

Isolated non-compaction of the left ventricle

V Y Kshirsagar, MD (Pediatrics); M Ahmed, MD (Pediatrics); S M Colaco, MBBS; S V Patil, MBBS

Krishna Institute of Medical Sciences, Deemed University, Karad, India

Corresponding author: V Y Kshirsagar (drkshirsagarvy@yahoo.com)

Isolated left ventricular non-compaction (LVNC) is a rare congenital disorder caused by failure of normal embryogenesis of the endocardium and myocardium *in utero*, resulting in prominent ventricular trabeculations with deep intertrabecular recesses. Patients may be asymptomatic or may present with refractory cardiac failure. The best modality of diagnosis is based on echocardiographic diagnostic criteria. Prognosis remains poor for patients with impaired systolic left ventricular function, as treatment options are very limited. We report the case of 2-month-old infant presenting with congestive cardiac failure diagnosed as isolated LVNC on echocardiography.

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Left ventricular non-compaction (LVNC) is a congenital dysfunction of ventricular morphogenesis. The World Health Organization has categorised it as an unclassified cardiomyopathy.^[1] It is thought to be the result of an arrest in the normal endomyocardial embryogenesis, which leads to the persistence of intertrabecular recesses and the development of a spongy myocardium.^[2] LVNC is now being diagnosed with increasing frequency, either in isolation or combined with congenital heart disease or neuromuscular disorders.^[3] Here we report the case of a 2-month-old male infant who presented with congestive heart failure (HF) where isolated LVNC was found on echocardiography.

Case report

A 2-month-old male infant was admitted to the paediatric intensive care unit (ICU) of Krishna Hospital, Karad, with shortness of breath and respiratory distress which commenced one day prior. The baby was born preterm at 30 weeks of gestation to non-consanguineous parents and was admitted for preterm care in the neonatal ICU. The hospital stay was unremarkable and the infant was discharged at a weight of 1.6 kg after 21 days. The baby was asymptomatic thereafter.

On admission the patient had features of congestive HF. Cardiovascular examination revealed a decreased volume peripheral pulse and no obvious murmur was appreciated. There was hepatomegaly 4 cm below the right costal margin. No dysmorphic features were observed. A chest X-ray revealed substantial cardiomegaly. Electrocardiography showed left ventricular overload, but no signs of ventricular arrhythmias. No neuromuscular abnormality was found.

An echocardiogram showed situs solitus, left aortic arch and atrioventricular-ventriculoarterial concordance; pulmonic veins drained normally to the left atrium and inferior vena cava to the right atrium. The inter-atrial and interventricular septums were intact. The left ventricle (LV) was markedly dilated with a severely decreased global systolic function (ejection fraction 30%). Hypertrabeculation was evident with multiple crypts at the apical posterolateral aspects of the LV (Fig. 1). Colour Doppler examination showed prominent LV trabeculations with deep intertrabecular recesses in continuity with the LV cavity. The ratio of non-compacted to compacted layer of the myocardium was >2 (Fig. 2).

The patient was treated with inotropes, decongestive therapy and captopril. His condition stabilised within 1 week; he was discharged and prescribed captopril and frusemide, and advised to follow-up regularly.

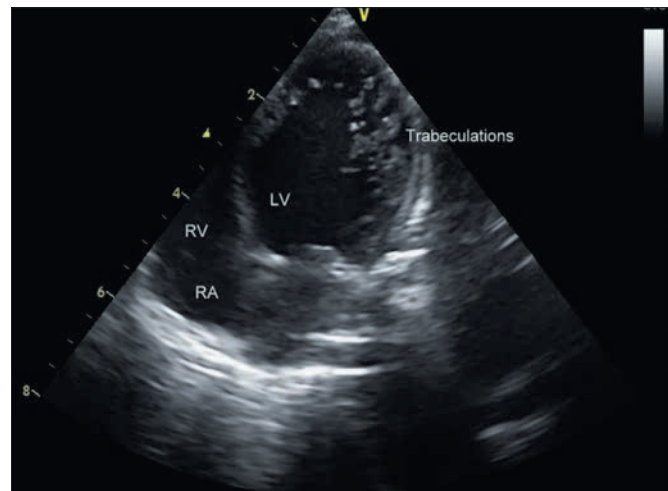


Fig. 1. Hypertrabeculations on the apical and posterolateral aspect of the left ventricle. (LV = left ventricle; RV = right ventricle; RA = right atrium.)

