Two Patients with Coincident Noncompacted Myocardium and Hypertrophic Cardiomyopathy

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Summary

To our knowledge, left ventricular noncompaction (LVNC) and hypertrophic cardiomyopathy (HCM) commonly occur as separate disorders in different patients; however, LVNC associated with HCM, which is called hypertrophic LVNC, is relatively rare.1) Here we report two sporadic cases of hypertrophic LVNC which were diagnosed by echocardiography and cardiac magnetic resonance (CMR).

Key words: Primary cardiomyopathy, Cardiac magnetic resonance

Left ventricular noncompaction is a rare genetic cardiomyopathy that affects both children and adults. Its prevalence in adults is 0.014%,2) and it rarely coexists with other cardiomyopathies. Previous studies have reported only case reports or small cohorts of LVNC which coexisted with cardiomyopathies.1,3-5) This is the first study to report the prevalence of this mixed phenotype. Relatively, in the hybrid cardiomyopathies, hypertrophic LVNC was less than LVNC associated with dilated cardiomyopathy.5) Currently, diagnosis of cardiomyopathies depends on non-invasive imaging studies—transthoracic echocardiography and CMR.6,7)

Case Reports

Case 1: An 18-year-old man presented clinically with chest distress and polypnea for three years. His family history was negative. The positive signs were as follows: the apical impulse increased, cardiac dullness boundary expanded, and a systolic grade III blowing murmur which was detected in the second auscultation aortic area. In addition, the hepatojugular reflux sign was positive. The electrocardiogram revealed pre-excitation syndrome. The laboratory results were non-specificity.

Echocardiography revealed the following: left ventricular (LV) dilatation while other atrioventriculars were normal; interventricular septal (IVS) hypertrophy with a thickness of 18 mm at end of diastole; prominent trabecular network in the LV apex wall (Figure A); LV dysfunction with an ejection fraction (EF) of 27%. After 10 months, the patient’s symptoms aggravated with more frequent dyspnea and abdominal distension, reexamination of echocardiography demonstrated the whole heart dilatation and the hypertrabeculation in the right ventricular apex wall except in LV (Figure B).

CMR was performed to further confirm the features. The fast imaging employing steady-state acquisition (FIESTA) cine images revealed non-obstructive HCM (IVS with a thickness of 22 mm at end-diastole), a two-layer myocardium in the biventricular apex, and LV anterior and lateral walls with deeply intertrabecular recesses communicating with blood in the cavity, and the maximum ratio of noncompacted to compacted myocardium (NC/C) was 3.6 at end-diastole (Figures C, D). Late gadolinium enhancement (LGE) showed moderate heterogeneous patchy enhancement in the myocardium of IVS and ring enhancement on the endocardial side (Figure E).

Case 2: A 40-year-old woman came to our hospital with a history of chest distress and polypnea, which aggravated for nearly 1 month. Her mother died of cardiovascular disease and her daughter was diagnosed with HCM. Coronary arteriography was normal. A systolic grade IV blowing murmur was detected in the apex and the second auscultation aortic area. The 24-hour Holter monitoring showed premature atrial contraction, premature ventricular beat, and ST-T segment abnormality. The laboratory results were unremarkable.

Echocardiography demonstrated the left atrioventricular enlargement, hypertrophic IVS (Figure F), and normal LV systolic function with EF of 70%; however, the operator did not find any abnormal echo in all myocardium.

A further examination of CMR showed noncompacted myocardial in the LV lateral and apex walls with maximum NC/C of 3.0 at end-diastole, basal, and middle IVS hypertrophy with maximum thickness of 19 mm (Figures G, H). LGE showed multiple patchy enhancement in IVS myocardium (Figure I). In addition, the signal intensity in the intertrabecular recesses was similar to the cavity.
APPLICATION OF CMR TO PRIMARY CARDIOMYOPATHY

Discussion

In our study, the features of LVNC associated with HCM were demonstrated by CMR. Typically, the non-compacted myocardium often occurred in LV apex and lateral walls rather than RV according to the arrest of the normal compaction process in embryonic development and the previous reports.2,3,8-9) Our CMR findings showed one rare case of noncompacted myocardium involving the biventricular, which when coexisting with HCM is even rarer.10

LVNC and HCM were both genetically determined diseases that presented different phenotypes. Previous studies proved that nearly all of the genes related to LVNC are linked with other phenotypes.1,3-5,11,12) Two families with noncompaction cardiomyopathy showed deficiency of cardiac beta-myosin heavy chain, which was the mutated form of HCM with a restrictive phenotype.11) Analogously, alpha cardiac actin genetic mutation was detected in families with LVNC, HCM, and septal defects.12) In the pedigree maps of 4-generation family, 8 members were diagnosed with hypertrophic LVNC using contrast echocardiography, which showed a link between LVNC and HCM.9

Despite the hypertrophic LVNC, LVNC can also be associated with dilated and restrictive cardiomyopathy, or with congenital heart disease like Ebstein’s anomaly, pulmonary stenosis, and pulmonary atresia, or some other diseases.3-15

Heart failure and arrhythmias influence mortality in LVNC, and hybrid subtypes are often accompanied by diastolic dysfunction and hypercontractile systolic dysfunction.3,10,14) In adults, some hybrid phenotypes seemed to have similar outcomes to LVNC alone, but the progno-
sis in neonates or in hypertrophic dilated LVNC phenotype was worse than LVNC alone. In our cases, case 1 was a young patient, experiencing heart failure with a LV EF of 27%. According to the characteristics of images, the patient had the evidence of myocardial hypertrophy, non-compacted myocardium, and ventricular dilatation, whereas case 2 was hypertrophic LVNC with normal systolic function. Therefore, different phenotypes necessitate different surveillance, and treatment is based on making the correct phenotypic diagnosis.

There had no standard diagnostic criteria for the hybrid subtypes. In our report, we propose a multimodal diagnostic approach. Echocardiography is used to assess the location of hypertrophic myocardium, atrioventricular size, thickness, systolic and diastolic function. Although echocardiography is the most frequently used imaging modality for cardiac diseases, it may be limited by the dependence on an adequate acoustic window and the skill of the operator. These limitations make it insufficient at identifying LVNC, and echocardiography missed some noncompaction segments in our study.

CMR is an effective adjunctive diagnosis method, largely because of superior soft tissue contrast and multiplane scanning. Cine images could clearly show the location of trabeculae and visible blood flow into the deep intertrabecular recesses, highlighted in the description of apical trabecular. In our two cases, the CMR findings match the diagnosis of both LVNC and HCM: two-layered structure with a compacted epicardial and non-compacted endocardial layer with NC/C > 2.3 at end-diastole; hypertrophic interventricular septum with maximum thickness more than 15 mm; and there were no risk factors of the disease. Furthermore, by the specific affected myocardial segments and gadolinium enhancement, we can assess the degree of ventricular burden. Above all, CMR can make up for the defects of echocardiography and help us to get more information for clinical intervention.

Conclusions
In our study, we confirmed two patients with LVNC who associated with HCM that was diagnosed by echocardiography and CMR. In the clinical work, awareness of the existence of hybrid phenotypes should be paid more attention during echocardiographic examination and should encourage the use of CMR.

Disclosures
Conflicts of interest: none.

References