Hypertrophic cardiomyopathy, non-compaction cardiomyopathy or non-compaction phenotype — another diagnosis, other further treatment

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Kardiol Pol. 2021; 79 (6): 700–701;
DOI: 10.33963/KP.15935

Received:
February 23, 2021

Revision accepted:
April 6, 2021

Published online:
April 13, 2021

Cardiomyopathy phenotype established using imaging methods determinates a diagnosis and further treatment in patients with cardiomyopathies. Hypertrophic cardiomyopathy (HCM) has been clearly defined whereas characterization of left ventricular non-compaction cardiomyopathy (LVNC) is still debated [1]. Apart from LVNC, a non-compaction pattern may accompany other cardiomyopathies, acute myocarditis, and athlete's heart [1, 2].

According to the recent data, HCM and LVNC are diseases with potent overlapping genetic defects as well as phenotypes [3, 4]. We present an atypical patient with a baseline diagnosis of HCM converted after a 3-month follow-up into the diagnosis of LVNC. Re-evaluation was crucial both for the final diagnosis, pharmacotherapy, and sudden cardiac death (SCD) risk re-assessment.

A 51-year-old man with hypertension and prior ischemic stroke was hospitalized due to atypical chest pain. Laboratory tests did not show any abnormalities. Resting ECG showed persistent ST-segment elevations up to 2 mm on the lateral wall leads. Transthoracic echocardiography (TTE) revealed increased thickness of the left ventricle (LV) wall (intraventricular septum [IVS] up to 17 mm, the maximal thickness of LV wall up to 24 mm) without LV gradient and with normal LV ejection fraction (LVEF, 55%) (Figure 1A–B); left atrial area 29.5 cm^2. There was no stenosis on coronary angiography. In 48-hour Holter monitoring several episodes of nsVT (max. 8 QRS) were recorded. His family history of SCD was positive. Family TTE screening allowed to recognize HCM in the patient's son (max. LV wall thickness, 29 mm; SCD risk score, 5.1%) (Figure 1C). Finally, HCM was diagnosed. The calculated SCD risk score was 7.8% and the patient was qualified for cardioverter-defibrillator (ICD) implantation. After three months an ambulatory cardiac magnetic resonance showed LVNC with LVEF 33%. In the 3rd month after the 1st assessment patient was re-hospitalized for ICD implantation. The control TTE showed: the maximal thickness of LV wall up to 15 mm, abnormal LV endocardial trabeculation with the index of spongy/compacted layers 2.0 and LVEF of 35% (Figure 1D–E). The baseline HCM diagnosis was verified by recognizing LVNC. Regardless of the other diagnosis, the patient still had the indications for ICD implantation, which was performed. Pharmacotherapy was optimized according to the heart failure guidelines and because of low LVEF, LV trabeculations and the ischemic stroke incident, anticoagulation was administered.

As it was mentioned, HCM and LVNC may present potent overlapping genetic defects as well as phenotypes [3, 4], therefore simultaneous screening for both HCM and LVNC is necessary. Until now, the overlapping phenotypes have been observed in different family members [3, 4]. Only two cases of coincident non-compacted myocardium and HCM were reported [5]. Based on our case, it might be speculated that the phenotype may change over time. The classical form of HCM has been either converted to LVNC or non-compaction pattern of HCM. Due to a short 3-month follow-up, it is unlikely that the presented asymptomatic patient demonstrated the decompensated HCM. Finally, the re-evaluation was necessary not to overlook details essential for the current

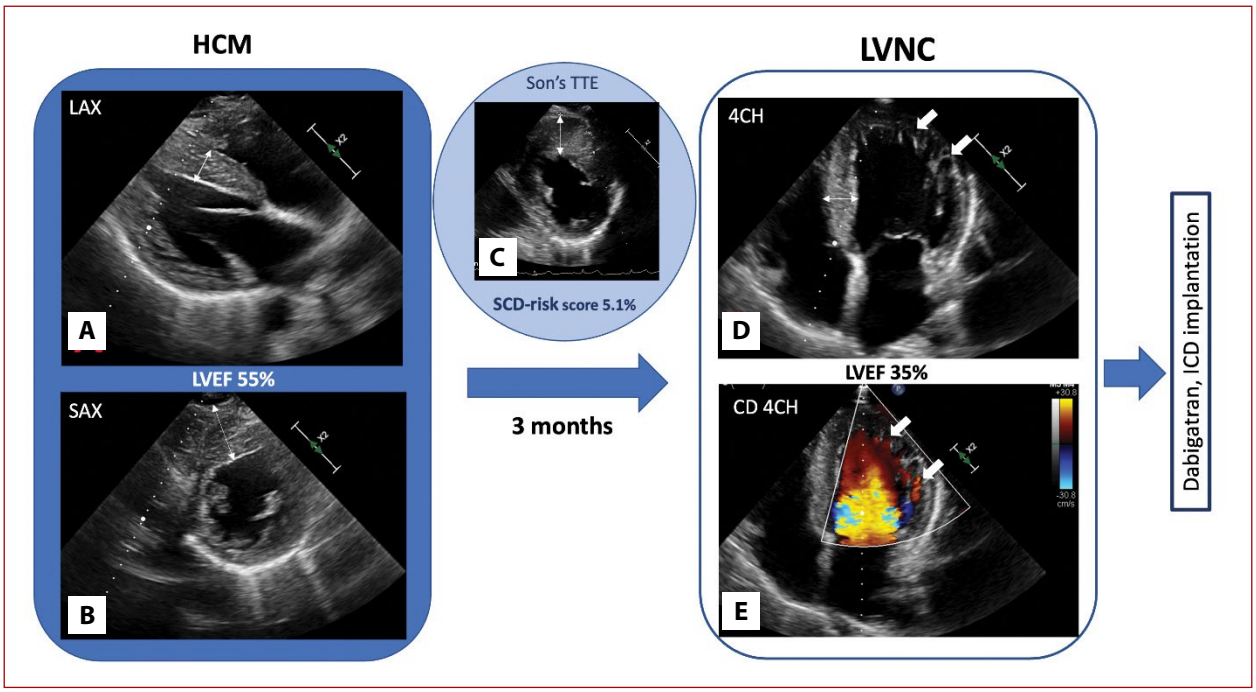

Figure 1. Transthoracic echocardiography (thin arrows — maximal thickness of LV segments). **A.** Long parasternal axis: hypertrophy of the LV wall (IVS thickness, 17 mm). **B.** Short parasternal axis: hypertrophy of the LV wall (maximal LV wall thickness, 24 mm). **C.** Short parasternal axis (echocardiography of the patient's son): hypertrophy of the LV wall (maximal LV wall thickness, 29 mm). **D.** Four-chamber view: the LV trabeculations (IVS thickness, 15 mm; thick arrows — trabeculations). **E.** Four-chamber view, color doppler: blood flow between the LV trabeculations (arrow — blood flow through trabeculations).

Abbreviations: HCM, hypertrophic cardiomyopathy; IVS, intraventricular septum; LV, left ventricle; LVEF, left ventricular ejection fraction; LVNC, left ventricle non-compaction cardiomyopathy; SCD, sudden cardiac death; TTE, transthoracic echocardiogram

approach. That prompts further clinical surveillance supported by genetic tests.

Article information

Conflict of interest: None declared.

How to cite: Bednarek A, Stec M, Mizia-Szubryt M, et al. Hypertrophic cardiomyopathy, non-compaction cardiomyopathy or non-compaction phenotype — another diagnosis, other further treatment. Kardiol Pol. 2021; 79(6): 700–701, doi: 10.33963/KP.15935.

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