The different clinical scenarios of left ventricular non-compaction: report of three cases

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Abstract

Introduction

Left ventricular non-compaction (LVNC) is a myocardial disorder characterized by prominent ventricular trabeculations and deep recesses extending from the left ventricular cavity to the sub-endocardial surface of the left ventricular wall with or without left ventricular dysfunction1,2. LVNC can be diagnosed in all stages of life. It has been identified as a distinct entity over the past few decades3–4. When the disease is first recognized, which usually occurs in later stages of life, it is often not clear whether the diagnosis represents late recognition of long-standing non-compaction or delayed morphological manifestation of an underlying cardiomyopathy. LVNC may be associated with other diseases such as neuromuscular disorders5. Although the American Heart Association has classified LVNC as a primary genetic cardiomyopathy6, the European Society of Cardiology refers to LVNC as an ‘unclassified cardiomyopathy’6 based on the fact that LVNC is a morphological manifestation of several distinct cardiomyopathies. Currently, there is no definite evidence to confirm whether LVNC is a distinct cardiomyopathy, an epiphenomenon or a phenotypic variant of other cardiomyopathies. However, the absence of specific genotype-phenotype association, the occurrence of LVNC morphology in various metabolic diseases and other cardiomyopathies, and the finding that the LVNC phenotype is not necessary for the development of cardiomyopathy, all suggest it is unlikely that it could be a specific cardiomyopathy7–9. Traditionally, diagnosis of LVNC is based on 2D trans-thoracic echocardiography8. Although there are three different diagnostic criteria, there is no universally accepted definition of LVNC. All criteria are based on morphological findings and require the presence of prominent trabeculations with deep intertrabecular recesses communicating with the ventricular cavity and a two-layered appearance of the myocardium (trabecular myocardium as one layer, and compacted myocardium as the second layer)10–12. If a ratio of trabeculated to compact myocardium of more than 2.3 is common in a large-population-based cohort, a critical re-evaluation of the current cardiac magnetic resonance (CMR) criteria for LVNC may be needed13. This article discusses the different clinical scenarios of LVNC.

Case report

The first case concerns a 50-year-old female who came to our medical centre. Subject complained of dyspnea and palpitation on effort for several months, which was attributed to her obesity and lack of exercise. The electrocardiography (ECG) showed a sinus rhythm interrupted by frequent ventricular premature beats, aspecific interventricular conduction delay (QRS 140 ms) with a left heart axis. Two dimensional trans-thoracic echocardiography performed with MyLab 30 Gold (ESAOTE Instruments) showed a marked dilatation of the left ventricle with global hypokinesis and very poor systolic function (EF 20%), complicated by severe functional mitral regurgitation by tethering and apical displacement of mitral leaflets; also, a moderate left atrial dilatation and mild tricuspid regurgitation was present without an increase in the systolic pulmonary pressure. Prominent trabeculation was evident on the anterolateral wall,
with deep intertrabecular recesses communicating with the ventricular cavity, and a two-layered appearance of the myocardium (trabecular myocardium as one layer, and compacted myocardium as the second layer; see Figure 1). In addition, intertrabecular space was filled by direct blood flow from the ventricular cavity as visualized on colour Doppler imaging. The diagnosis of non-ischemic dilated cardiomyopathy in LVNC was made and subsequently a cardiac magnetic resonance confirmed this disease (see Figure 2).

The second case concerns a 35-year-old elite triathlete in good health condition and highly competitive in sports with unremarkable clinical history. No abnormalities in physical examination were found. Resting ECG showed sinus bradycardia, early repolarization and a sign of left ventricular hypertrophy that required further work through two-dimensional trans-thoracic echocardiography performed with MyLab 30 Gold (ESAOTE Instruments). This exam showed a non-dilated left ventricle with good systolic function (estimated EF 65%) and absence of valvular disease or other abnormalities, but the presence of a myocardial disorder characterized by prominent ventricular trabeculations and deep recesses extending from the left ventricular cavity to the sub-endocardial surface of the whole anterolateral left ventricular wall (see Figure 3). Mild right ventricular involvement was also shown (see red arrow). Subsequently, a CMR showed the typical morphologic findings of left ventricular non-compaction of the anterolateral wall (see Figure 4) without dysfunction.

The third case concerns the incidental finding of non-compacted myocardium in a 48-year-old female cyclist who was asymptomatic. This athlete was subjected to sports screening prior to participation. Subject had no family history of cardiac disease or sudden death. Physical examination revealed the presence of a slight heart murmur. Resting ECG showed normal sinus rhythm, and exercise testing was negative for arrhythmias. Echocardiography examination showed periapical and apical thickening with spongiform appearance.

Figure 1: Two-dimensional trans-thoracic echocardiography colour Doppler (apical four-chamber view) recorded in a patient with a dilated cardiomyopathy related to ventricular non-compaction showed a prominent trabeculation along the lateral wall as well as severe functional mitral regurgitation and mild tricuspid regurgitation.

Figure 2: Cardiac magnetic resonance (CMR) shows the typical finding in the short-axis view of non-compacted to compacted layer ratio of more than 2.3 in diastole on the lateral wall.
of the myocardium and deep intertrabecular recesses with the association of a false tendon inside the cavity (see Figures 5 and 6). The diagnosis of LVNC myocardium was made. In this case, the athlete has refused to undergo MRI.

Discussion

These three case reports describe different clinical scenarios of the same pathology and then three totally opposite sides of the same structural morphological abnormality of the myocardium. In the first case, the LVNC is related to an important base of cardiomyopathy such as dilated cardiomyopathy, which causes a severe dysfunction of the left ventricle and requires suitable treatments and therapies; in the second case, the same morphological alteration of the myocardium in a subject appears perfectly healthy and suitable to the athletic skills of that subject, who is completely asymptomatic. It follows that prominent left ventricular trabeculation can be found in asymptomatic, healthy athletes as well as in subjects with hypertrophic or dilated cardiomyopathy. Thus, the difference between variants and LVNC may often be challenging. What makes the difference, in our opinion, is not the morphology but the function of the myocardium affected and the absence of other associated anomalies that determine the prognosis of these patients.

Conclusion

The variety in clinical presentation, genetic heterogeneity and phenotype of the first transgenic animal model of an LVNC-associated mutation question the hypothesis that LVNC be a distinct cardiomyopathy. It seems to be a distinct phenotype or phenotypic, morphological expression of different underlying diseases rather than a distinct cardiomyopathy. We believe that any person can have non-compacted myocardium and that this development is not only about the genetic
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Competing interests: none declared. Conflict of interests: none declared.

Consent

Informed consent was collected from the patients for the procedures performed. Consent for data publication was also collected from the patients. Written informed consent was obtained from the patients for publication of this manuscript and accompanying images.

References


Figure 5: Two-dimensional trans-thoracic echocardiography (in apical four-chamber view) shows periapical and apical thickening with spongiform appearance of the myocardium and deep intertrabecular recesses with the association of a false tendon inside the cavity.

Figure 6: A zoom on the left ventricular apex showed excessive myocardial trabeculation with deep recesses and false tendon (arrows head).
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