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Left Ventricular Non-Compaction Cardiomyopathy: Case Report and Review of Literature

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Non-compaction cardiomyopathy (NCC) is characterized by trabeculations in either one or both ventricles. Clinical presentation is highly variable: dyspnea, palpitation, thromboembolic events, arrhythmia, or sudden cardiac death. There are currently no universally-accepted criteria for classifying and diagnosing left ventricular non-compaction (LVNC) cardiomyopathy. Transthoracic echocardiography (TTE) is the diagnostic exam of choice. The diagnosis is often missed or delayed because of a lack of knowledge about this uncommon disease. Progression of LVNC is highly variable and prognosis is very difficult to predict.

We report a case of a 50-year-old female patient with a history of total thyroidectomy under hormonal supplementation who consults for dyspnea and paroxysmal palpitations revealing an isolated LVNC.

This case emphasizes the importance of imaging techniques, which are, TTE and cardiac magnetic resonance imaging (MRI) in early diagnosis, management, and follow-up.

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Keywords: Cardiomyopathy; left ventricular non-compaction cardiomyopathy; cardiac magnetic resonance imaging; ventricular trabeculations; spongy myocardium.

1. INTRODUCTION

Non-compaction cardiomyopathy (NCC), also called “spongy myocardium”, is a rare genetic disorder characterized by trabeculations in either one or both ventricles [1]. NCC is found on transthoracic echocardiography (TTE) and/or cardiac MRI. This rare disease is associated with an increased risk of ventricular thrombus formation and even sudden death.

Clinical presentation is highly variable and can range from asymptomatic disease to symptoms like palpitations, chest pain, shortness of breath, or revealing heart failure, thromboembolic events, atrial or ventricular arrhythmias, bundle branch blocks, and even sudden cardiac death. There is currently no universally-accepted definition for left ventricular non-compaction (LVNC) cardiomyopathy.

Left ventricular non-compaction (LVNC) cardiomyopathy is thought to be caused by the arrest of normal embryogenesis of the endocardium and myocardium. It may be associated with other congenital cardiac defects.

We report a case of a 50-year-old female patient with a history of total thyroidectomy under hormonal supplementation who consults for dyspnea and paroxysmal palpitations revealing an isolated left ventricular non-compaction (LVNC) cardiomyopathy suspected in transthoracic echocardiography (TTE) and confirmed with cardiac MRI.

2. CASE PRESENTATION

A 50-year-old female patient with a past medical history of total thyroidectomy under hormonal supplementation who consults for dyspnea two months prior without any aggravating or alleviating factors. She also complained of frequent and sustained episodes of rapid palpitations. There was no family history of cardiomyopathy.

Physical examination found a stable patient, with a blood pressure of 138/78mmHg and a heart rate of 73 beats per minute. Her electrocardiogram (ECG) demonstrated normal sinus rhythm, normal axis, and repolarization disorders with negative T waves in aVL and DI leads (Fig. 1). Biological assessment shows a normal troponin level as well as normal renal and liver function tests.

