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## Rare disease

# Non-compaction of the left ventricle and associated ventricular septal defect

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## Summary

A case report of a 28-year-old patient, who presented with symptoms and signs of congestive heart failure and had clinical signs of ventricular septal defect as well. On further work-up echocardiogram showed non-compaction of the left ventricle with severe left ventricular systolic dysfunction and a ventricular septal defect. He was treated with standard treatment of heart failure and is doing well.

## BACKGROUND

Left ventricular non-compaction (LVNC) also called hypertrabeculation syndrome or spongy myocardium is a rare disorder. It is classified as a primary genetic cardiomyopathy by the American Heart Association (AHA).<sup>1</sup> Isolated LVNC can be either sporadic or familial and very rarely can occur as a transient phenomenon during myocarditis. The prevalence of LVNC in the general population is not known but a review from Switzerland identified 34 cases within 15 years, which represented 0.014% of echocardiographic studies done over a 15-year period.<sup>2</sup> This may be an underestimate, since improved echocardiographic image quality and increasing awareness of this rare disorder will likely lead to enhanced recognition. Non-compacted myocardium is occasionally found accompanying other congenital cardiac anomalies such as ventricular septal defect (VSD).

Our case report is a very rare clinical entity that will enhance awareness about its echocardiographic recognition.

## CASE PRESENTATION

A 28-year-old man with no known comorbidities presented in clinic with complaints of breathlessness, swelling of feet and palpitations for 6 months. He had exertional breathlessness which had progressed and he was getting short of breath on just walking for approximately 5 min. He had paroxysmal nocturnal dyspnoea for 4 months and orthopnoea for last 2 weeks. He complained of occasional palpitations which had become more frequent for 1 week. He denied chest pain, fever or flu-like symptoms and had no history of heart problems. His family history was negative for premature coronary artery disease, heart failure, hypertension, diabetes mellitus and sudden cardiac death (SCD).

On examination, he was a young man lying comfortably in bed with a pulse of 110 beats/min regular, blood pressure of 125/80 mm Hg, raised jugular venous pressure and pitting pedal oedema. Apex beat was palpable in sixth intercostal space in anterior axillary line with no thrills or parasternal heave. A pansystolic murmur was audible at lower-left sternal edge. Bilateral crackles were

audible at the lung bases but no cyanosis or clubbing was present. Clinical impression of decompensated heart failure and VSD was made. Treatment was started with diuretics, low-dose carvedilol and candesartan.

## INVESTIGATIONS

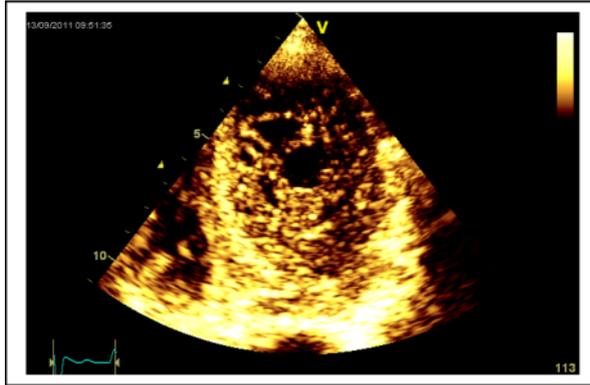
Echocardiogram showed spongiform cardiomyopathy with severe LV systolic dysfunction (LV ejection fraction, LVEF approximately 20%) and global hypokinesia. A small perimembranous VSD was seen measuring 5 mm with left to right shunt, a peak gradient of 60 mm Hg and QP:QS ratio of 1.3 (restrictive VSD). Right ventricular systolic function was normal. There was grade III LV diastolic dysfunction and a retracted posterior mitral valve leaflet with moderate eccentric mitral regurgitation. Holter monitor done for palpitations showed sinus tachycardia correlating with time of symptoms recorded in patient's diary. The rest of baseline reports were normal.

