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Heart Failure de novo in Left Ventricular Noncompaction Cardiomyopathy

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Abstract

Noncompaction Cardiomyopathy (NCC) is a rare cardiomyopathy that features a persistent fetal myocardium, a prominent trabecular meshwork and deep intertrabecular recesses, systolic dysfunction and left ventricular dilatation. There is no consensus on the definition and management of NCC. We report the case of a 64-year-old patient with NCC, with congestive heart failure de novo without known cardiovascular risk factors and family history of sudden cardiac death. An implantable cardioverter-defibrillator was inserted due to the patient's high risk for sudden cardiac death. Establishing definitive guidelines for the diagnosis of NCC should lead to earlier identification of patients, prompt treatment and improved survival.

Keywords: Heart Failure, Hypertrabeculation Syndrome, Left Ventricular Noncompaction, Noncompaction Cardiomyopathy, Spongy Myocardium

Introduction

While most cases of the patients who develop heart failure on the basis of previous known history of cardiovascular disease, there are rare cases of heart failure that occur without predisposing risk factors. Noncompaction cardiomyopathy (NCC) is characterized by a prominent left ventricular trabecula and deep intertrabecular recesses involving the left ventricle or both cardiac ventricles, resulting in heart failure.

The diagnosis of Noncompaction Cardiomyopathy (NCC) is rare but can be lethal if not recognized. NCC is defined by three main features: the thinning of the compact (C) myocardial layer, a prominent non-compact (NC) trabecular layer, with deep intertrabecular recesses. The ratio of NC to C thickening is at least 2 to 2.3. Finally, Doppler analysis features of blood flow passing through trabecular and crypts [1–3].

Case Report

64-year-old man with a four-month history of shortness of breath developed further progressing of symptoms, with dyspnea on mild exertion associated with palpitations. Family History was remarkable for multiple first and secondary male family members with non-specific cardiovascular disease, where his father and brother both suffered sudden cardiac death at young age, at 55 and 43 years of age respectively. The patient's cardiovascular exam revealed normal S1, S2 heart sounds, no S3 and no murmurs, with bilateral diffuse crackles over the lung fields. Lower extremities featured bilateral pedal edema. Electrocardiogram (ECG) showed a left bundle branch block (Figure 1).

A 2D-Echocardiogram exhibited diastolic dysfunction grade I, mild left ventricular dilation, Left Ventricular Ejection Fraction (LVEF) of 35% by Simpson criteria, and evidence of mesh of endocardial trabeculations.

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A coronary angiography was normal as well as a 24-hour Holter monitor recording (Figure 2,3).

Cardiac Magnetic Resonance Imaging (MRC) revealed a large diastolic myocardial ratio of non-compacted to compacted thickness suggestive of Noncompaction Cardiomyopathy. An implantation of a cardiac resynchronization defibrillator was performed, and patient was discharged home on chronic anticoagulation (Figure 4,5).

Discussion

The first account of postnatal persistence of spongy myocardium was a pathological description in 1975 [4]. Two decades later, an isolated, rare myocardial anomaly term "isolated non-compaction of left ventricular myocardium" was recognized, as product of an arrest of the normal compaction process during embryogenesis [5].

Embryogenetic etiologies are key in understanding the pathogenetic mechanisms in the development of NCC. Nonetheless, the description of acquired cases brings into question if it is appropriate to consider arrested maturation as the sole cause in all cases of NCC. It may be part of chronic kidney disease, pregnancy, in young athletes, Sickle Cell Anemia or even hypertension. It is considered that in these cases hemodynamic forces play a great role in disease pathogenesis, and some of these states of NCC may be reversible.

Epidemiology

Non-compaction of the left ventricular myocardium is considered a rare form of cardiomyopathy. The true prevalence of NCC is unknown; the prevalence of reported NCC in echocardiography laboratories is between 0.014 and 1.3% [6]. In a retrospective cohort study from the Texas Children's Hospital echocardiography database NCC accounted for 9.5% of cases, representing the third most frequent cardiomyopathy after ischemic heart disease, idiopathic dilated cardiomyopathy, and valvular disease [7] (Table 1).

