Isolated form of left ventricular myocardium noncompaction - a rare cause for ischemic stroke. Case report.

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ABSTRACT

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The article reports the case of a 32-year-old patient with no pathological history, hospitalized with a left sensory syndrome and lateral left homonymous hemianopsia which were suddenly installed in full apparent health status. Neuroimaging investigations confirmed right carotid ischemia, while all the paraclinical examinations that were conducted to determine the etiology of the stroke remained negative. The examination that allowed for the detection of a cause for cerebral ischemia was the cardiac echography, which revealed typical modifications for left ventricular noncompaction.

Key-words: left ventricular noncompaction, stroke

INTRODUCTION

Left ventricular noncompaction (LVNC) is a rare type of cardiomyopathy, characterized by the presence of an excessive trabeculation of the myocardium, which mostly affects the left ventricle. There are numerous excessive trabeculations and intertrabecular recesses which communicate with the left ventricular cavity.

Pathogenetically, the cardiomyocyte compaction process is expected to stop during embryogenesis.

Clinical aspects are similar to other types of cardiomyopathy and include systolic and diastolic dysfunctions of the left ventricle, tachyarrhythmias and systemic embolism.

CASE REPORT

In the following we report the case of a 32-year-old male patient without any personal or family pathological history who, in full apparent state of health, suddenly displayed a sight disorder that consisted in lateral left homonymous hemianopsia, associated with a sensory syndrome in the left part of the body.

Admission to our clinic was followed by a brain CT evaluation, which did not detect any damage occurring during the 24 hours after the onset. Subsequently, a brain MRI evaluation with angiographic component was carried out, which detected an area with a T_2 and FLAIR hypersignal with diffusion water restrictions on the diffusion sequences occurring on the right parietal side, parasagittal, under the aspect of a recent ischemic lesion with a normal venous sinus - fig.1,2:

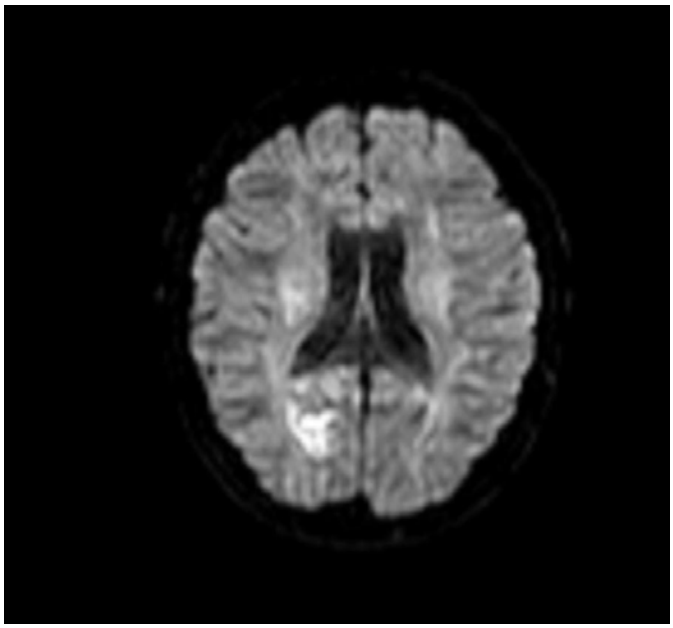

Fig.1: Area of T2 hypersignal, right parasagittal side under the aspect of recent ischemia

