Non-compacted myocardium in an adult with acute neurological deficit in the Emergency Department: a case report

Miocárdio não compactado em adultos com déficit neurológico agudo no Departamento de Emergência: um relato de caso

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ABSTRACT
Non-Compacted Myocardium (NCM) is an uncommon cardiac condition with a genetic predisposition, often characterized by trabeculae and distinct myocardial layers. This case report discusses a 59-year-old hypertensive, diabetic male with acute neurological symptoms. Diagnosed with an ischemic stroke, subsequent investigations revealed features indicative of NCM. Confirmatory Cardiac Magnetic Resonance Imaging (CMR) and echocardiography were pivotal for diagnosis. The patient received specialized outpatient follow-up, emphasizing the importance of early diagnosis and tailored treatment. This report contributes to the understanding of NCM's diverse clinical presentations and underscores the significance of a multidisciplinary approach for effective patient care.

Keywords: isolated non-compacted ventricular myocardium, stroke, heart failure.

RESUMO
O Miocárdio Não Compactado (MNC) é uma condição cardíaca rara com predisposição genética, caracterizada por trabéculas e camadas miocárdicas distintas. Este relato de caso descreve detalhadamente um homem de 59 anos, hipertenso e diabético, que apresentou sintomas neurológicos agudos. Após o diagnóstico de acidente vascular cerebral isquêmico, investigações subsequentes revelaram características sugestivas de MNC. Métodos diagnósticos de confirmação incluem Ressonância Magnética Cardíaca (RMC) e ecocardiografia. O paciente recebeu seguimento clínico adequado, destacando a importância do diagnóstico precoce e do tratamento individualizado. Este relato de caso apresenta um quadro clínico incomum de MNC e destaca a importância de uma abordagem multidisciplinar para o cuidado do paciente.

Palavras-chave: miocárdio não compactado ventricular, acidente vascular cerebral, insuficiência cardíaca.

1 INTRODUCTION

Non-Compacted Myocardium (NCM) is a rare condition, whose etiology is not well defined, but genetic factors seem to be associated. Genetic predisposition plays a significant role, with alterations in genes related to cardiac development potentially leading to this condition (1). Disturbances during embryonic cardiac development are also identified as potential triggers for non-compacted myocardium (2). Characterized by prominent trabeculae, deep intertrabecular recesses and myocardium composed of two distinct layers, compacted and non-compacted (1).

Recent studies suggest a prevalence of NCM of up to 43%, mainly in some individuals who present physiological adaptation due to pressure overload, such as those of African descent, athletes and pregnant women (3).

The clinical results of this condition can be quite heterogeneous, from asymptomatic patients to patients with important comorbidities such as heart failure, thromboembolism and sudden death (3). Studies in patients with CMN who developed heart failure show that the main clinical manifestations are dyspnea, palpitation, chest pain, syncope or pre-syncope and, more rarely, stroke (3).

2 CASE REPORT

Male patient, 59 years old, police officer, born in São Paulo, previously hypertensive and diabetic, practicing physical activity. He seeks the Emergency Department due to dysarthria and central facial paralysis for a few hours. Family history of mother with unknown heart disease.

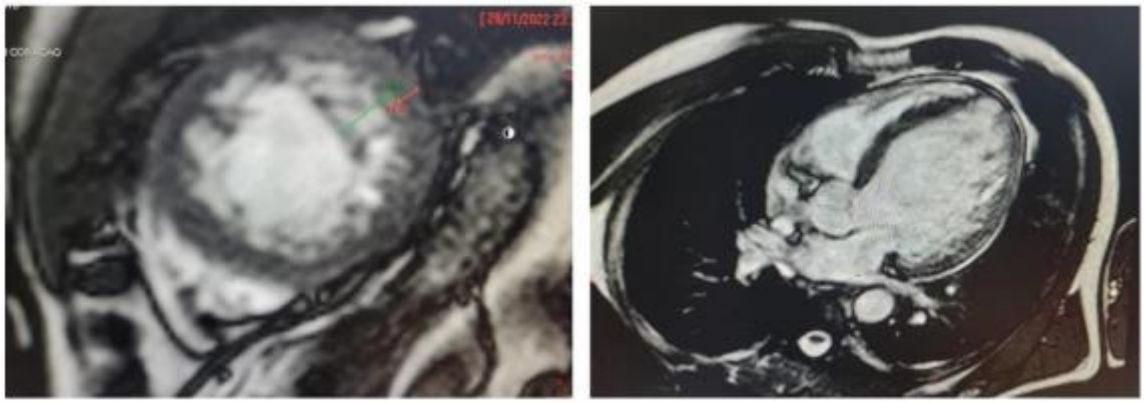
In the emergency department, vital signs were measured - without changes - and an electrocardiogram, which showed sinus rhythm and left bundle branch block. Due to neurological complaints, a head tomography was performed which showed an ischemic cerebrovascular accident (CVA). After carrying out initial measures, the patient was discharged from the emergency room to the ward and continued with the investigation of the etiology of the stroke with other complementary tests. The transthoracic echocardiogram revealed enlargement of the left chambers, eccentric hypertrophy of the left ventricle, diastolic dysfunction, diffuse hypokinesia with an ejection fraction of 36% (Simpson) and the presence of marked trabeculation.