## OUTCOME AND FOLLOW-UP

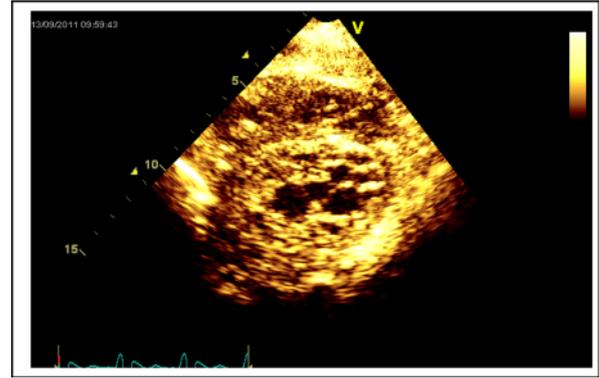
The patient was seen in the clinic for follow-up. Spironolactone and aspirin were added, doses of carvedilol, furosemide and candesartan were optimised and the patient was counselled regarding need for an implantable cardiac defibrillator (ICD) for primary prevention of SCD. Six months following diagnosis, the patient has remained stable on medical treatment.

## DISCUSSION

LVNC is a rare cardiac disorder, classified as a primary genetic cardiomyopathy by the AHA.<sup>1</sup> The European Society of Cardiology classified LVNC as an unclassified cardiomyopathy.<sup>3</sup> LVNC is characterised by an altered ventricular myocardium containing trabeculae and deep intertrabecular recesses resulting in thickened myocardium with two layers consisting of non-compacted and compacted myocardium. Also having direct communication between the LV cavity and the deep intratrabecular recesses, which are filled with blood from the LV cavity without connection to the epicardial coronary arteries (figure 1). Non-compacted myocardium is occasionally



**Figure 1** Echocardiographic image, left parasternal short-axis view at apical level.



**Figure 2** Echocardiographic image, left parasternal short-axis view at papillary muscle level.

found accompanying other congenital cardiac disorders, like bicuspid aortic valve, aorta-to-LV tunnel, Ebstein's anomaly, congenitally corrected transposition, hypoplastic left heart syndrome and isomerism of the left atrial appendage.<sup>4</sup> Non-compacted myocardium has also been seen in patients with atrial and VSDs as was the case in our patient, patent ductus arteriosus<sup>5</sup> and in cardiomyopathies due to neuromuscular disorders. LVNC can occur in genetic syndromes and metabolic diseases including Charcot-Marie-Tooth disease 1A, Barth syndrome and Melnick-Needles syndrome, as well as nail-patella syndrome. It has been postulated that LVNC may be due to intrauterine arrest of compaction of the loose interwoven meshwork or pronounced hypertrabeculation may be due to altered regulation in cell proliferation, differentiation and maturation during LV wall formation.<sup>6</sup> The prevalence of LVNC in the general population is not known but has been described among patients undergoing echocardiographic studies. A review from Switzerland identified 34 cases within 15 years, which represented 0.014% of echocardiograms that were performed.<sup>7</sup> This may be an underestimation, since improved echocardiographic image quality and increasing awareness of LVNC will perhaps lead to enhanced recognition of LVNC. LVNC can be either sporadic or familial. In various reports, 12–50% of patients with LVNC had a family history positive of this condition.<sup>7</sup> Mutations have been identified in at least nine genes in LVNC patients including genes encoding LIM domain-binding protein 3 (LDB3),  $\alpha$ -dystrobrenin (DTNA), tafazzin (TAZ), lamin A/C (LMNA),  $\beta$ -myosin heavy chain (MYH7),  $\alpha$ -cardiac actin (ACTC), cardiac troponin T (TNNT2), SCN5A and tropomyosin 1 (TPM1).<sup>8,9</sup> The main clinical presentations of LVNC are congestive cardiac failure, atrial and ventricular arrhythmias and thromboembolic events including stroke.<sup>7</sup> The ECG is usually abnormal but there are no characteristic changes.<sup>7</sup> ECG abnormalities that can be seen include right or left bundle branch block, fascicular block, atrial fibrillation (AF) and ventricular tachycardia.

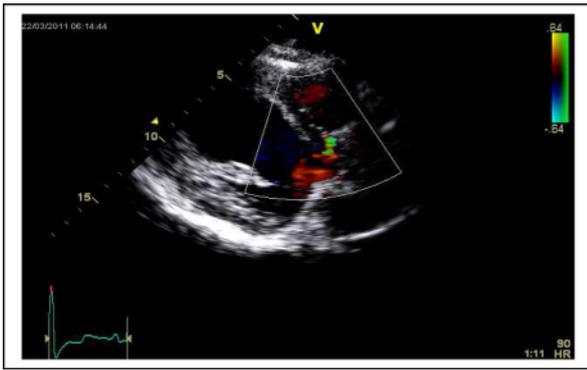
The diagnosis of LVNC is usually established by echocardiography. Cardiovascular MRI, cardiac CT and left ventriculography are other imaging modalities that may be diagnostic or raise the initial clinical suspicion. Echocardiography has been utilised both to establish the diagnosis and as an aid during patient follow-up.<sup>2,7</sup>