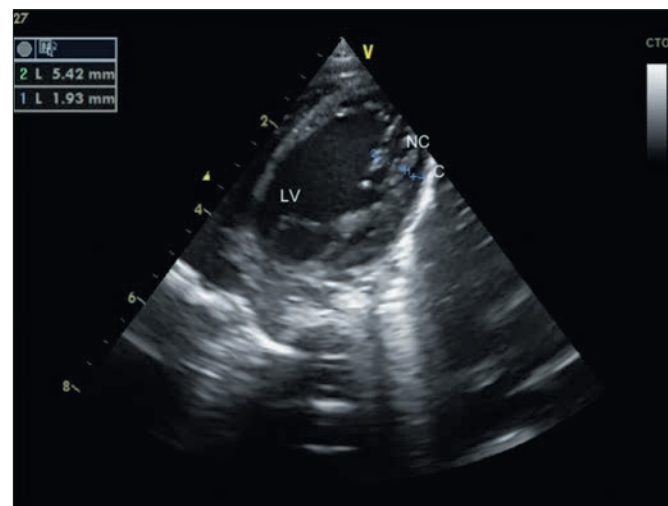


Fig. 2. Ratio of compacted to non-compacted myocardium. (LV = left ventricle; NC = non-compacted; C = compacted.)

Discussion

Non-compaction of the myocardium was first described by Bellet *et al.*, from an autopsy carried out on a newborn in whom aortic

atresia and coronary-ventricular fistula were also observed. In 1984, Engberding *et al.* reported a case with a diagnosis of isolated non-compaction, a condition characterised by the absence of other associated cardiopathies.^[4] Although it is a well-known pathology in adults, there are only a few case reports and series in the literature regarding LVNC in children.

The inheritance of isolated LVNC may be sporadic or familial. In familial cases, autosomal dominant inheritance is more common than X-linked inheritance.^[5] To date, mutations in seven different genes have been found; however, at present, gene G4.5 (*TAZ* gene) on the Xq28 chromosomal region identified in neonatal isolated LVNC is the only confirmed disease-causing locus.^[6] Owing to a lack of facilities, our patient could not undergo genetic testing.

LVNC represents the persistence of multiple trabeculations in the ventricular myocardium with deep intratrabecular spaces due to arrested compaction of the wall. LVNC is associated with a broad spectrum of clinical and pathophysiological findings with an unclear natural history.^[7] The clinical presentation may range from an incidental echocardiographic diagnosis without symptoms to disabling HF. The three most common clinical presentations include HF, arrhythmias and embolic events. Additional presenting symptoms include chest pain and syncope. Our patient presented with HF as seen in 67% of cases. Arrhythmia, rarer in children, manifests as ventricular tachyarrhythmia, atrial fibrillation, left bundle branch block, paroxysmal supraventricular tachycardia, complete heart block and Wolf-Parkinson-White syndrome.^[8]

Echocardiography is the diagnostic tool of choice for LVNC; however, different echocardiographic criteria are currently been used.^[11] Jenni *et al.*^[9] have described criteria that include: (i) the absence of co-existing cardiac abnormalities; (ii) a two-layered structure of the ventricular wall with an end-systolic ratio of non-compacted to compacted layer of >2 ; (iii) the finding of this morphological presentation in apical and mid-ventricular areas; and (iv) direct blood flow from the ventricular cavity into the deep intertrabecular recesses, as assessed by Doppler echocardiography. Cardiac magnetic resonance imaging (MRI) has been used in the developed world for confirmation of the diagnosis where a non-compacted to compacted myocardium layer ratio >2.3 would reliably diagnose the disease entity.^[11] Our diagnosis was made entirely on echocardiography findings, but the use of cardiac MRI can confirm the diagnosis.

The prognosis for patients with LVNC is highly variable; the spectrum ranges from a prolonged, asymptomatic course to rapid, progressive HF. Based on a paediatric case series, LV dysfunction inevitably develops in patients with LVNC over a 10-year period,

regardless of the presence of symptoms at the time of diagnosis.^[6] The most common causes of death are intractable HF and sudden cardiac death.

The treatment of LVNC consists of management of HF and arrhythmias, and oral anticoagulation to prevent thromboembolic episodes. The use of beta-blockers such as carvedilol has proved to have beneficial effect on LV function. Cardiac transplantation is advised in patients with refractory HF. The use of long-term anti-thrombotics and intracardiac defibrillation is also advisable.^[10] Our patient was stabilised and continued treatment with captopril and frusemide. Regular follow-up was advised and the parents were educated to assess for the danger signs of worsening HF.

Conclusion

Isolated LVNC is a rare, genetic, congenital disorder. Due to easy availability of diagnostic modalities and awareness of this new type of cardiomyopathy, early recognition and prompt treatment are possible. Patients should also be screened for thromboembolism and arrhythmias. Due to the genetic transmission of LVNC, screening of close relatives is recommended, and the screening of the patient for neuromuscular diseases is also advised.

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