

# Isolated noncompaction of myocardium in adults

Não compactação isolada do miocárdio no adulto

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## Resumo

*A não compactação isolada do miocárdio ventricular, ou miocárdio esponjoso, é uma forma congénita rara de miocardiopatia, caracterizada pela presença de trabeculações proeminentes e recessos intertrabeculares profundos em comunicação com a cavidade ventricular. Pode manifestar-se pela presença de insuficiência cardíaca, disritmias ventriculares e auriculares e tromboembolismo sistémico. Apesar de se tratar de uma situação congénita, o início da sintomatologia pode surgir tardiamente em adultos. Os autores apresentam um caso clínico com diagnóstico tardio: um homem, de 62 anos de idade, cuja avaliação imagiológica revelou as alterações patognomónicas da doença.*

*Palavras chave: Não compactação, ecocardiografia, miocárdio esponjoso*

## Abstract

*Isolated noncompaction of ventricular myocardium, or spongy myocardium, is a rare form of congenital cardiomyopathy, characterized by the presence of prominent trabeculations and deep intertrabecular recesses which communicate with the ventricular cavity. Heart failure, ventricular arrhythmias and systemic embolism are the major clinical manifestations. Although this is a congenital disorder, onset of symptoms*

*can occur late in adults. The authors present a case of late diagnosis: a 62-year-old male, in which image evaluation showed the pathognomonic features of the disease.*

*Key words: Noncompaction, echocardiography, spongy myocardium*

## Introduction

Isolated noncompaction of left myocardium (INCM), also known as spongy myocardium, is a rare congenital form (0,05% incidence in adults)<sup>1</sup> of dilated cardiomyopathy. Resulting from an arrest in the normal endomyocardial embryogenesis<sup>2</sup>, without any coexisting congenital cardiac abnormalities, it's characterized by the presence of numerous and prominent trabeculations and deep intertrabecular recesses, lined by ventricular endothelium<sup>3</sup>. Initially described as a congenital cardiomyopathy, there have been reports of late presentation, both isolated and familial<sup>4</sup>. Being categorized as one of the unclassified cardiomyopathies by WHO<sup>5</sup>, it is associated with high mortality and morbidity, due to the appearance of congestive heart failure, ventricular arrhythmias and systemic embolism. Treatment includes the management of heart failure, oral anticoagulation, eventual implantation of automated defibrillator/cardioverter<sup>6</sup> and, heart transplantation, in certain patients. The degree of ventricular dysfunction seems to be related with the extension of ventricular wall involvement<sup>7,8</sup>.

## Case report

A 62-year-old eurocaucasian male, construction worker, was complaining, two weeks before admission, of non-productive cough, dyspnoea with low efforts and leg oedema, being brought to our hospital due to worsening of complaints and sensation of precordial discomfort, admitted in grade III of NYHA classification of heart failure.

It was a patient with known hypertension, admitted in 1994 in the ICU due to acute pulmonary oedema, with hypertensive crisis and pulmonary infection. Echocardiogram at the time showed "left ventricular hypertrophy and dilatation, with global systolic function depression".

Followed as outpatient since 1994, he showed periods of dyspnoea and asthenia alternating with periods of higher effort toleration. Electrocardiography showed sinus rhythm, alternating with sporadic ventricular ectopic beats, bigeminism and atrial fibrillation with high ventricular rate. Another echocardiography showed dilatation of left cavities, hypertrophy of interventricular septum and depressed systolic function. In 1999, he was re-admitted due to ischemic stroke, overt heart failure and atrial fibrillation with high ventricular rate.

Physical examination in the last admission showed: AP - 138/100 mmHg; HR - 109 bpm.; Cardiac sounds: S1+S2, rhythmic, with no murmurs; Pulmonary sounds: symme-

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tric respiratory murmur, with rales in the inferior pulmonary area, bilaterally; Inferior limbs: bilateral oedema, with Godet sign.

Complementary exams showed: ECG – sinus rhythm, with ST segment elevation in V2 and V3, ST depression in V5 e V6 with T wave inversion, left ventricular hypertrophy and frequent polyfocal ventricular ectopic beats; Laboratory - CK: 73 U/L, CK-mb: 29 U/L, AST: 4 U/L, Troponin I: 1,4 ng/mL, Mioglobin: 164,3 ng/mL, CK-mass: 17,7 ng/mL. Thorax X-ray (*Fig.1*) showed enlarged cardio-thoracic index due to left cavities dilatation, and hilar ingurgitation. Echocardiogram (*Fig.2*) showed left ventricular dilatation, abnormally trabeculated in the apex, lateral ventricular wall and inter-ventricular septum. These aspects were also seen in the apex and adjacent area of the lateral wall of the right ventricle, suggestive of non-compaction of the myocardium, with poor global systolic function (FE – 15%). Spiral CT scan (*Fig.3*) and NMR confirmed the diagnosis, showing thickened wall, with two areas of different density in the non-compacted segments. Ventriculography showed abnormal retention of contrast in the deep trabecular recesses. Coronariography didn't show any coronary lesion. The electrocardiographic signs were considered related to previous history of hypertension and to systolic and diastolic surcharge.

A rapid regression of the heart failure manifestations was achieved with the institution of captopril, diuretics and digoxin. Life long oral anticoagulation was started.