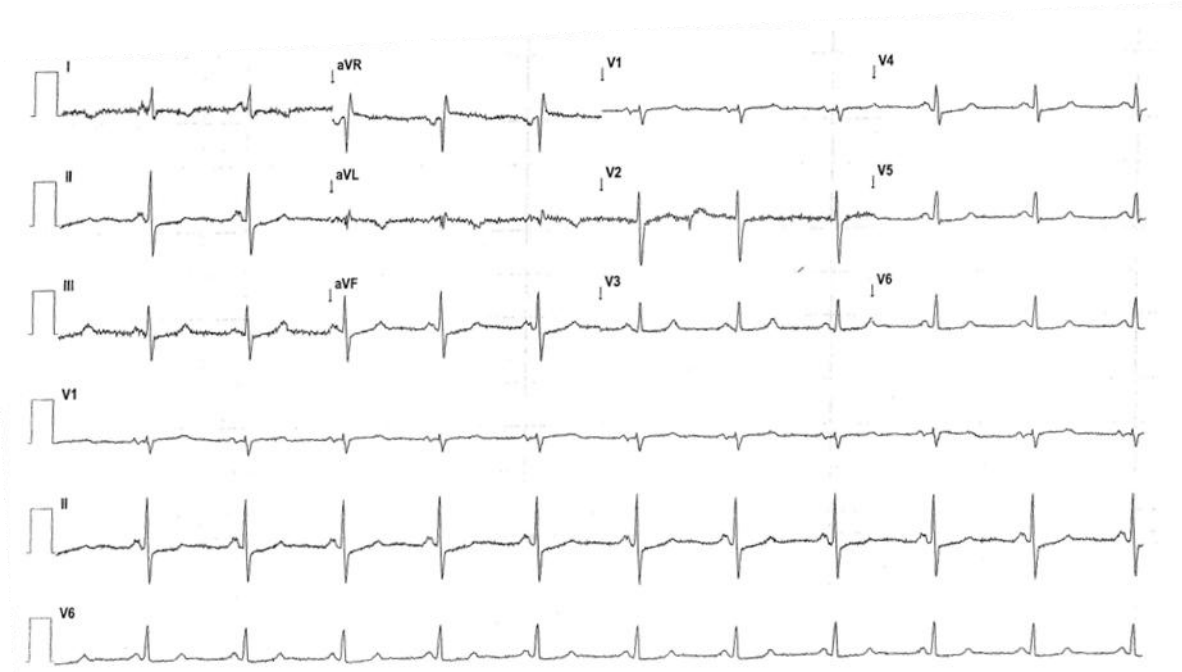

Fig. 1. Twelve-lead ECG showing sinus rhythm, normal QRS duration, and negative T-wave in aVL, DI

A TTE was performed demonstrating marked thickening trabeculation of the left ventricle predominantly at the apex and also in the apical and mid portion of the lateral and anterior wall. Color doppler displayed flow within the deep intertrabecular recesses. The left ventricle was not dilated. The left ventricle ejection fraction was mildly reduced to 45%. The right ventricle was normal. No additional abnormalities were seen. These echocardiographic findings were concerning for an isolated LVNC (Figs. 2-3). A cardiac catheterization did not reveal any evidence of obstructive coronary artery disease. Twenty-four-hour Holter monitoring identified frequent premature ventricular contractions.

Given the strong suspicion of non-compaction, a cardiac MRI was indicated and it demonstrated a trabeculated aspect of the papillary muscles and hyper trabeculation of the left ventricle with a ratio of non-compacted to compacted myocardium of 2.7. The tagging sequence showed an alteration of the intrinsic contractility of the anterior wall. There was no late gadolinium enhancement suggestive of myocardial infarction or fibrosis (Figs. 4-5).

Our patient was started on treatment for heart failure (beta blocker and angiotensin-receptor blocker) and acenocoumarol 4 mg daily with an international normalized ratio (INR) goal of 2-3. Our patient was having regular follow-ups for arrhythmia screening and monitoring of her left ventricular function.

Familial screening of first-degree relatives was negative for LVNC and congenital heart disease.

Today, after two years of follow-up under treatment, we noted a good evolution of the symptoms, and regression of the episodes of palpitations without any clinical or echocardiographic thrombo-embolic incident.

