CASE REPORT

ISOLATED LEFT VENTRICULAR NON COMPACTION IN A 45-YEAR OLD PATIENT

• *clinical presentation, multimodality imaging findings and follow up* •

Carma Karam*, Stephen Binsse**, Nicolas Mansencal, Pascal Lacombe****, Olivier Dubourg********

Corresponding author: Carma Karam <carma.karam@apr.aphp.fr>

* MD. Cardiology Department, University of Versailles - Saint Quentin en Yvelines, Ambroise Paré Hospital, Assistance Publique-Hôpitaux de Paris (AP-HP), Centre de référence des Maladies Cardiaques Héréditaires, Boulogne-Billancourt, France. UMR-EpReC.

** MD. Radiology Department, University of Versailles - Saint Quentin en Yvelines, Ambroise Paré Hospital (AP-HP), Boulogne-Billancourt, France

*** MD, PhD. Cardiology Department, University of Versailles - Saint Quentin en Yvelines, Ambroise Paré Hospital, Assistance Publique-Hôpitaux de Paris (AP-HP), Centre de référence des Maladies Cardiaques Héréditaires, Boulogne-Billancourt, France. UMR-EpReC.

**** MD. Radiology Department, University of Versailles - Saint Quentin en Yvelines, Ambroise Paré Hospital (AP-HP), Boulogne-Billancourt, France

***** Cardiology Department, University of Versailles - Saint Quentin en Yvelines, Ambroise Paré Hospital, Assistance Publique-Hôpitaux de Paris (AP-HP), Centre de référence des Maladies Cardiaques Héréditaires, Boulogne-Billancourt, France. UMR-EpReC.

A 45 year old patient, with a previous history of tobacco smoking, and treated hypertension, presented with hemoptysis on New Year's Eve. In the emergency room, his EKG showed a left bundle branch block with a heart rate of 110 bpm along with ST-segment abnormalities.

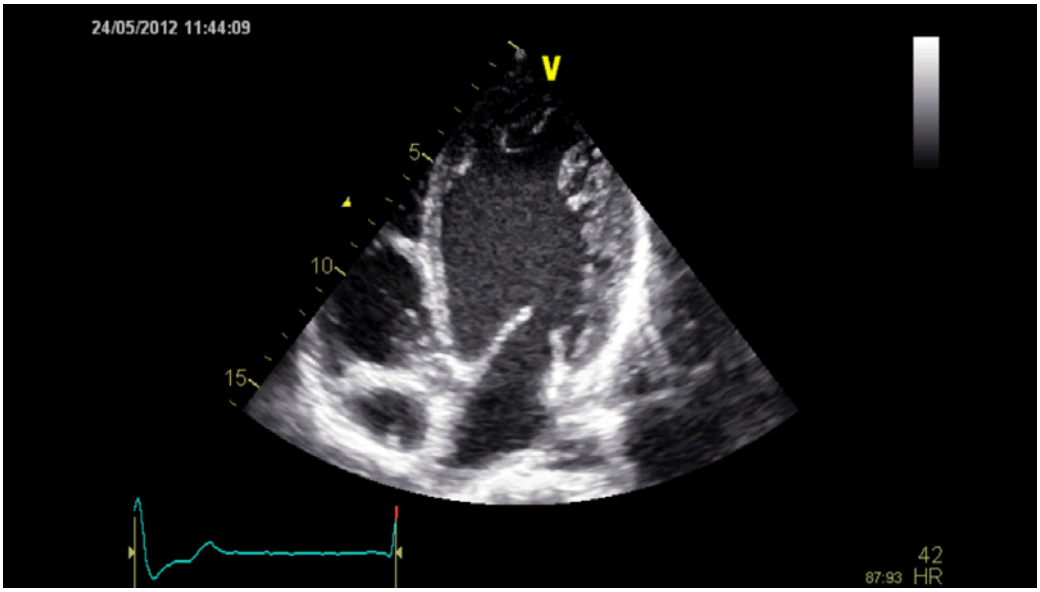
Blood troponin was in the normal range. He received amoxicillin and was referred to the cardiologist four days later. The physical examination was normal with a blood pressure (BP) of 130 over 80 mm Hg. He was scheduled for hospital day care to perform cardiac and pulmonary investigations.

