Case report: a 55-year-old patient with hypertrophic cardiomyopathy and heterozygous missense variant of TTN and MYH6 genes

Lucija Klobučar[1,2]†, Željka Breškić Ćurić[1,3]†, Ivana Lukić[1,2]†, Kristina Selthofer-Relatić[1,2]*

[1]University Hospital Centre Osijek, Osijek, Croatia
[2]Josip Juraj Strossmayer University of Osijek, Faculty of Medicine Osijek, Osijek, Croatia
[3]General County Hospital Vinkovci, Vinkovci, Croatia

KEYWORDS: hypertrophic cardiomyopathy; genetic testing; left ventricular hypertrophy.
CITATION: Cardiol Croat. 2023;18(5-6):149. | https://doi.org/10.15836/ccar2023.149

***ADDRESS FOR CORRESPONDENCE**: Kristina Selthofer-Relatić, Kliničkі bolnički centar Osijek, Josipa Huttlera 4, HR-31000 Osijek, Croatia. / Phone: +385-98-623-403 / E-mail: selthofer.relatic@gmail.com
ORCID: Lucija Klobučar, https://orcid.org/0000-0003-4333-1048 • Željka Breškić Ćurić, https://orcid.org/0000-0002-6077-4329
Ivana Lukić, https://orcid.org/0000-0001-9832-6700 • Kristina Selthofer-Relatić, https://orcid.org/0000-0002-9890-6489

Introduction: Left ventricular hypertrophy (LVH) diagnosed by echocardiography has broad differential diagnosis. LVH is most frequently caused by abnormal loading conditions (i.e. uncontrolled arterial hypertension and valvular heart disease). When LVH cannot entirely be explained by abnormal loading conditions the diagnosis of hypertrophic cardiomyopathy (HCM) is made. The most common cause of HCM is mutation in cardiac sarcomere protein genes. Some of the less common causes of HCM are metabolic, infiltrative and neuromuscular diseases[1]. Genetic testing is important in recognizing family members at risk of developing HCM. Detected variants of cardiac sarcomere protein genes can be classified (and reclassified) as pathogenic/ likely pathogenic, benign/ likely benign and variant of unknown significance (VUS)[2].

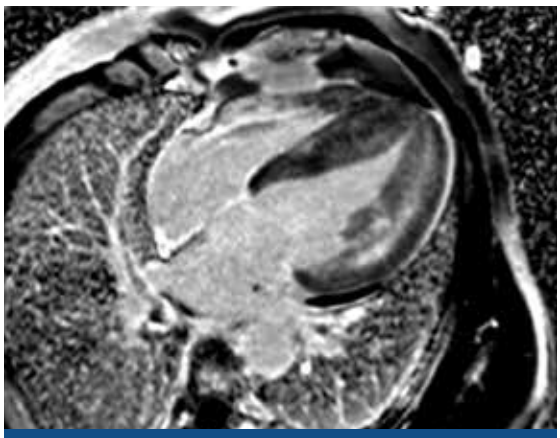

FIGURE 1. Cardiac magnetic resonance image of presented patient.

Case report: We present the case of 55-year-old, hypertensive patient who was admitted to the hospital because of recurrent presyncope and signs of LVH with typical load signs in the ECG that could not be related to hypertensive heart disease. Non-obstructive HCM with preserved ejection fraction was diagnosed by echocardiography, elevated hsTn and NTproBNP were detected and atherosclerotic coronary artery disease was excluded (**Figure 1**). By ECG monitoring ventricular nonsustained tachycardia was detected and an implantable cardioverter defibrillator was implanted in primary prevention of sudden cardiac death. Genetic testing identified a heterozygous missense variant rs201381085 in TTN gene and heterozygous missense variant in MYH6 gene. Currently, both of these variants are classified as VUS but with known HCM cases in literature related to these mutations. Genetic screening also revealed the same heterozygous variant in TTN gene in his 26-year-old son who is currently phenotype-negative (**Figure 2**).

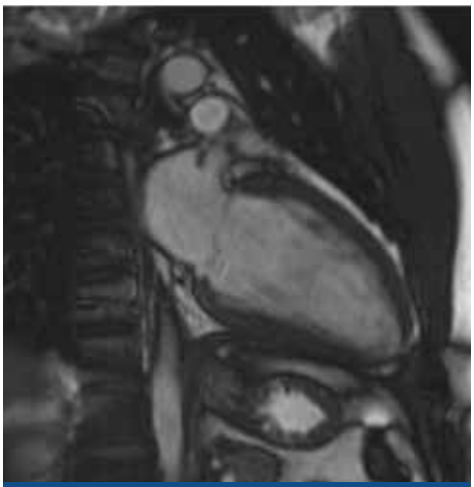

FIGURE 2. Cardiac magnetic resonance image of patient's phenotype-negative son.

Conclusion: In daily clinical practice, genetic testing of HCM patients is still unfortunately underutilized due to work overload, administrative and organizational difficulties. Knowing the importance and potential benefits of genetic testing for the patient, as for their families, the goal is to simplify the entire process.

†Equally contributing first authors

RECEIVED:
March 14 2023
ACCEPTED:
March 29, 2023

LITERATURE

1. Authors/Task Force members; Elliott PM, Anastasakis A, Borger MA, Borggrefe M, Cecchi F, Charron P, et al. 2014 ESC Guidelines on diagnosis and management of hypertrophic cardiomyopathy: the Task Force for the Diagnosis and Management of Hypertrophic Cardiomyopathy of the European Society of Cardiology (ESC). Eur Heart J. 2014 Oct 14;35(39):2733-79. https://doi.org/10.1093/eurheartj/ehu284
2. Ommen SR, Mital S, Burke MA, Day SM, Deswal A, Elliott P, et al. 2020 AHA/ACC Guideline for the Diagnosis and Treatment of Patients With Hypertrophic Cardiomyopathy: A Report of the American College of Cardiology/American Heart Association Joint Committee on Clinical Practice Guidelines. Circulation. 2020 Dec 22;142(25):e558-e631. https://doi.org/10.1161/CIR.0000000000000937