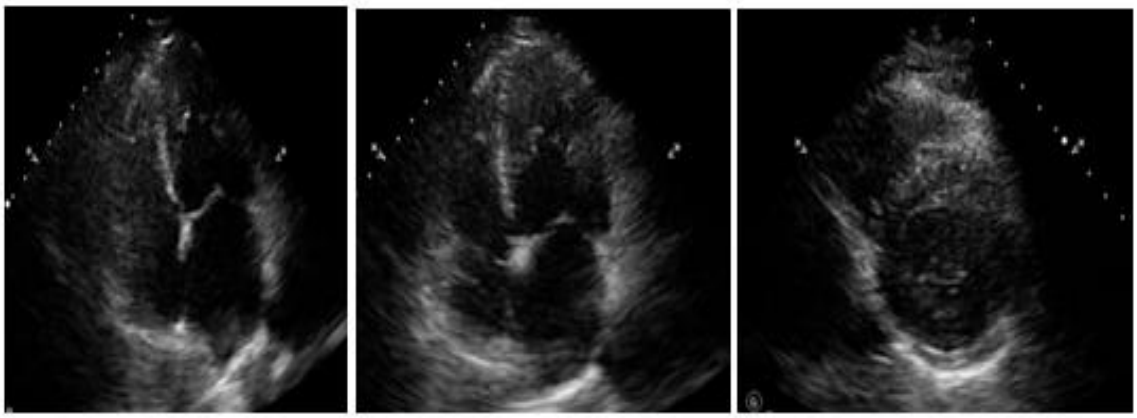

Fig. 2. 2D echocardiography: apical four-chamber (A4c) and short axis showing trabeculation of the apex, the medium and apical segment of the interventricular septum, and the lateral wall

