

## CASE REPORT

# An interesting case of cryptogenic stroke in a young man due to left ventricular non-compaction: role of cardiac MRI in the accurate diagnosis

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## SUMMARY

A 28-year-old man arrived for an outpatient cardiac MRI (CMR) study to evaluate cardiac structure. At the age of 24 the patient presented with acute onset expressive aphasia and was diagnosed with ischaemic stroke. Echocardiography at that time was reported as 'apical wall thickening consistent with apical hypertrophic cardiomyopathy'. CMR revealed a moderately dilated left ventricle with abnormal appearance of the left ventricular (LV) apical segments. Further evaluation was consistent with a diagnosis of LV noncompaction (LVNC) cardiomyopathy with a ratio of noncompacted to compacted myocardium measuring 3. There was extensive delayed hyperenhancement signal involving multiple segments representing a significant myocardial scar which is shown to have a prognostic role. Our patient, with no significant cerebrovascular risk factors, would likely have had an embolic stroke. This case demonstrates the role of CMR in accurately diagnosing LVNC in a patient with young stroke where prior echocardiography was nondiagnostic.

## BACKGROUND

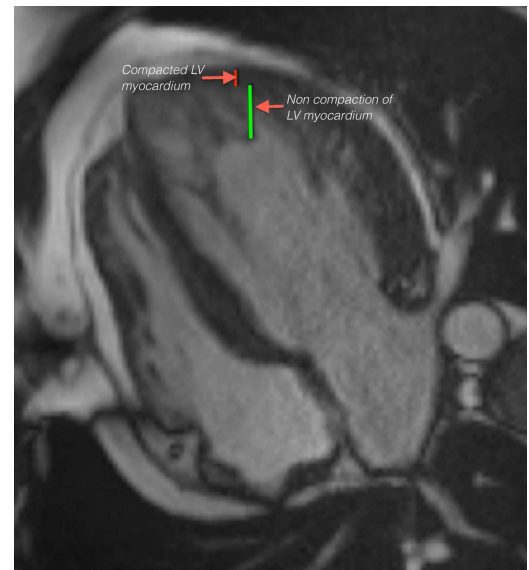
This case emphasises the importance of accurate diagnosis of left ventricular non-compaction (LVNC) as it is associated with poor prognosis. We believe that all patients with suspected cardiomyopathy should have a thorough work-up to make a proper diagnosis.

## CASE PRESENTATION

A 28-year-old man arrived for an outpatient cardiac MRI (CMR) study to evaluate cardiac structure. At the age of 24 the patient presented with an acute onset expressive aphasia. A CT of the head without contrast was obtained and the patient was diagnosed with an acute left middle cerebral artery territory ischaemic stroke. Echocardiography at that time was reported as 'apical wall thickening consistent with apical hypertrophic cardiomyopathy (HCM)', left atrial dilation and an ejection fraction (EF) of 55%. The patient was lost to follow-up for 4 years. He was later referred to our institution by his new primary care physician for CMR to evaluate cardiac structure and function. There was no family history of cardiomyopathy and sudden cardiac death.

## INVESTIGATIONS

CMR revealed a left ventricle that appeared moderately dilated with moderately reduced systolic function. LVEF was 38%. There was akinesia of the



**Figure 1** Four-chamber SSFP view showing the ratio of non-compacted to compacted of left ventricular myocardium of  $>2.3$  in diastole.

apical parts of the septal and anterior walls. An excessive amount of trabeculation was observed, consistent with a diagnosis of LVNC cardiomyopathy. The ratio of non-compacted to compacted myocardium in diastole measured 3 in inferior septum, anterolateral and apical inferior segments of the left ventricle (figure 1; video 1). A ratio more than 2.3 in diastole is consistent with LV non-compaction.<sup>1</sup> The ratio of trabeculated LV mass measured more than 22% compared with global LV mass. There was no evidence of LV thrombus in our patient. There was significant delayed hyperenhancement signal involving the anterior and inferior septum, anterolateral and apical inferior walls, representing significant myocardial scar/fibrosis in those areas (figure 2). Since the first echocardiogram was performed at an outside hospital a few years prior to his index presentation to our institution, we were unable to obtain the images.