Genetics

Non-compaction of the left ventricular myocardium is a heterogeneous disorder with a sporadic and familial form. Upon identification of an index case of NCC, the familial occurrence rate is estimated at about 30% [8], although 60-70% of cases appear to be sporadic [9].

There is no single gene defect responsible for NCC; many of the of the genetic phenotypes coincide clinically with cardiomyopathies or genes encoding for sarcomeric proteins, and even for disorders of the mitochondria and cytoskeleton [10–12]. NCC is associated with many genetic conditions, including muscular dystrophies, ion channelopathies that may also cause arrhythmias (e.g. long QT Syndrome), aberrant chromosomal disorders (e.g. trisomies), and as part of metabolic syndromes [13]. In some familial cohorts, several members may present with NCC and others with dilated or hypertrophic cardiomyopathies (Table 2).

Clinical presentation

The spectrum of the initial presentation of NCC is broad. Major clinical manifestations are symptoms of systolic,

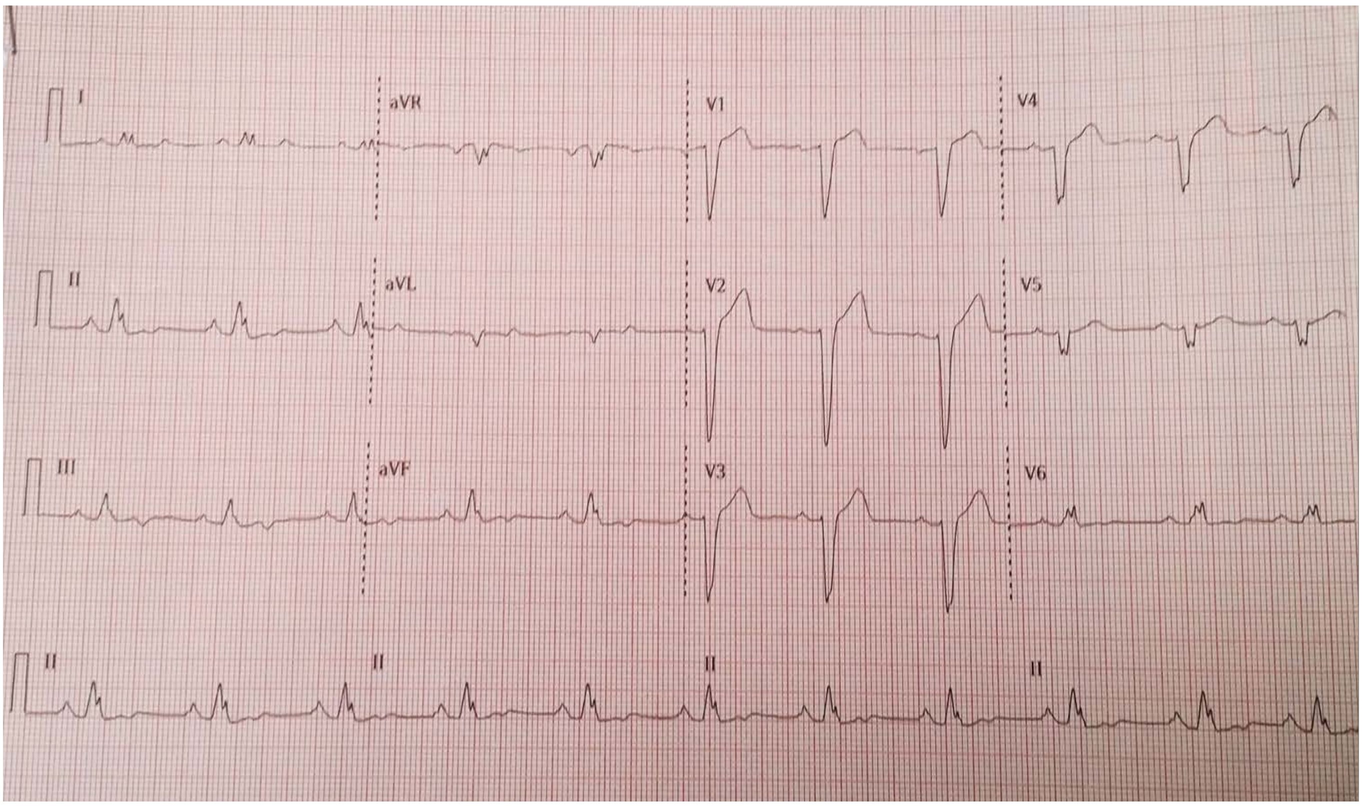

Figure 1: ECG: Sinus rhythm with LBBB.

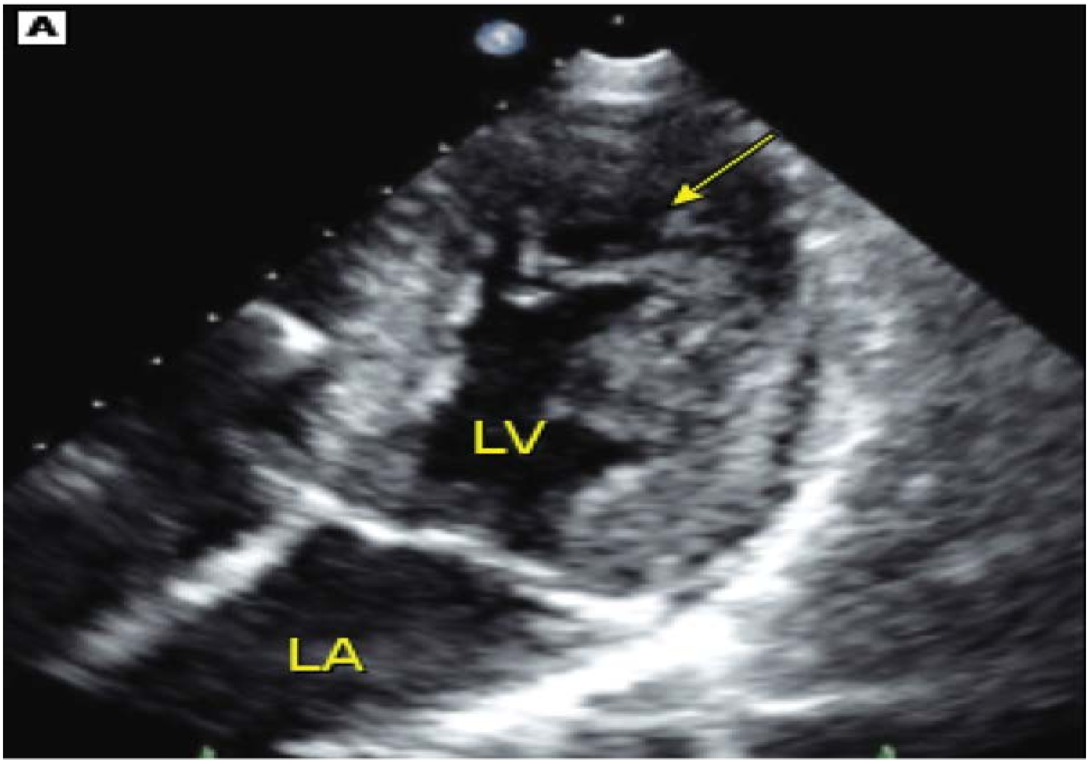

Figure 2: 2D-Echocardiogram showing multiples endocardial trabeculations within a thickened myocardium.