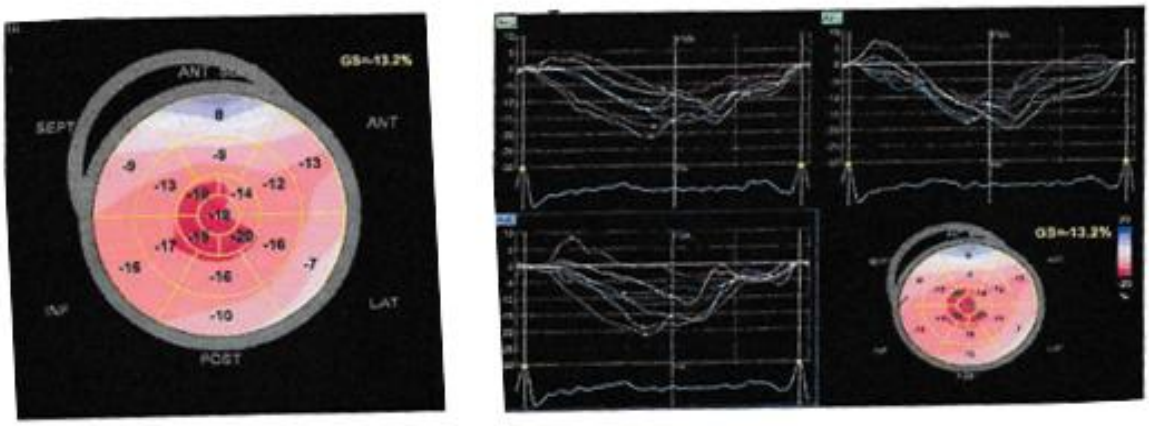

Fig. 3. 2D speckle tracking echocardiography

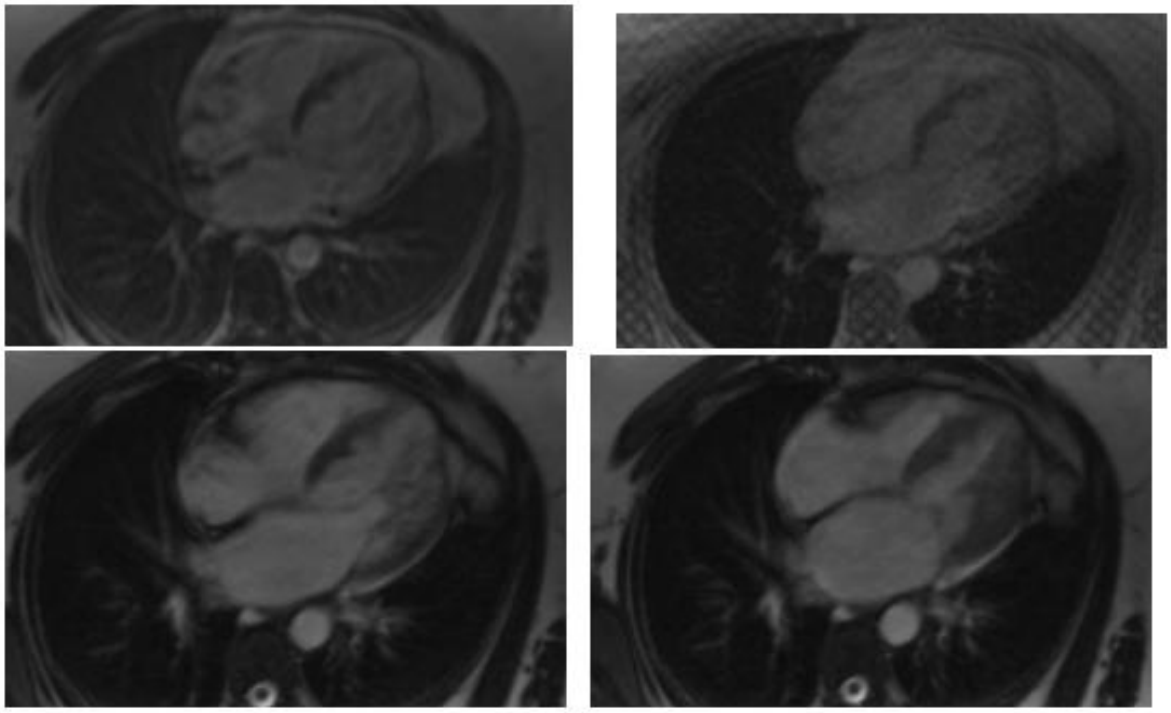

Fig. 4. Four-chamber view of cardiac MRI revealing an LVNC (systole and diastole)

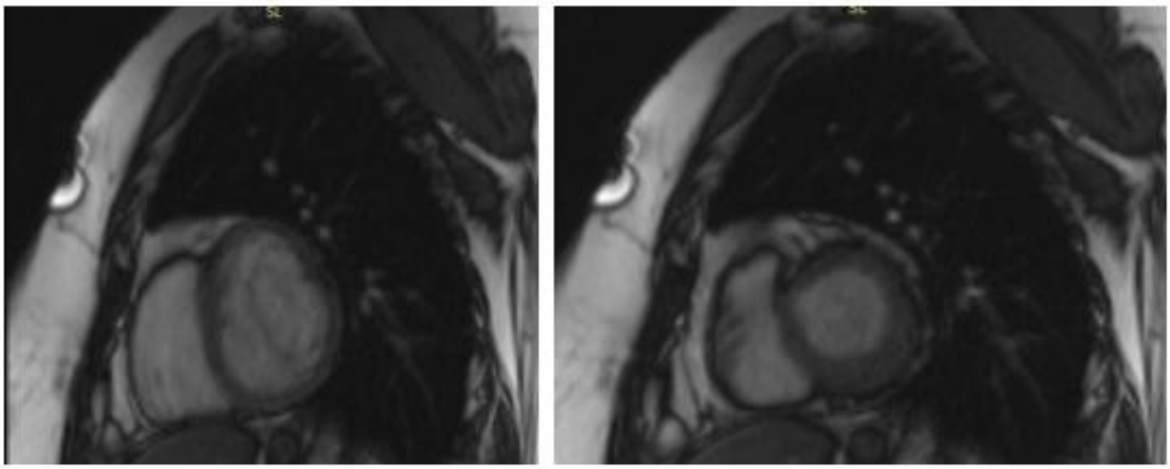

Fig. 5. Short axis view of cardiac MRI revealing an LVNC (diastole and systole)

3. DISCUSSION

"NCC is a rare genetic cardiomyopathy [1] first described in 1932 when Bellet et al reported this anomaly in newborns with aortic atresia and coronary artery-left ventricular fistula". "It is characterized by excess trabeculations in the endocardium and can affect one or both ventricles" [2].

"LVNC is a rare cardiomyopathy. Its prevalence is less than 0.02% and is male predominant" [1]. "It is often familial with an autosomal dominant mode with defects in genes coding for sarcomere, cytoskeleton, and mitochondrial proteins or sporadic" [3].

ECG can be normal, or show early repolarization abnormalities, QTc prolongation repolarization, atrial fibrillation, paroxysmal supraventricular tachycardia, or even complete heart block [4].

The most used method of diagnosis is TTE [5]. "The cardiac MRI can confirm the TTE finding

when the ratio between the end-systolic thickness of the compacted vs noncompacted tissue is greater than 2.0. Recent reports indicate high sensitivity and specificity using a ratio of >2.3" [5].