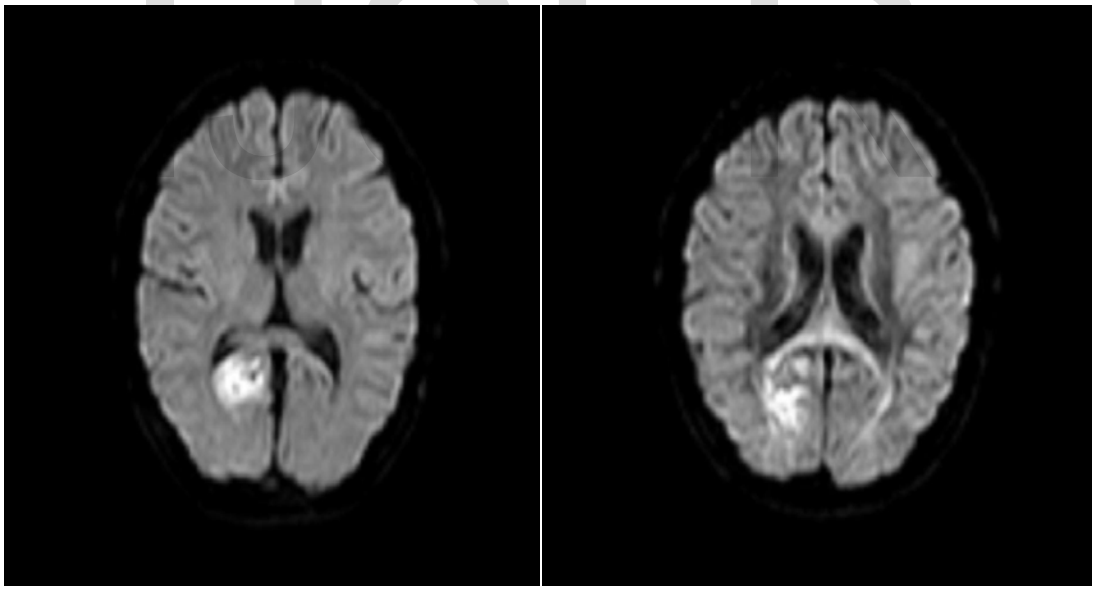

Fig.2: Right parasagittal parietal ischemia

The usual blood tests – including total cholesterol, low density lipoproteins - LDL-cholesterol, triglycerides were within normal limits. Several tests were conducted including the antiphospholipid antibody profile, cerebral vasculitis profile, collagenosis profile, thrombophilia profile, Borreliosis test, S protein

determination, C active protein resistance, homocysteinemia, without detecting any pathological modifications.

The Holter rhythm electrocardiogram scan registered over a 24-hour time lapse was also within normal limits, which allowed for the exclusion of paroxysmal rhythm disorders. Coronary arteriography evaluation was also normal.

The extracranial Doppler of cervical arteries examination revealed an age-appropriate appearance, without lesions regarding the arteries.

However, pathological changes were diagnosed during the cardiac echography exam. The examination revealed numerous trabeculations in the left ventricle apical area through which we could observe the penetration of the Doppler color signal: compact area/compact+ non-compact area, section SAX = 9/21 + 9 = 0.3. Conclusions: the left ventricle is undiluted, regarding the body surface, but excessively presenting trabeculations in the apical area, with systolic function conserved, Ejection Rate = 67%; mild diastolic dysfunction, delayed relaxation type, no significant hemodynamic valvulopathy, no pulmonary hypertension estimated by surrogate parameters, free pericardium- fig. 3:

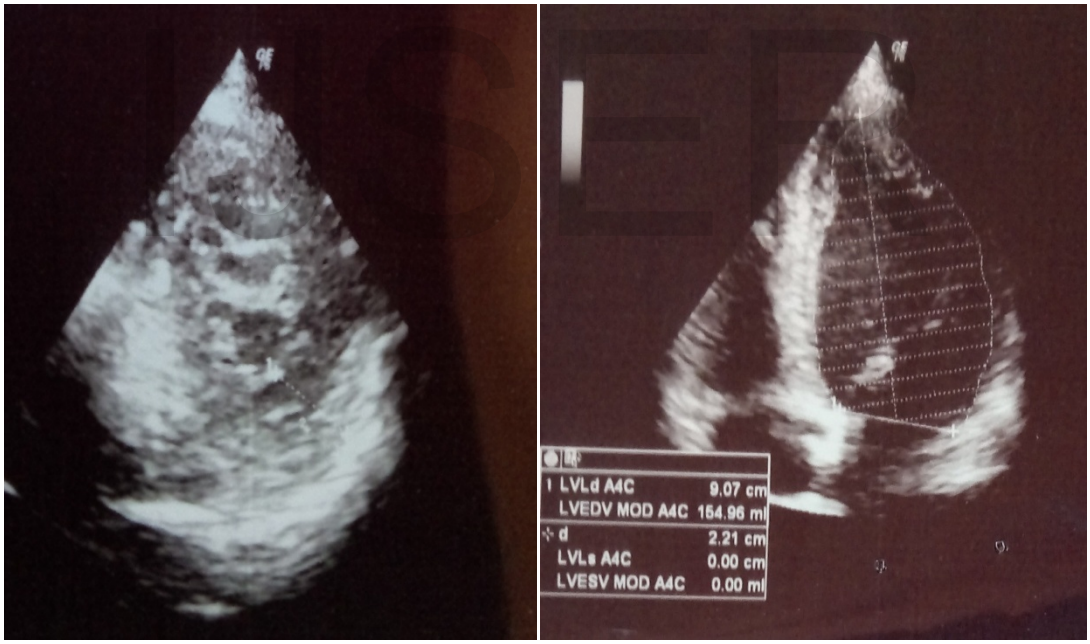

Fig. 3: Cardiac echography that shows left ventricular noncompaction

Taking into account the cardiac echography, as well as the exclusion of other etiologies for cerebral ischemia following our investigations, the parietal ischemic lesion was interpreted as representative of a cerebral embolic event, determined by the phenomenon of left ventricular noncompaction.

Such embolic events within the LVNC are rare, which is also confirmed by different studies.

Anticoagulation treatment was started. The evolution was favorable, with improvements in the visual field and on the sensory syndrome.

The patient is under neurological and cardiological supervision.

DISCUTIONS

Left ventricular noncompaction (LVNC) is a rare cardiomyopathy with an incidence of 0,05-1,3/100.000 births[1], characterized by the persistence of the fetal spongiform structure with an excessive trabeculation of the myocardium, which mostly affects the left ventricle[2,3], but possibly also the right ventricle[3].There are numerous excessive trabeculations and intertrabecular recesses, which communicate with the left ventricular cavity[2], but not with the coronary circulation. It occurs in adults as well as in children.

In the human heart, the left ventricle has up to three prominent trabeculations and it has less trabeculations than the right ventricle[4].

Two forms of the disease have been described: an isolated form and a non-isolated one, associated with other congenital diseases: ventricular septal defects, atrial septal defects, pulmonic stenosis, hypoplastic left ventricle, facial dysmorphia [5].

The American Heart Association has classified LVNC as a primary genetic type of cardiomyopathy [6], while The European Society of Cardiology considers it is unclassified type of cardiomyopathy, based on the fact that NCV may be a morphological manifestation of other severe distinct cardiomyopathies.

The mechanism leading to LVNC is unclear. LVNC is not considered a distinct cardiomyopathy, as some authors consider it is either congenital or an acquired morphological characteristic of other dilatative cardiomyopathies.

Pathogenetically, the cardiomyocyte compaction process is expected to stop during embryogenesis. It was suggested that the most important fact may be the arrest of myocardial fibers in the intrauterine development, resulting in two different myocardial layers: one compacted and one non-compacted, trabeculated[3].

It can be either familial (Barth's syndrome) [7] or secondary to congenital ventricular outflow tract obstructions. Studies of the genetic of the LVNC have strongly suggested that the disease has an inheritance pattern (18% to 50% of cases are familial) [6].

Genetics in LVNC: several studies suggest that LVNC is a genetic and heterogeneous disease with a sporadic and also a familial form, with pathogenic mutations in the genes encoding proteins such as cytoskeletal, mitochondrial,

sarcomeric and Z-line proteins. Various autosomal dominant, recessive, X-linked or mitochondrial transmissions were described[8].

New mutations can appear in sporadic cases which occur most frequently [8].

Specific genotype-phenotype association are absent in LVNC, as mutations of the same gene can not only determine LVNC, but also dilatative or hypertrophic cardiomyopathies, which limit the results of genetic testing.