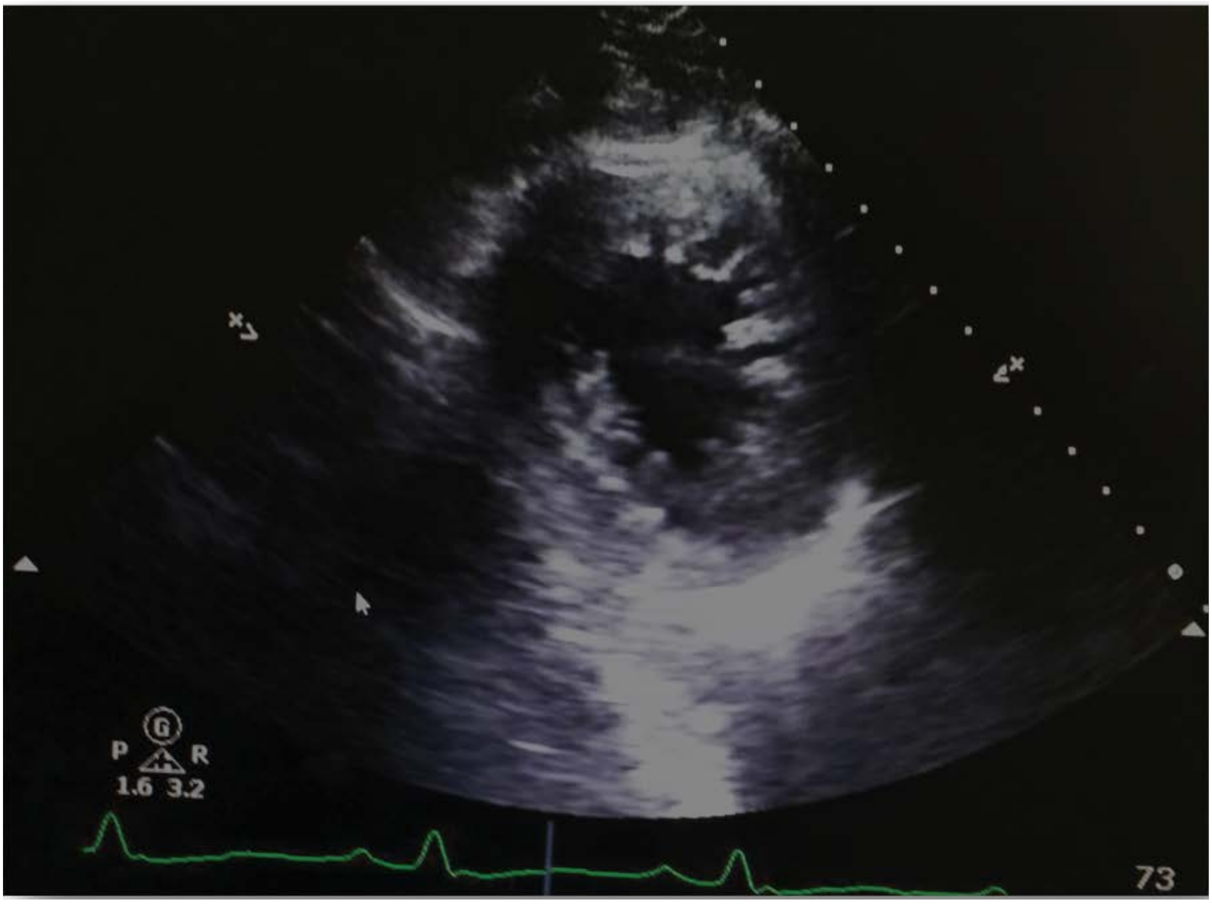

Figure 3: 2D-Echocardiogram showing the Noncompaction over Compaction myocardial thickness ratio (NC/C ratio) of 1.4 to 0.68, calculated at over 2 to 1 in the in End-Diastolic (ED) short-axis view.