"There are two sets of echocardiographic criteria: the Jenni criteria focused on the presence of a two-layered structure [6], and the Chin criteria focused on the depth of the recess compared with the height of the trabeculations" [7]. "Jenni criteria are the most accepted echocardiographic criteria and consist of evidence of a two-layer structure: a compacted layer and a noncompacted endocardial layer. In the short-axis view, the end-systolic ratio of noncompacted to compacted layers (NC/C) > 2.0 is compatible with the diagnosis" [6]. "Additional criteria that must be met include the absence of any coexisting cardiac abnormalities and color doppler evidence of deep perfused intertrabecular recesses" [6]. "Chin criteria considered for diagnosis are the presence of numerous excessively prominent trabeculations and deep intertrabecular recesses with the ratio of the distance from the epicardial surface to the trough of the trabecular recesses and distance from the epicardial surface to the peak of trabeculation ≤ 0.5, assessed at end-diastole on short-axis parasternal views and/or apical views" [7]. "It is also important that no other cardiac structural abnormalities be present" [7]. Stollberger et al. defined "LVNC as trabeculations >/3, prominent formations along the left ventricular endocardial border, located apically to the papillary muscles, visible in end-diastole, in one imaging plane, moving synchronously with the compacted myocardium, distinct from the papillary muscles, false tendons, or aberrant bands" [8]. Ghebhard et al. considered "compacted myocardium systolic thickness < 8 mm for diagnosis of LVNC" [9].

"In difficult cases, other echocardiographic techniques can be used for the diagnosis: contrast enhancement, three-dimensional echocardiography, speckle tracking, and tissue Doppler imaging. Speckle-tracking echocardiography is used in borderline cases because LVNC affects the left ventricle twist" [10].

LVNC is usually associated with cardiac structural abnormalities, reduced ejection fraction or systolic dysfunction, due to pathogenic ischemic and damaged endocardium, as evidenced by fibrotic changes on cardiac MRI and also due to asynchronism of contraction between the compacted and noncompacted myocardial layers [4]. Isolated NCC can occur as well.

"Cardiac MRI plays a pivotal role in the diagnosis of LVNC. *Patersen et al* suggest that LVNC is diagnosed accurately with cardiac MRI using the NC/C ratio in diastole. An NC/C ratio of >2.3 in diastole distinguishes pathological non-compaction, with a sensitivity, specificity, and positive and negative predictions of 86%, 99%, 75%, and 99%, respectively" [11]. **"**For the *Grothoff* criterion, the NC/C ratio is measured in diastolic short-axis views, and the cutoff for LVNC is NC/C ≥3" [12]. "The *Stacey* criterion is calculated from end-systolic short-axis views, with NC/C ≥2 considered positive for LVNC" [13]. "The Jacquier criterion is estimated from the noncompacted myocardial mass as a percentage of total left ventricular mass; a noncompacted mass ≥20% is considered to indicate an LVNC phenotype" [14].

Herein, we expose some studies based on TTE and cardiac MRI to define an LVNC (Table 1) [15].

These definitions highlight variations in current definitions of excessive trabeculation.

"There is no specific therapy for patients with LVNC. Treatment is focused on complications, which are, heart failure, systemic embolism, and sudden cardiac death" [16]. "Management of symptomatic patient's heart failure is based on digoxin, diuretics, angiotensin-converting enzyme inhibitors, and beta-blockers. Some patients undergo cardiac transplantation. Cardiac rhythm abnormalities are managed with standard protocol, while some patients may benefit from an implanted cardiac defibrillator for severe ventricular tachyarrhythmias to prevent sudden death" [17].