The thoracic Computed Tomography and the bronchoscopy did not reveal any intrinsic pulmonary disease, except fragility and bleeding in the bronchial mucosa.

The ambulatory monitoring for BP showed satisfactory BP control. A 2D Echocardiography was performed and found a large left ventricular (LV) end diastolic diameter along with global hypokinesia and an impaired LV function with a LV ejection fraction (EF) of 35% calculated by Simpson's biplane method, a mild mitral insufficiency as well as a systolic pulmonary pressure of 41 mm Hg . It also showed prominent ventricular trabeculations in the apical and lateral LV wall and deep recesses extending from the LV cavity to the subendocardial surface of the LV wall (Figure 1). The coronary angiogram was normal.

DOI: http://dx.doi.org/10.17267/2317-3386bjmhh.v3i2.693

Figure 1 - Two D Echocardiography in the apical view shows prominent ventricular trabeculations in the apical and lateral LV wall and deep recesses extending from the LV cavity to the subendocardial surface of the LV wall, suggesting ILVNC.



An isolated left ventricular non-compaction (ILVNC) was suspected and a cardiac magnetic resonance (CMR) was indicated. It showed global hypokinesia, with an LV end diastolic volume of 239 ml and an LVEF of 24%. On steady-state free precession (SSFP) acquisitions, the apical and mid lateral LV wall showed multiple trabeculations with the presence of a distinct two-layered appearance of the myocardium, and a non-compacted to compacted myocardial ratio of >2.3 in diastole (Figure 2).

Figure 2 - Cardiac magnetic resonance images on steady-state free precession (SSFP) acquisitions in the axial (Figure 2 A) and short axis (Figure 2 B) show the presence of a distinct two-layered appearance of the myocardium, with a non-compacted to compacted myocardial ratio of >2.3 measured in diastole.

Figure 2A

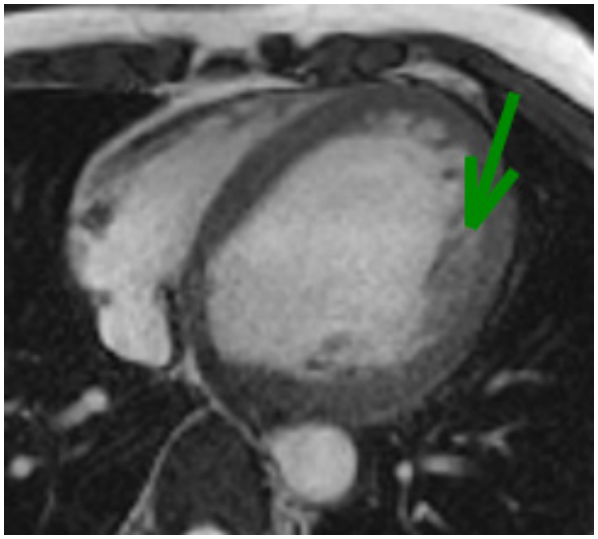
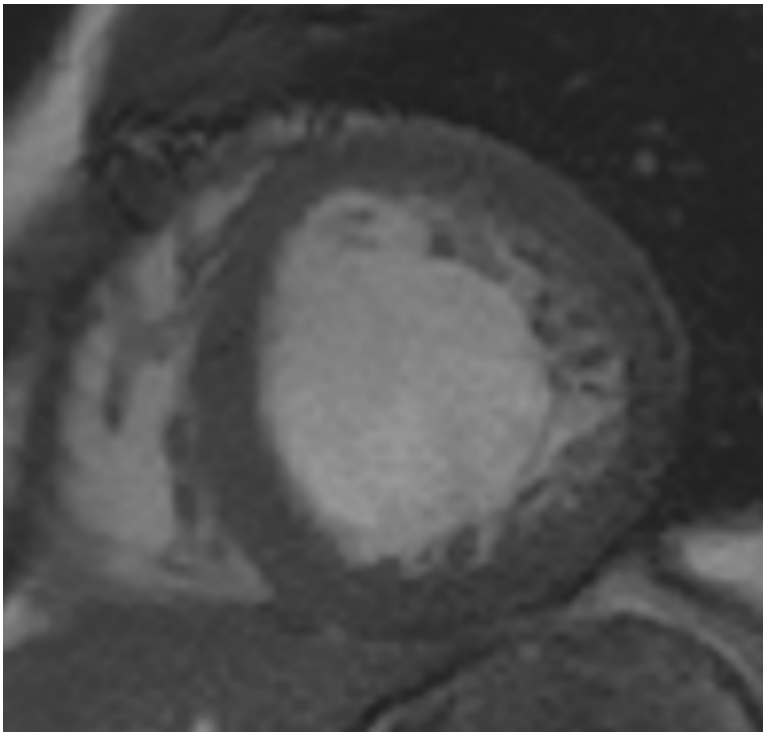