Given the echocardiogram findings, the hypothesis of non-compacted myocardium with neurological complications was considered. The diagnosis was confirmed through Cardiac Magnetic Resonance Imaging (CMR). Other complementary exams were also performed, such

as CT angiography of coronary arteries without significant lesions, Holter monitoring with sinus rhythm, left bundle branch block and rare ventricular extrasystoles, and an electrophysiological study that did not induce ventricular arrhythmias.

During hospitalization, the patient showed improvement in the neurological deficit, remaining asymptomatic during the hospital period, including without cardiac symptoms despite the reduced ejection fraction. He was discharged from hospital for specialized outpatient follow-up, with full anticoagulation and pharmacological treatment for heart failure with reduced ejection fraction.

Figure 1: Magnetic Resonance Images of the Heart, with the presence of myocardium composed of two distinct layers, compacted and non-compacted.



Source: authors.

3 DISCUSSION

Patients with Non-Compacted Myocardium (NCM) may exhibit a range of symptoms, including asymptomatic cases, as well as presentations with heart failure, arrhythmias, and thromboembolic events, such as stroke. Currently, there is no universally accepted standard test for diagnosis, with echocardiography serving as a common diagnostic tool, and cardiac resonance imaging being employed when echocardiographic findings prove inconclusive (4).

Echocardiography is frequently selected as the initial diagnostic method to assess the presence of NCM. This method provides real-time visualization of the heart, facilitating the identification of deep trabeculations and intertrabecular recesses (4). Cardiac Magnetic Resonance Imaging (CMR) assumes a crucial role in confirming the diagnosis and characterizing the extent of NCM. CMR offers a more detailed view of myocardial architecture and distinctly identifies the compacted and non-compacted layers of the myocardium (5). A non-compacted/compacted ratio of >2.3 in diastole distinguished pathological non-compaction, with values for sensitivity, specificity, and positive and negative predictions of 86%, 99%, 75%, and 99%, respectively (6).

In specific cases, Computed Tomography (CT) can be employed to evaluate cardiac anatomy, especially when echocardiography and CMR yield inconclusive results. CT provides additional insights into heart structure, proving particularly valuable in challenging clinical scenarios where other diagnostic methods may not offer conclusive findings (3).

Regarding risk factors, it is necessary to understand the genetic basis of this condition. Genetic inheritance can determine this pathology, making a thorough assessment of family history essential to identify possible indicators. Understanding these risk factors is crucial not only for early diagnosis but also for the implementation of targeted preventive strategies based on the specific genetic characteristics of each patient (1).

The therapy aims to treat complications and prevent sudden death, in addition to reducing hospitalizations, as repeated hospital admissions for HF have a negative impact on prognosis, being an independent predictor of mortality. (7)

Therefore, early diagnosis is extremely important to reduce complications, guaranteeing a better prognosis for the patient and tracking first-degree relatives.

4 CONCLUSION

The diversity of clinical presentations, coupled with its genetic origins, underscores the significance of a holistic approach in the care of these patients. Advances in imaging techniques, notably echocardiography and cardiac magnetic resonance, play a pivotal role in early and precise identification of non-compacted myocardium. However, challenges persist, and ongoing research is paramount to refine diagnostic methods, gain a deeper comprehension of the natural history of the disease, and optimize therapeutic approaches.

As we consolidate our knowledge, it is crucial to remain attentive to clinical nuances and the latest scientific findings, thereby providing a solid foundation for advancements in the treatment and management of this intricate cardiac condition.

In conclusion, non-compacted myocardium emerges as an intriguing and complex cardiac condition that necessitates a profound understanding to guide effective diagnostic, treatment, and management strategies.

INFORMED CONSENT

Written informed consent for patient information and images to be published was provided by the patient.

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