## TREATMENT

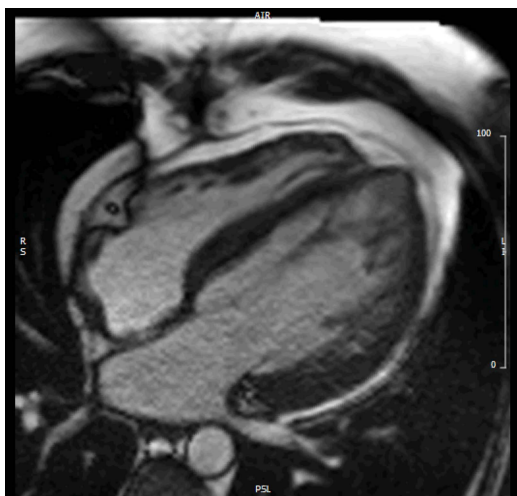
Since the patient remained asymptomatic, he was advised to follow-up with a cardiologist.

## OUTCOME AND FOLLOW-UP

The patient was advised to have a serial follow-up with a cardiologist. He was counselled regarding the necessity of future anticoagulation and



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**Video 1** 4 chamber (4CH) Steady-state free precession (SSFP) cine view revealing the decreased LV systolic function and left ventricular non-compaction.

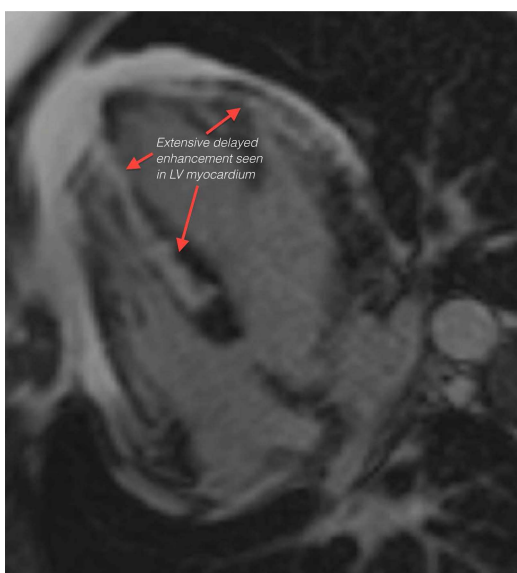
implantable cardioverter defibrillator (ICD) placement if his EF were to drop below 35%.

**DISCUSSION**

LVNC is categorised as an unclassified cardiomyopathy by WHO and as a genetic cardiomyopathy by the American Heart Association. LVNC (defined in the literature under different names including spongy myocardium or hypertrabeculation) is thought to be due to failure of myocardial maturation and, hence, failure of trabecular remodelling during embryogenesis.

Various cases of genetic (with an autosomal preponderance)<sup>2</sup> and sporadic forms of LVNC have been defined. The genetic mutations associated with LVNC were noted in the  $\alpha$ -dystrobrevin gene G4.5 (also known as the Tafazzin gene), CSX, FKBP12,  $\alpha$ -cardiac actin gene and in the absence of the Peg1 gene.

It is unclear whether the LVNC is a distinct cardiomyopathy or associated with other cardiomyopathies. The majority of them are associated with dilated cardiomyopathy followed by



**Figure 2** Four-chamber delayed enhancement images showing extensive delayed enhancement of left ventricular myocardium.