Figure 2B



The patient received heart failure medications, i.e. ramipril 5 mg daily and furosemide 40 mg daily. He did not tolerate beta blockers. Six months later, he was admitted for elevated brain natruretic peptide (BNP) level and a control 2D Echocardiography showed worsening of LVEF and elevated pulmonary artery systolic pressure of 60 mm Hg, without patent signs of heart failure. He received intravenous diuretics and beta blockers (nebivolol 5 mg) were initiated during hospital stay.

We discussed the use of a cardiac resynchronisation therapy with an implantable cardioverter defibrillator (ICD), in case of clinical or arrhythmic worsening.

Two years later, the ICD was not implanted, the patient was asymptomatic and the BNP level was normal. The patient was still receiving oral rampril, furosemide and nebivolol daily.

ILVNC is a myocardial disorder characterised by prominent ventricular trabeculations and deep recesses extending from the LV cavity to the subendocardial surface of the LV wall with or without LV dysfunction.[1, 2]

The American Heart Association classified LVNC as a primary genetic cardiomyopathy,[3] while the European Society of Cardiology refers to ILVNC as an 'unclassified cardiomyopathy',[4] possibly a morphological manifestation of several distinct cardiomyopathies.

The origin of ILVNC is unknown. The most widely believed hypothesis is the disturbance in compaction of myocardium during embryogenesis, between 5 and 10 weeks of gestation.

Widespread availability of cardiovascular imaging leads to increased diagnosis of ILVNC. However, there is poor agreement between the various diagnostic criteria[5] and a trend towards overdiagnosis.[6-8] Rarity of the disease as well as the absence of a non-pathological gold standard for diagnosis may explain the lack of large studies in the field.

Usually, the diagnosis of ILVNC is based on 2D echocardiography and requires the presence of prominent trabeculations with deep intertrabecular recesses communicating with the ventricular cavity, and a two-layered appearance to the myocardium (trabecular myocardium, and compacted myocardium).[1,2, 9-11]

Jenni et al criteria consist of four components that evolved over several publications.[1, 12,13] In the

absence of other cardiovascular disease, a two-layered myocardium with intertrabecular spaces filled by blood from the ventricular cavity visualised by colour Doppler is required. The mid-lateral and inferior walls and the apex were shown to be most commonly involved. A non-compacted to compacted segment ratio measured at the site of maximal wall thickness of >2.0 at any level at end-systole using a short-axis view was suggested as a parameter that differentiated ILVNC from patients with LV hypertrophy or dilated cardiomyopathy.

The use of echo contrast agent can aid visualisation of deep recesses in the trabeculated myocardium.

