13]:

-The presence of at least 4 trabeculations with deep intertrabecular recesses;

-Segmental thickening of the left ventricle walls, which has two layers:one epicardial (compact and thin), and one endocardial (thick, with permanent trabeculae and very deep recesses);

-The ratio of compacted and non-compacted myocardium to be greater than 2 (measured in end-systole);

-Localization of the changes in the middle areas of the lower, lateral and apical wall;

-Highlighting the presence of blood flow in the recesses by color Doppler;

- Lack of congenital abnormalities.

Regarding treatment, there is currently no specific one for this entity, adapting it according to the clinical presentation of the patient. Light, uncomplicated forms require no treatment. In advanced forms, with cardiac insufficiency and severe systolic dysfunction, treatment is consistent with the pathology guide [14], with the indication of cardiac transplantation at the final stage of the disease. Prophylaxis of cardioembolic events by administration of oral anticoagulants as well as the reduction of the risk of malignant arrhythmias and sudden death by implantation of cardiac defibrillator (ICD) should also be considered [15,16].

The prognosis is determined by the degree of severity and rapidity of progression of cardiac insufficiency, the severity of arrhythmias and the occurrence of embolic phenomena [17]. Negative prognostic factors are: increased left ventricular end-diastolic diameter (at first patient presentation), NYHA class III or IV heart failure, presence of permanent atrial fibrillation or major left bundle branch block. As long-term management of the disease, symptomatic or high-risk patients will be evaluated every 6 months. It is also very important to inform patients about the illness, complications, risks and the need for a regular check.

Conclusions

LVNC belongs to the category of primary cardio-myopathy, with genetic transmission, rarely discussed in the differential diagnosis of heart failure. Clinical

manifestations of the disease vary in severity and include: signs and symptoms of heart failure, malignant arrhythmias, embolic events and sudden death. Echocardiography remains the gold standard in diagnosing this entity. In this case, complete echocardiographic evaluation in this patient has allowed for non-invasive LVNC diagnosis. The special echocardiographic aspect, demonstrated by standard echocardiography and confirmed with contrast echocardiography, completed the correct and accurate diagnosis for this rare cardiomyopathy entity. New approaches and imaging technologies can continue to redefine the diagnostic criteria for this unusual and distinct cardiomyopathy. Positive diagnosis remains a challenge by the presence of protruding trabecularities in the left ventricle to a normal cord [18,19]. At present, there is no specific therapy for patients with LVNC, based primarily on disease complications (treatment of heart failure, rhythm disorders, embolic prophylaxis, ICD, cardiac transplantation). The long-term prognosis of these patients could be improved by early diagnosis of this pathology, family screening and better disease management, consisting of periodic assessment and appropriate treatment. However, many questions remain about this pathology, with the need for further investigations in the future.

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