"Hypertrabeculation predisposes patients with reduced left ventricular function to a risk of clot formation. Some literature supports the use of anticoagulation in patients with LVEF≤ 40%, a history of atrial fibrillation, or a prior cardioembolic event. Whether anticoagulants should be administered to every LVNC patient is, however, still debated. Anticoagulation therapy must be targeted to the individual patient after careful assessment of the benefit and risks. Oral anticoagulation therapy (target INR 2.0–3.0) is

Table 1. Examples of echocardiographic and cardiac MRI approaches to determining the extent of left ventricular trabeculations

	Jenni et al. [6]	Petersen et al. [11,15]	Jacquier et al. [14]	Stacey et al. [13]	Captur et al.
Modality	**TTE**	**MRI**	**MRI**	**MRI**	**MRI**
Sample size	NC* (n = 34) No control group	NC (n = 7) Control subjects (n = 170)	NC (n = 16) Control subjects (n = 48)	NC (n = 122) No control group	NC (n = 30) Control subjects (n = 105)
Study design/ external validation	Retrospective/ no external validation cohort	Retrospective/ no external validation cohort	Retrospective/ no external validation cohort	Retrospective/ no external validation cohort	Retrospective/ no external validation cohort
Definition of NC	Absence of coexisting cardiac disease Numerous excessively prominent trabeculations and deep intertrabecular recesses Intertrabecular spaces filled by direct blood flow from the ventricular cavity, on colour Doppler imaging	Bilayered appearance on TTE combined with increased pretest probability (eg, similar appearance in first-degree relatives, associated neuromuscular disorder, or complications, such as systemic embolization and regional wall motion abnormalities)	Diagnosis of NC was established on echocardiographic criteria	Consecutive patients from MRI reports that mention trabeculation or NC	Diagnosis of NC echocardiographic criteria and at least 1 of the following: positive family history, associated neuromuscular disorder, regional wall motion abnormality, NC-related complications (arrhythmia, heart failure, or thromboembolism)
Description	NC to compaction ratio Decreased thickening and hypokinesia present within, but not limited to the noncompacted segments	Two-layered myocardium measured at the most pronounced trabeculations, avoiding apex measurement perpendicular to compact myocardium	Short-axis cines for total LV** mass and compact mass to define trabecular mass Papillary muscle included in the myocardial mass	Apical short-axis views 16-24 mm from the true apical slice Region with the largest NC to compaction ratio	Loss of base-to-apex fractional dimension gradient
Cardiac phase	End-systole	End-diastole	End-diastole	End-systole	End-diastole
Cardiac view	Short axis	Long axes (4-chamber, 2-chamber, 3-chamber)	Short-axis stack	Apical short axis	Short-axis stack
Excessive trabeculation cutoff	NC to compaction ratio >2	NC to compaction ratio >2.3	Trabecular mass >20%	NC to compaction ratio ≥2	Fractal dimension ≥1.30

** NC = non-compaction, ** LV = left ventricular*

recommended in patients with impaired systolic function, previous history of embolism, transient ischemic attack, atrial fibrillation, and intracardiac thrombi identified on echocardiogram or another cardiac imaging modality" [20]. "Thromboembolic risk assessment based on $CHADS_2$/$CHADS_2$-VASc scores similar to that used to stratify stroke risk in patients with atrial fibrillation is recommended" [18,19]. "However, the length of anticoagulation therapy in these patients has not been established. If using warfarin, a targeted INR range of 2-3 is proposed" [9].

"Patients with LVNC and sustained ventricular tachycardia or ventricular fibrillation require an implantable cardioverter defibrillator (ICD). These patients are at higher risk for sudden cardiac death, even with normal ejection fraction. Current implantable cardioverter defibrillator primary and secondary prevention guidelines are to be followed. An implantable cardioverter defibrillator for primary prevention of sudden cardiac death is indicated for patients with LVNC who present with LVNC and LVEF ≤ 35%. Patients with malignant ventricular tachyarrhythmia should receive implantable cardioverter defibrillator for secondary prevention" [20].

The outcomes of transplanted patients have yet to be established.

Follow-up consists of symptom assessment, LVEF monitoring, and arrhythmia screenings, including electrophysiology evaluation if needed [9,20].

All patients should receive family and genetic counselling [9].

4. CONCLUSION

LVNC is an uncommon cause of cardiomyopathy. There are still no universally established criteria for its diagnosis. The management and treatment of each case of LVNC should be individualized for each patient. All patients should receive family and genetic counselling.

2D echocardiography and cardiac MRI play a pivotal role in the diagnosis and management of LVNC. However, more studies are needed to establish the choice and duration of anticoagulants, including direct oral anticoagulants.

CONSENT

As per international standard or university standard, patient(s) written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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