## Discussion

Clinical manifestations of INCM are the development of heart failure, with systolic and diastolic dysfunction, atrial and ventricular taquidysrhythmias, conduction abnormalities (mainly intraventricular) and systemic tromboembolism.

Diagnosis is based mostly on bidimensional echocardiographic criteria, where myocardium segmentar thickening of the left ventricular wall is found (sometimes also on the right ventricle), with two layers: one small compacted epicardic layer; another thick inner layer, with multiple prominent trabeculations and deep intertrabecular recesses, in communication with the ventricular cavity<sup>6</sup>. Colour codified Döppler shows the blood flow between the ventricular cavity and the deep recesses.

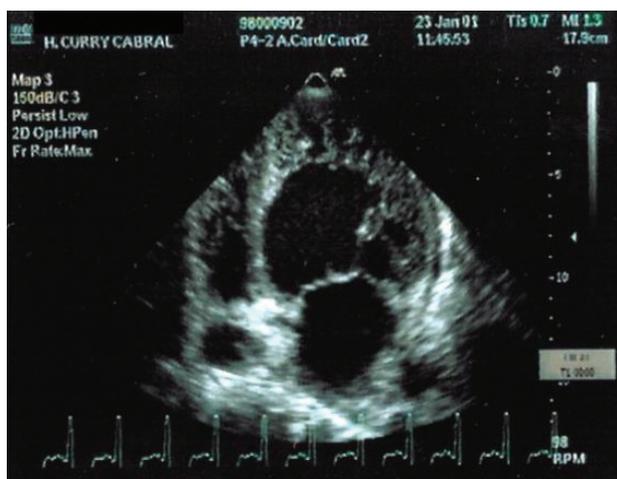
The diagnosis can be completed with CT scan, NMR and ventriculography, which show similar findings.

Spiral CT scan shows thickening of several segments of ventricular wall and, sometimes, of the septum. The thickened wall is composed by two areas of different density: one outer layer, consisting on a stripe of uniform tissue with a density similar to the muscle; the inner layer, thicker, composed by a mixture of contrasted blood with soft tissue<sup>7</sup>.

NMR clearly defines areas of myocardium non-compaction, showing the deep recesses and prominent trabecula-



*Fig. 1 – Thorax X-ray: enlarged cardio-thoracic index; hilar ingurgitation*



*Fig. 2 – Echocardiogram: multiple prominent trabeculations and deep intratrabecular recesses, in communication with the ventricular cavity.*

tions. In T2, it can show high-density areas in the left ventricular apex, which are consistent with microcirculation alterations due to fibrosis, thrombus formation and hypokinesia in the affected areas.

Ventriculography shows honeycomb pattern, or spongy like, of the inner contour of the noncompaction area during diastole and marked contrast retention in the intertrabecular recesses during systole. The hypokinesia of the non-compaction areas is well recognised in the vast majority





Fig. 3 – Spiral CT Scan: thickened wall composed by two areas of different density.

of cases, with left ventricular systolic function depression and elevation of the tele-diastolic and tele-systolic volumes. Coronariography is, usually, normal.

Positron Emission Tomography, with ammonium N-13 as flow marker, and dipiridamole, as stress study, is a new method of myocardium perfusion and coronary reserve flow study, used recently to demonstrate the abnormalities found in INCM<sup>9</sup>. The underlying mechanism seems to be a faulty development of the coronary microcirculation when the myocardium mass increases, or the compression of the intramural coronary bed by the hypertrophic myocardium, or both. Anyhow, these impairments seem to be more prevalent in the subendocardic ventricular area. QT segment prolongation and late potentials appearance identify a slow conduction area, possibly capable of re-entry phenomenon, which is the basis of ventricular monomorphic tachycardia episodes occurrence in these patients<sup>9</sup>.

Differential diagnosis must be made with normal variants (less trabeculations), with myocardium hypertrophy (hypertrophy pattern can simulate trabeculations, but recesses are never found), other miocardiopathies, with less trabeculations, and with thrombus in the apex, simulating trabeculations.

In our patient, coronary-ventriculography was made due to the electrocardiographic and enzymologie changes, suggestive of ischemia, but no coronary lesions were found. With improvement of the electrocardiographic data, non-

confirmed abnormalities of cardiac enzymologie, and progressive recovery of the symptous due to heart failure, the imagiologique data were enough to confirm the diagnosis of isolated noncompaction of myocardium.

Treatment is based on heart failure control, when there is dilated miocardiopathy, on disrhythmias early diagnosis and treatment, due to the possible malignant outcome and on cardioembolic prevention. An evaluation of family members is mandatory. Rarely, INCM can be diagnosed in several members of the same family, frequently in association with facial dismorphism<sup>10</sup>. Clinical and echocardiographic study made to our patient only son was negative; likewise, there was no family history of cardiac disease in patient parents.

### Conclusion

Although congenital, INCM or spongy myocardium diagnosis can occur late in life, as shown in our patient. Diagnosis is based on echocardiographic study, with all the other imagiologic exams being reserved to differential diagnosis of doubtful situations. Patients good outcome is closely linked with the efficacy in management of the clinical manifestations, namely, heart failure, taquidishrhythmias and tromboembolism.

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