HCM and restrictive cardiomyopathy.<sup>3</sup> Coexisting apical HCM and LVNC have rarely been reported.<sup>4</sup> The association was thought to be due to common genetic mutations that occur in HCM and LVNC.<sup>5</sup>

Although the prevalence of LVNC is thought to be very low, it is increasingly understood that they are under-diagnosed as patients present late, once they are symptomatic. The most common reason for presentation is heart failure. However, the clinical symptoms vary depending on different phenotypes. They range from asymptomatic status to chest pain, shortness of breath and transient ischaemic attack symptoms. They may progress to heart failure, arrhythmias including ventricular tachycardia and systemic thromboemboli. Reports of cerebrovascular events, pulmonary embolism and LV thrombus have been reported.<sup>6</sup> LVNC is generally associated with poor long-term prognosis.<sup>7</sup>

Initial clues to the diagnosis of LVNC are usually from transthoracic echocardiography (box 1). Although various echocardiographic criteria have been suggested for diagnosis, the accuracy of diagnosis improves when these are used in conjunction.<sup>8</sup> CMR provides a more accurate visualisation in distinguishing myocardial trabeculations from non-compacted LV mass.<sup>1</sup> It has improved the yield of diagnosis for those on whom prior testing was non-diagnostic.<sup>9</sup> Various CMR criteria (box 1) have been proposed including trabeculated LV mass >20% of the global LV mass and ratio of non-compacted to compacted of >2.3 in diastole,<sup>1</sup> but they need to be validated in a large population as they have varied specificity.<sup>10</sup> Late gadolinium enhancement, as observed in our patient, is found in 40% of patients with LVNC. The reason for such enhancement is not clearly understood. Jenni<sup>7</sup> noted that coronary flow reserve, an indirect measure of coronary circulation, is decreased in segments with wall motion abnormalities but not in non-compacted segments. The decrease in coronary microcirculation is thought to cause delayed enhancement in gadolinium images.

**Box 1 Various echocardiographic and cardiac MRI (CMR) criteria proposed in the diagnosis of left ventricular noncompaction (LVNC)**

*Echocardiographic criteria for diagnosis of LVNC*

- ▶ Chin *et al*<sup>13</sup> LVNC is defined by a ratio of distance from the epicardial surface to the trough of the trabecular recess/ distance from the epicardial surface to peak of trabeculations  $\leq 0.5$ .
- ▶ Jenni *et al*<sup>14</sup>
  - A two-layer structure with a thin compacted layer and a thick non-compacted layer measured in end systole at parasternal short-axis views.
  - LVNC is defined by a ratio of non-compacted layer of myocardium/compacted layer of myocardium >2.
- ▶ Stollberger *et al*<sup>15</sup>
  - > Three trabeculations protruding from the left-ventricular wall.
  - Trabeculations perfused from the ventricular cavity.

*CMR criteria for diagnosis of LVNC*

- ▶ Peterson *et al*<sup>1</sup> A ratio of non-compacted layer of myocardium/compacted layer of myocardium >2.3 at long axis SSFP cine images at diastole.
- ▶ Jacquier *et al*<sup>16</sup>
  - A trabeculated LV mass greater than 20% of global LV mass.

Such enhancement is of importance as recent studies have reported the prognostic value of delayed enhancement in cardiomyopathies.<sup>11</sup> But it should be noted that the prevalence of gadolinium enhancement in LVNC is quite variable. This may well be attributed to underlying heterogeneity in genotypes.<sup>12</sup>

Owing to its genetic preponderance, a thorough family history of  $\geq 3$  generations is recommended by the Heart Failure Society. If a familial pattern is suspected or confirmed, pedigree analysis is suggested. If familial LVNC is suspected based on clinical or genetic testing, frequent screening is recommended in asymptomatic first-degree relatives.

The treatment of LVNC is dictated by clinical features and LV function, as there is no specific therapy for LVNC. Patients with heart failure or arrhythmias are treated according to standard guidelines. Also, ICDs are placed depending on indications such as systolic dysfunction or history of sudden cardiac death. Although current guidelines do not support any clinical intervention in LVNC without systolic dysfunction, these patients should be kept under close surveillance to monitor and address complications.

