

Cardiac magnetic resonance imaging in the diagnosis of biventricular non-compaction in a young but failing heart

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A 15-year-old Chinese girl with a history of scoliosis presented to Queen Mary Hospital, Hong Kong in April 2016 with an incidental finding of an ejection systolic murmur at the left lower sternal border. No history of chest pain, syncope, reduced effort tolerance, or significant family history of congenital cardiac disease was present. Echocardiography revealed heavy trabeculations over the left ventricular (LV) apical region. Colour Doppler revealed abnormal in-and-out flow at the deep crypts. Overall features were suspicious of LV non-compaction (NC). The patient subsequently underwent cardiac magnetic resonance imaging for further assessment, using a 1.5-T magnetic resonance imaging scanner (Magnetom Aera; Siemens Healthcare, Forchheim, Germany). Cardiac magnetic resonance confirmed the diagnosis of biventricular NC with diffuse involvement (Fig 1). The ratio of non-compacted to compacted diastolic myocardium was 2.95 (>2.3). The LV ejection fraction (EF) was 32.6%, and the right ventricular (RV) EF was 32.5%. Moderate global hypokinesia of both ventricles was observed. Mild mitral regurgitation was present. No significant left to right cardiac shunt or late gadolinium enhancement was seen at the LV wall to suggest presence of scar or fibrosis (Fig 2).

No abnormal thinning of the wall, focal/regional RV wall motion abnormality, or aneurysmal change was noted. No thrombus was present within the cardiac chambers. A normal configuration of a left-sided aortic arch was observed with absence of coarctation. Subsequent genetic testing for pathogenic mutations for NC was negative in this patient.

Ventricular NC of the myocardium, also known as spongiform cardiomyopathy, is a rare cardiomyopathy arising from arrested endomyocardial development during embryogenesis,¹ with an incidence of approximately 0.05%.² Non-compaction is a group of genetically heterogeneous disorders and can be inherited in autosomal dominant, autosomal recessive and X-linked recessive pattern.³ However, the majority of NC have idiopathic pathogenesis, and the diagnostic yield of gene panel testing in LVNC is low (~9%). Patients with isolated NC are less likely to have a positive genetic test result.³ Morphologically, NC is characterised by an altered myocardial wall with resultant prominent trabeculae and deep intertrabecular recesses,⁴ leading to an abnormal thickened bilayer of compacted and non-compacted myocardium. The LV is more frequently involved and biventricular involvement is less commonly encountered. The absence of wall thinning, RV wall motion abnormality or aneurysmal change, although not diagnostic, suggests an alternative diagnosis to that of arrhythmogenic RV dysplasia. Principal clinical manifestations of NC include: heart failure, arrhythmia, cardioembolic events, syncope, and sudden cardiac death.⁴ Even though our patient had remained asymptomatic prior to diagnosis, there were notable reductions in LVEF and RVEF, suggesting heart failure.

Cardiac magnetic resonance in establishing suspected NC cases would be crucial in: (1) confirming the diagnosis, (2) establishing residual cardiac function, and (3) determining presence of other associated cardiac malformations, such as LV outflow tract abnormalities (eg, bicuspid aortic valve), Ebstein anomaly, tetralogy of Fallot (more commonly diagnosed at a younger age-group) or coarctation of the aorta. Cardiac magnetic resonance is also superior to echocardiography in delineating RV anatomy and function, evaluating RV involvement of NC, determining presence of intracardiac thrombus and myocardial scarring. After the above diagnostic considerations have been addressed, management

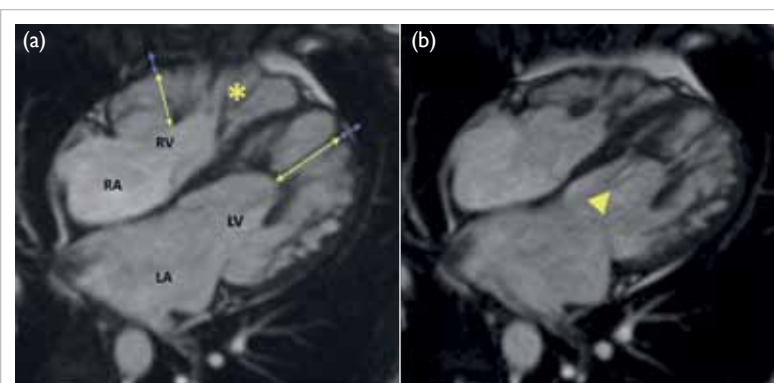


FIG 1. (a) Four-chamber steady-state free precession cine cardiac magnetic resonance image at end-diastole showing biventricular non-compaction in a 15-year-old girl. Prominent and excessive trabeculations (yellow asterisk) are observed at the non-compacted layer (yellow arrows). The ratio of non-compacted to compacted myocardium (blue arrows) at end-diastole measured >2.3, confirming the diagnosis of biventricular non-compaction. (b) Four-chamber image at systole showing the presence of a "jet" (yellow arrowhead) at the left ventricle near the non-compacted layer, mirroring turbulent in-and-out flow seen on echocardiography

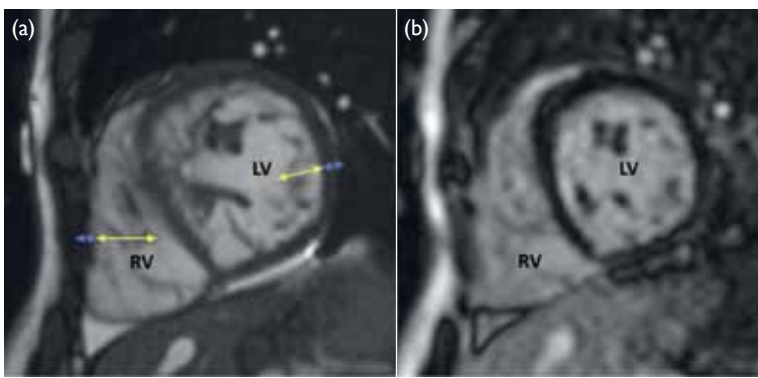


FIG 2. (a) Short-axis cardiac magnetic resonance image at the mid-ventricular level showing biventricular non-compaction. (b) No late gadolinium enhancement is noted at the left ventricular wall to suggest the presence of scar tissue or fibrosis

of biventricular NC may include anticoagulation, treatment of heart failure, and the placement of implantable cardioverter defibrillator or pacemaker where clinically appropriate. However, cardiac transplantation remains as the only definitive treatment of biventricular NC.

Author contributions

All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity. All authors contributed to the concept, image acquisition, image and data interpretation, drafting of the article, and critical revision for important intellectual content.

Conflicts of interest

The authors have no conflicts of interest to disclose.

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Ethics approval

Patient consent was obtained for the purpose of this case report.

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