Purpose Non-compaction of the ventricular myocardium (NVM) is a cardiomyopathy caused by arrest of normal embryogenesis of the endocardium and myocardium with some gene disorders, including hypertrophic cardiomyopathy (HCM). However, familial incidence of NVM associated with hypertrophic cardiomyopathy is extremely rare. The aim of our study was to assess familial incidence of NVM associated with hypertrophic cardiomyopathy.

Methods The study consisted of 7 individuals in 3-generation kindred (2 female adults, 3 male adults and 2 children; 2.5–48 years). We investigated detailed clinical presentations and imaging findings of the kindred. Each family member was evaluated by 2-dimensional echocardiography, 4 of which further underwent magnetic resonance imaging. Electrophysiological tests were performed on two inpatients of this kindred.

Results (1) Out of 3 inpatients in the kindred, 2 of them presented with symptoms of systolic dysfunction. One individual underwent chemical ablation treatment because of left ventricular outflow tract (LVOT) obstruction, and obtained an obvious improvement. The rest were clinical asymptomatic. (2) All the family members fulfilled the previously proposed echocardiographic diagnostic criteria for NVM; six of them had echocardiography evidence of HCM. Classic echocardiographic features of NVM and HCM were identified in the proband who was hospitalised due to symptoms of congestion heart failure at his age of 48. His brother, sister, daughter, son and nephew were found to have similar imaging findings as the proband. Six members of NVM associated with HCM displayed hypertrophy in interventricular septum and myocardial non-compaction in mid segments of inferior, posterior, and lateral walls, and apex. Among them, a 40-year-old female showed severe obstruction in LVOT. Only the granddaughter of the proband had non-compaction in the apex, in absence of HCM. (3) Hypertrophy and non-compaction of LV myocardium were clearly displayed in MRI which were performed in 4 individuals. The location of lesions showed a good correlation with that of echocardiography. (4) Abnormal electrophysiological activities were found in two inpatients. One was detected multifocal premature ventricular contraction was detected in one and atrial fibrillation, left anterior fascicular block in another one.

Conclusions This kindred of NVM associated with HCM is reported to share similar familial characteristics among the family members. Echocardiography is a sensitive and effective examination in diagnosis of NVM.