Since there is a well-recognised association of thromboembolic phenomena in LVNC, we postulate that our patient, with no significant cerebrovascular risk factors, would likely have had an embolic event resulting in his prior stroke.

In summary, we report a case of LVNC in a patient originally diagnosed as 'apical HCM' and a prior stroke. The diagnosis of LVNC was confirmed on CMR, which also demonstrated significant delayed enhancement in the trabeculae of the non-compacted myocardium, which has prognostic value. Our case emphasises the importance of multimodality imaging in evaluating patients with suspected cardiomyopathy. There is a growing wealth of evidence that CMR is helpful in diagnosing cardiomyopathies on whom prior echocardiograms were non-diagnostic. Therefore, it would be worthwhile to consider CMR as a part of the standard diagnosing modality in patients with cardiomyopathy.

### Learning points

- ▶ We present an interesting case of stroke thought to be due to thromboembolic phenomena associated with left ventricular non-compaction.
- ▶ The case emphasises the importance of prompt recognition of left ventricular non-compaction as it is associated with poor prognosis.
- ▶ Our case also demonstrates the role of multimodality imaging, including cardiac MRI, which is required for appropriate diagnosis of suspected cardiomyopathy.

**Contributors** AK wrote the case report and discussion, RJ provided critical revision of the manuscript, MRI and final approval, and AD wrote the introduction and proofread the report.

**Competing interests** None.

**Patient consent** Obtained.

**Provenance and peer review** Not commissioned; externally peer reviewed.

### REFERENCES

- 1 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.
- 2 Sasse-Klaassen S, Gerull B, Oechslin E, *et al.* Isolated noncompaction of the left ventricular myocardium in the adult is an autosomal dominant disorder in the majority of patients. *Am J Med Genet A* 2003;119A:162–7.
- 3 Biagini E, Ragni L, Ferlito M, *et al.* Different types of cardiomyopathy associated with isolated ventricular noncompaction. *Am J Cardiol* 2006;98:821–4.
- 4 Alizadeh-Sani Z, Madadi S, Sadeghpour A, *et al.* Cardiac MRI in a patient with coincident left ventricular non-compaction and hypertrophic cardiomyopathy. *J Tehran Heart Cent* 2011;6:214–16.
- 5 Monserrat L, Hermida-Prieto M, Fernandez X, *et al.* Mutation in the alpha-cardiac actin gene associated with apical hypertrophic cardiomyopathy, left ventricular non-compaction, and septal defects. *Eur Heart J* 2007;28:1953–61.
- 6 Oechslin EN, Attenhofer Jost 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.
- 7 Jenni R, Wyss CA, Oechslin EN, *et al.* Isolated ventricular noncompaction is associated with coronary microcirculatory dysfunction. *J Am Coll Cardiol* 2002;39:450–4.
- 8 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.
- 9 Duncan RF, Brown MA, Worthley SG. Increasing identification of isolated left ventricular non-compaction with cardiovascular magnetic resonance: a mini case series highlighting variable clinical presentation. *Heart Lung Circ* 2008;17:9–13.
- 10 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.
- 11 Green JJ, Berger JS, Kramer CM, *et al.* Prognostic value of late gadolinium enhancement in clinical outcomes for hypertrophic cardiomyopathy. *JACC Cardiovasc Imaging* 2012;5:370–7.
- 12 Wan J, Zhao S, Cheng H, *et al.* Varied distributions of late gadolinium enhancement found among patients meeting cardiovascular magnetic resonance criteria for isolated left ventricular non-compaction. *J Cardiovasc Mag Reson* 2013;15:20.
- 13 Chin TK, Perloff JK, Williams RG, *et al.* Isolated noncompaction of left ventricular myocardium. A study of eight cases. *Circulation* 1990;82:507–13.
- 14 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.
- 15 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.
- 16 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.

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