CMR has an emerging role in the diagnosis of ILVNC, especially when echocardiographic image quality is poor. Currently, two CMR diagnostic criteria exist. The first criteria published by Petersen et al[14] include the presence of a distinct two-layered appearance of the myocardium, with a non-compacted to compacted myocardial ratio of >2.3 measured using SSFP acquisitions, in diastole, perpendicular to the compacted myocardium in one of the three long-axis views at a location with the most prominent trabeculations. The maximal ratio should be used for the diagnosis.

However, the Petersen's criteria may result in overdiagnosis in low pretest probability population, and the trabecular mass percentage measurement can have low reproducibility.

Jacquier et al[15] suggested other diagnostic criteria using CMR. The LV trabecular mass was calculated on SSFP short-axis acquisitions, as the difference between the global LV mass (including the trabeculae) and the compacted LV mass (excluding the trabeculae) in end-diastole.

Several studies have shown the presence of myocardial fibrosis in the trabecular and compacted myocardium in up to 55% of patients with ILVNC using late gadolinium enhancement imaging (LGE),[16-17], that can also be used to detect the presence of intertrabecular thrombus.

Most patients identified as having LVNC are asymptomatic. However, symptoms include heart failure, arrhythmias and embolic events.

The most common, clinically significant arrhythmias in patients with ILVNC are ventricular tachycardia and atrial fibrillation. Thromboembolism may include the cerebrovascular, peripheral vascular and mesenteric systems.

In patients with LVNC, Oechslin et al [18] reported a prevalence for ILVNC of 0.014% in patients referred to the echocardiography laboratory.and a mortality of 35% over a mean follow-up period of 3.7 years. However, recent studies with similar mean follow-up periods report mortality in the range of 2% to 15%, suggesting that although the prognosis may be bad it is not as poor as originally described.[19,21]

Based on consensus guidelines,[22] asymptomatic patients with normal LV systolic function may be followed every 2 to 3 years with clinical assessment and echocardiography, as their prognosis is usually good. Asymptomatic patients with echocardiographic LV systolic and/or diastolic dysfunction should be treated with heart failure medications,[22] and followed every 1 to 2 years and symptomatic patients should be managed according to their clinical presentation.

The issue of ICD implantation in all LVNC patients has been proposed due to the high risk of sudden cardiac death, but is also highly controversial.[18] Implantation of an ICD is indicated for secondary prophylaxis in patients with sustained ventricular arrhythmia with haemodynamic compromise, and those with aborted sudden cardiac death.[23]

Finally, screening is recommended in asymptomatic first-degree relatives of patients diagnosed with ILVNC.[24]

REFERENCES

1. Jenni R, Oechslin E, Schneider J, et al. Echocardiographic and pathoanatomical characteristics of isolated left ventricular non-compaction: a step towards classification as a distinct cardiomyopathy. Heart 2001;86:666-71.
2. Stollberger C, Finsterer J, Blazek G. Left ventricular hypertrabeculation/noncompaction and association with additional cardiac

abnormalities and neuromuscular disorders. Am J Cardiol 2002;90:899-902.