Three genes were identified as correlating to NCV: dystrobrevin-alpha (DTNA), cipher/ZASP (Z-line component which is both found in the skeletal muscle as well as in the cardiac muscle), TAZ (a gene with unknown dysfunction which is involved in the X-linked dilatative cardiomyopathy) [8].

Clinical aspects are similar to othertypes of cardiomyopathy and include systolic and diastolic dysfunctions of the left ventricle, tachyarrhythmias and systemic embolism [9].

Although the asymptomatic period varies significantly, it is considered that the average duration from the onset of the symptoms to the diagnosis is three years[10].

The triad formed by symptoms of heart failure, arrhythmia and cardioembolic events is the clinical manifestation in patients with diastolic dysfunction of the left ventricle [11]. Different types of arrhythmias can occur, from atrial fibrillation - 7 – 26% to sustained ventricular tachycardia [1].

The main symptom is dyspnea, due to low cardiac output. Tachyarrhythmias in Wolf-Parkinson-White syndrome, ventricular tachycardias, atrioventricular blocks, bundle branch blocks and even sudden death have been reported [5].

As a follow up, cardiac events occurred in a number of 36 patients over a period of four years. There were five cardiac deaths, 16 heart failure hospitalizations, 10 ventricular arrhythmias and five thromboembolic events [12].

Another study made by Chin et al. on a number of 8 patients with LVNC described a cardioembolic event of a type of stroke in a 2,3-year-old child leading to his death [2].

Transitory ischemic attacks or stroke in 25% of all patients with LVNC have been reported in two series [1,2]. Considering the incidence of atrial fibrillation in these patients, anticoagulation treatment is recommended in 29% of the cases. Patients with LVNC have various prognosis. Some studies associate the disease with high mortality due to heart failure and sudden cardiac death [5]. Others patients have better prognosis [8].This depends on the stage of the disease at the moment ofdiagnosis, on the severity of the heart failure andthe improvements due to treatment.

While echocardiogram is the gold standard for paraclinical diagnosis, heart computed tomography and heart magnetic resonance are also used to detect left

ventricular trabeculations and recesses, which serve as a nidus for mural thrombus[6].

The transthoracic echocardiogram TTE (two dimensional TTE and three dimensional TTE) reveals a dilated left ventricle, with multiple intravascular trabeculations and recesses, commonly involving the apical and mid ventricular segments[13].The left ventricle has two distinct layers: one which is compacted and thin towards the epicardiumand the other, thicker, noncompacted, with deep trabeculations towards the endocardium, which can communicate directly with the left ventricular cavity. For positive diagnosis, the ratio between non-compacted and compacted myocardium must be above 2 (Jenny Criteria) [3].

In the past years, better diagnosis with more performant echocardiography led to a rise in the prevalence of LVNC [13].

There is no specific therapy for LVNC. The treatment addresses the three types of clinical manifestations: heart failure (beta-blockers, angiotensin converting enzyme inhibitors, diuretics), arrhythmias (antiarrhythmic drugs) and systemic embolic events (anticoagulation) [9].

CONCLUSIONS

1. Establishing the cause of a stroke in young people without pathological history and without vascular risk factors is challenging and requires extensive paraclinical investigations, both for finding out the etiology as well for the differential diagnosis.

2. In the clinical case of the patient we report, the essential examination was cardiac echography, this being the one that detected the LVNC, which explains the occurrence of the cerebral ischemia by means of embolic mechanism.

3. This case is clinically important due to the low incidence of LVNC cases. Only 25% out of all cases are quoted to have displayed a stroke episode which occurred through embolic mechanism during their evolution.

4. Even in the absence of associated atrial fibrillation, preventing systemic embolic recurrence involvesthe administration of anticoagulant medication.

5. Lack of familial history suggestiveof cardiomyopathies allowed for the case to be isolated as an LVNC form.

6. Both neurological and cardiological follow-up of the patient is important in order to promptly apply necessary treatment in the case of a possible complication (heart failure, atrial fibrillation).

This article does not contain any studies with human participants, performed by any of the authors.

Informed consent: the informed consent of the patient was obtained and written in the observation sheet.

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