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One genotype, two phenotype: Hypertrophic cardiomyopathy with left ventricular non-compaction

Luísa Gonçalves, Inês Pires, João Santos, Joana Correia, Vanda Neto, Davide Moreira, Miguel Correia, Costa Cabral

Cardiology Department, Centro Hospitalar Tondela-Viseu, Viseu, Portugal

A 65-year-old man with a family history of sudden cardiac death (SCD; mother and two maternal uncles, aged between 52 and 60) is referred for palpitations, non-sustained ventricular tachycardia, and non-obstructive hypertrophic cardiomyopathy (HCM) criteria on echocardiogram (Fig. 1A–F). A comprehensive study is performed. Coronary disease is excluded by coronariography, and cardiac magnetic resonance (CMR) is carried out, disclosing left ventricle (LV) ejection fraction of 55%, mildly dilated LV (101 mL/m^2), asymmetric LV hypertrophy (max 25 mm, septal), and non-ischemic late enhancement associated within hypertrophied segments. Additionally, there was an overlapping feature of LV non-compaction (LVNC) phenotype with a ratio between non-compacted and compacted layer > 2.3 on medium-apical lateral segments (Fig. 1G–J). The pathogenic variant c.2670G>A in heterozygosity in the MYBPC3 gene was present, and cardioverter-defibrillator was implanted for secondary SCD prevention, based on the HCM risk-SCD model > 6%. One year after diagnosis and medication with bisoprolol 5 mg a day, the patient remains eventless.

The phenotypic expression of LVNC and HCM in the same patient is rare. Mutations in the genes encoding the sarcomere proteins have been linked with HCM and LVNC and may justify the overlap of the two cardiomyopathies in this patient. Although the relation between the two cardiomyopathies is still unclear, the presence of LVNC may be associated with ventricular tachycardia in HCM patients, and may provide a new phenotypic marker for adverse prognosis. There are no gold-standard diagnostic criteria for LVNC. It is a myocardial phenotype that ranges from a normal variant to clear LVNC cardiomyopathy. CMR has an increasing role on cardiomyopathies providing associated phenotypes that are missed with echocardiography.

Conflict of interest: None declared

Address for correspondence: Dr. Luísa Gonçalves, Cardiology Department, Centro Hospitalar Tondela-Viseu, Avenida Rei D. Duarte, Viseu, 3504-509, Portugal, tel: +351934120142, e-mail: malvar.luisa@gmail.com

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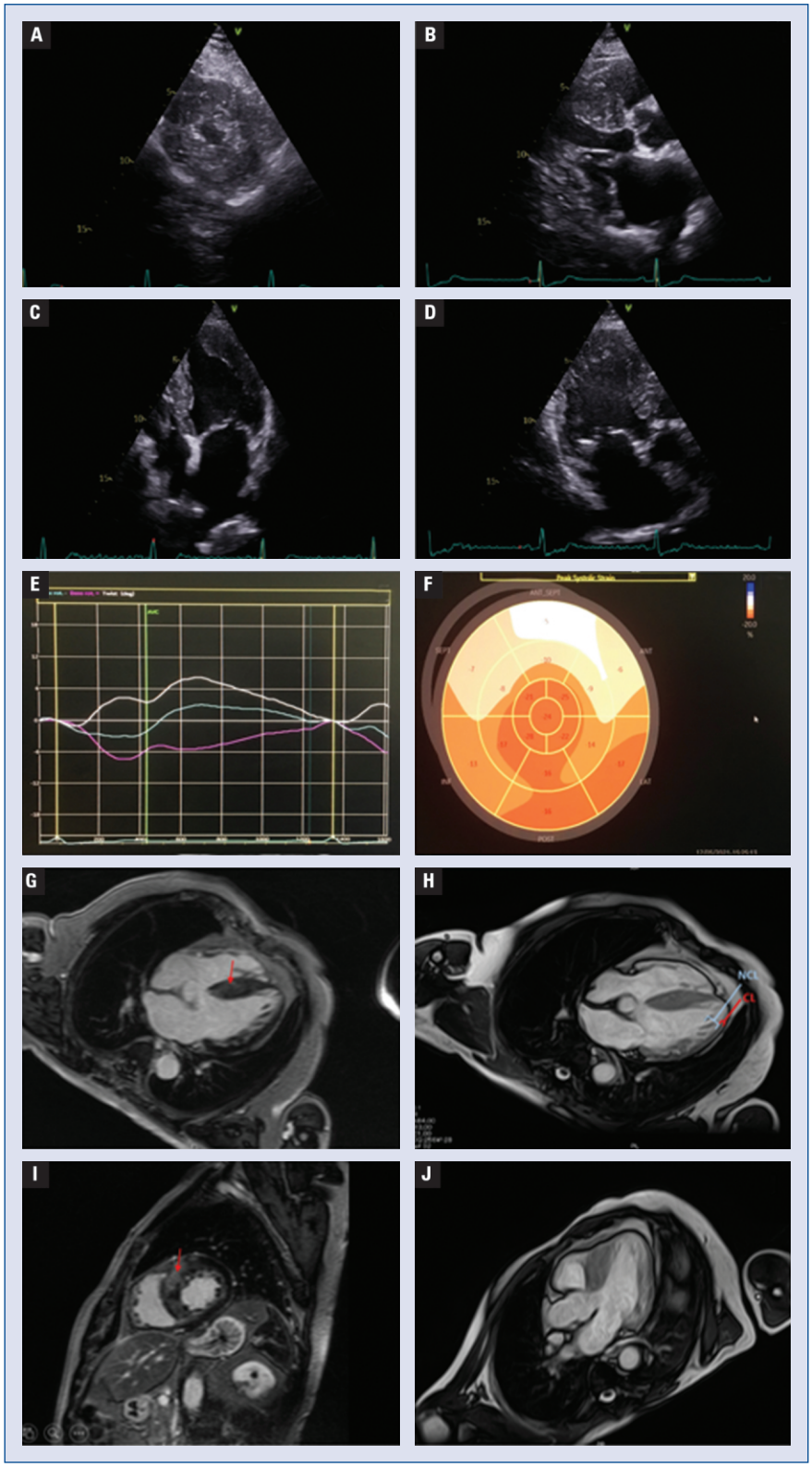

Figure 1. Transthoracic echocardiography (**A**, **B**, **C**, **D**) — two-dimensional: showing severe left ventricular hypertrophy and trabeculations in the left ventricle, visible only in the three chamber view (**D**). Circunferencial (**E**) and longitudinal (**F**) strain analysis: showing reduced global longitudinal strain in the hypertrophied segments, and reduced twist provoked by the base and apex rotating in the same direction. Late gadolinium enhancement (LGE) cardiac magnetic resonance (CMR) sequences (**G**, **I**): marked asymmetric septal hypertrophy associated with diffuse intramural non-ischemic LGE (fibrosis) within the hypertrophied segments (arrows), translating the diagnosis of hypertrophic cardiomyopathy. CINE CMR sequences (**H**, **J**), demonstrating the non-compacted and compacted layers of the myocardium with a trabecular/thin layer ratio of 2.3 at end-diastole in medium-apical lateral segments, a feature of left ventricle non-compaction; NCL — non-compact layer; CL — compact layer.