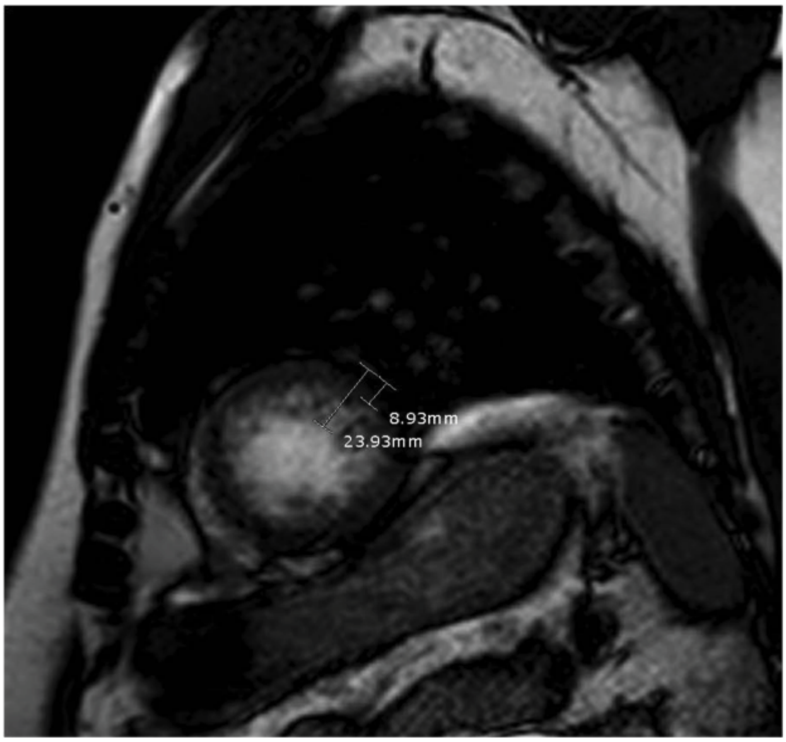

Figure 4: Cardiac MRI (CMR) views feature large diastolic myocardial ratio of non-compacted to compacted thickness, at the ED Short axis the NC/C ratio was calculated as greater than 2.3.

Figure 5: Cardiac MRI (CMR) Long axis four chambers view at ED NC/C myocardial thickness ratio of >2.3.

Table 1: Estimated Prevalence of LVNC.

Infants	1:100,000
Children	1:1,000,000
Adults	1:10,000

Table 2: Associated forms of LVNC.

Familial LVNC
LVNC associated with Genetic or Metabolic Congenital Syndromes (e.g. Barth Syndrome)
Acquired LVNC: Pregnancy, Sickle Cell Anemia, Young Athletes

diastolic heart failure, and include nonspecific chest pain, arrhythmias like atrial fibrillation, ventricular arrhythmias, sudden cardiac arrest, and thromboembolic events, including stroke.

The electrocardiographic findings are nonspecific for NCC. It can range from entirely normal electrocardiogram to intraventricular conduction delay, left ventricular hypertrophy, and repolarization abnormalities.

Table 3: Echocardiography features in LVNC.

Chin et al. [5]	Jenni et al. [16]	Stöllberger and Finsterer [17]
Two-layered structure of the myocardium (epicardial compacted, endocardial non-compacted layer)	Thickened myocardium with a two-layered structure consisting of a thin compacted epicardial layer/band (C) and a much thicker, non-compacted endocardial layer (N) or trabecular meshwork with deep endomyocardial spaces; N/C ratio 2.0	More than three trabeculations protruding (within 1 imaging plane) protruding from the LV free wall, apically from the papillary muscles
Determination of the X-to-Y ratio (≤0.5) X—Distance between the epicardial surface and through of intertrabecular recess	Predominant location of the pathology: mid-lateral, mid-inferior, and apex Color Doppler evidence of deep intertrabecular recesses filled with blood from the left ventricular cavity.	Trabeculations with the same echogenicity with synchronous movement with ventricular contractions.
Y—Distance between epicardial surface and peak of trabeculations.	Absence of coexisting cardiac abnormalities (in the presence of isolated LVNC)	Ratio of non-compacted to compacted segment 2.0 at end-diastole.
Acquisition of the images: parasternal short-axis view, measurements of the X-to-Y ratio at end diastole.	Acquisition of the images: short-axis views, measurements of the N/C ratio at end systole.	Acquisition of the images: apical four chamber view.