However, there is no universally accepted definition of LVNC. Proposed echocardiographic criteria for LVNC are based on observations from different centres. Jenni *et al* proposed the criteria which included: (1) a thickened LV wall consisting of two layers: a thin compacted epicardial layer; and a markedly thickened endocardial layer with numerous prominent trabeculations and deep recesses with a maximum ratio of non-compacted to compacted myocardium  $>2:1$  at end-systole in the parasternal short-axis view, (2) colour Doppler evidence of flow within the deep inter-trabecular recesses and (3) prominent trabecular meshwork in the LV apex or mid-ventricular segments of the inferior and lateral wall. All three echocardiographic criteria are required for diagnosis and the criteria are assessed in the parasternal short-axis views at base, mid- and apical levels.<sup>7</sup> Other findings that can be seen on echocardiography include reduced global LV systolic function, diastolic dysfunction, LV thrombi and abnormal papillary muscle structure (figures 2 and 3).<sup>7</sup>

The echocardiographic image of isolated LVNC can be very heterogeneous including dilated forms, hypertrophic variant and restrictive types (figures 4 and 5).<sup>10</sup> Published series have found that LVNC is linked with high rates of morbidity and mortality in adults. In a study of 34 patients (mean age 42) the probability of survival free of death or heart transplantation at 5 years was 58%.<sup>7</sup>

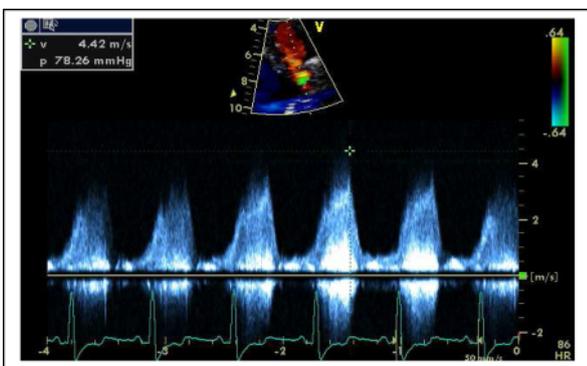


**Figure 3** Echocardiographic image, left parasternal short-axis view at midleft ventricular cavity level with colour Doppler.



**Figure 4** Echocardiographic image, left parasternal long axis view with colour Doppler showing ventricular septal defect.

However, this study population represents a group of severely affected patients with a poor prognosis but hopefully with the increasing awareness of LVNC, more subtle forms in minimally symptomatic patients or severe forms in asymptomatic patients will be detected, which may change the prognosis. Clinical data on treatment of LVNC are limited, and there is no specific therapy for LVNC. Medical management depends on clinical manifestations, LVEF, the presence or absence of arrhythmias and perceived risk of thromboembolism. LVNC patients with reduced LVEF, heart failure and asymptomatic systolic dysfunction are treated according to standard guidelines. LVNC patients with or without AF are at high risk for thromboembolism. In addition, given the high thromboembolic risk, chronic anticoagulation therapy is recommended in patients with LVNC and AF who do not otherwise have an indication for anticoagulation. Anticoagulation is also recommended in patients with LVNC without AF with LVEF <40%. LVNC patients should be advised to refrain from competitive endurance sports or weight lifting. LVNC patients should receive ICD therapy according to standard indications for ICD therapy in patients with non-ischaemic cardiomyopathy. Patients with LVNC who have end-stage heart failure are candidates for cardiac transplantation evaluation.



**Figure 5** Echocardiographic image with continuous wave Doppler showing pressure gradient across the ventricular septal defect.

**Learning points**

- ▶ Left ventricular non-compaction (LVNC) is a rare sporadic or familial cardiomyopathy.
- ▶ The clinical presentations and prognosis of such patients are variable; including heart failure, chest pain, thromboembolic events and atrial and ventricular arrhythmias.
- ▶ LVNC patients who have end-stage heart failure are candidates for cardiac transplantation evaluation.

**Competing interests** None.

**Patient consent** Obtained.

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