3. Maron BJ, Towbin JA, Thiene G, et al. Contemporary definitions and classification of the cardiomyopathies: an American Heart Association Scientific Statement from the Council on Clinical Cardiology, Heart Failure and Transplantation Committee; Quality of Care and Outcomes Research and Functional Genomics and Translational Biology Interdisciplinary Working Groups; and Council on Epidemiology and Prevention. Circulation 2006;113:1807-16.
4. Elliott P, Andersson B, Arbustini E, et al. Classification of the cardiomyopathies: a position statement from the European Society Of Cardiology Working Group on Myocardial and Pericardial Diseases. Eur Heart J 2008;29:270-6.
5. Kohli SK, Pantazis AA, Shah JS, et al. Diagnosis of left-ventricular non-compaction in patients with left-ventricular systolic dysfunction: time for a reappraisal of diagnostic criteria? Eur Heart J 2008;29:89-95.
6. Kawel N, Nacif M, Arai AE, et al. Trabeculated (noncompacted) and compact myocardium in adults: the multi-ethnic study of atherosclerosis. Circ Cardiovasc Imaging 2012;5:357-66.
7. Stanton C, Bruce C, Connolly H, et al. Isolated left ventricular noncompaction syndrome. Am J Cardiol 2009;104:1135-8.
8. Habib G, Charron P, Eicher JC, et al. Isolated left ventricular non-compaction in adults: clinical and echocardiographic features in 105 patients. Results from a French registry. Eur J Heart Fail 2011;13:177-85.
9. Chin TK, Perloff JK, Williams RG, et al. Isolated noncompaction of left ventricular myocardium. A study of eight cases. Circulation 1990;82:507-13.
10. Stollberger C, Finsterer J. Left ventricular hypertrabeculation/noncompaction. J Am Soc Echocardiogr 2004;17:91-100.
11. Finsterer J, Stollberger C. No rationale for a diagnostic ratio in left ventricular hypertrabeculation/noncompaction. Int J Cardiol 2011;146:91-2.
12. Frischknecht BS, Jost Attenhofer CH, Oechslin EN, et al. Validation of noncompaction criteria in dilated cardiomyopathy, and valvular and hypertensive heart disease. J Am Soc Echocardiogr 2005;18:865-72.
13. Jenni R, Oechslin EN, van der Loo B. Isolated ventricular non-compaction of the myocardium in adults. Heart 2007;93:11-15.
14. Petersen SE, Selvanayagam JB, Wiesmann F, et al. Left ventricular non-compaction: insights from cardiovascular magnetic resonance imaging. J Am Coll Cardiol 2005;46:101-5.
15. Jacquier A, Thuny F, Jop B, et al. Measurement of trabeculated left ventricular mass using cardiac magnetic resonance imaging in the diagnosis of left ventricular non-compaction. Eur Heart J 2010;31:1098-104.
16. Dodd JD, Holmvang G, Hoffmann U, et al. Quantification of left ventricular noncompaction and trabecular delayed hyperenhancement with cardiac MRI: correlation with clinical severity. AJR Am J Roentgenol 2007;189:974-80.
17. Nucifora G, Aquaro GD, Pingitore A, et al. Myocardial fibrosis in isolated left ventricular non-compaction and its relation to disease severity. Eur J Heart Fail 2011;13:170-6.
18. Oechslin EN, Jost Attenhofer CH, Rojas JR, et al. Long-term follow-up of 34 adults with isolated left ventricular noncompaction: a distinct cardiomyopathy with poor prognosis. J Am Coll Cardiol 2000;36:493-500.
19. Aras D, Tufekcioglu O, Ergun K, et al. Clinical features of isolated ventricular noncompaction in adults long-term clinical course, echocardiographic properties, and predictors of left ventricular failure. J Card Fail 2006;12:726-33.
20. Lofiego C, Biagini E, Pasquale F, et al. Wide spectrum of presentation and variable outcomes of isolated left ventricular non-compaction. Heart 2007;93:65-71.
21. Murphy RT, Thaman R, Blanes JG, et al. Natural history and familial characteristics of isolated left ventricular non-compaction. Eur Heart J 2005;26:187-92.
22. Hunt SA, Abraham WT, Chin MH, et al. 2009 focused update incorporated into the ACC/

AHA 2005 Guidelines for the Diagnosis and Management of Heart Failure in Adults: a report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines: developed in collaboration with the International Society for Heart and Lung Transplantation. Circulation 2009;119:e391-479.

23. Kobza R, Steffel J, Erne P, et al. Implantable cardioverter-defibrillator and cardiac resynchronization therapy in patients with left ventricular noncompaction. Heart Rhythm 2010;7:1545-9.
24. Hershberger RE, Lindenfeld J, Mestroni L, et al. Genetic evaluation of cardiomyopathy–a Heart Failure Society of America practice guideline. J Card Fail 2009;15:83-97.