Table 4: Cardiac Magnetic Resonance Imaging descriptions in LVNC.

Petersen et al. [2]	Jacquier et al. [18]
Ratio between the non-compacted and compacted layer >2.3 Measurement: at end-diastole	Trabeculated left ventricular mass >20% of the global left ventricular mass Measurement: left ventricular trabeculation and global/ compacted LV mass were defined at end-diastole the

Table 5: Diagnostic Features in LVNC.

NC/C>2.3
+ Family History of LVNC

Table 6: Complications of LVNC.

AV Arrhythmias
Cardiac failure
Thromboembolism

The amount of normal myocardium will determine the functional impact of the pathology and the highly variable clinical course. The most severe cases present during the first years of life. At the other end of the severity spectrum are patients with very localized forms of myocardial noncompaction that can be asymptomatic for prolonged periods, with the diagnosis only being made at an old age.

Diagnostic Approach

There is much debate regarding the diagnostic criteria for NCC but there may be a predilection for overdiagnosis if criteria are not set strictly enough. For example, remodeling after myocarditis may be misdiagnosed as NCC [14], and similarly with many other well defined clinical entities, caution advised to avoid a diagnosis purely based on visualized estimate of echocardiography.

There is no gold standard test or genetic tests confirmatory of NCC. Multiple modalities may be required for a complete assessment. 2D-Echocardiography is the basic tool for the evaluation of NCC. The classical echocardiographic features of NCC are the presence of multiple prominent trabeculations with deep intertrabecular recesses invaginating deeply into the outer one third of the ventricular myocardium. High resolution imaging of cardiac magnetic resonance (CMR) delivers anatomy in greater detail, as well as functional information of the compacted and noncompacted enhancement for the evaluation of fibrosis (Table 3,4).

Misdiagnosis may be secondary to the presence of compacted papillary muscles of phenomena associated to dilated cardiomyopathy. The diagnostic criteria continue to be debated. It is based on both anatomic and functional characteristics from cardiac Magnetic Resonance Imaging (CMR) or two-dimensional echocardiography (2D-Echo) with the proposed cutoff measures of Non Compacted to Compacted ratio (NC/C) equal or more than 2.23 at the end of systole. Contrast echocardiography enhancing the cardiac wall may be especially helpful. Family history helps in establishing a more precise diagnosis (Table 5).

Management

There is no specific therapy for patients with NCC. There is limited data prospectively assessing specific agents for long-term outcomes in NCC. The standard of care for patients NCC should include evidence-based guideline directed medical therapy for the management of patients with cardiomyopathy, including anticoagulation and primary prevention of sudden cardiac death. The current guidelines for insertion of an implantable cardioverter-defibrillator should be considered in patients with NCC presenting with syncope, symptomatic ventricular arrhythmias or with severely impaired LV systolic function (LVEF<35%).

Another important management goal is the prevention of thromboembolic complications. Anticoagulation should be instituted when a definite left ventricular clot has been identified independent of ventricular systolic function and in patients with atrial fibrillation and or LVEF <40% with high CHA2DS2-VASc score. For the patients who do not fall into either of these categories, risk assessment using the CHA2DS2-VASc score is to be used as guidance and followed by a discussion with the patient regarding the risks and benefits of anticoagulation (Table 6).

Conclusions

Four decades later after its original description, the controversy continues about the etiologic mechanism of NCC, challenging the notion that there is single etiologic mechanism [15]. While morphologic assessment is the only available diagnostic workup, with no single definitive genetic pathway identified, NCC or isolated left ventricular noncompaction remains a diagnostic and management challenge. Consensus for diagnostic criteria are needed as well as further development of patient registries in order to capture data of both clinical and genetic sources. The diagnosis of NCC needs to be established with caution. If a Mendelian association is made, genetic counseling may be considered.

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