Hypertrophic and noncompacted cardiomyopathy of left ventricle: different manifestations of the same disease

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Hypertrophic cardiomyopathy (HCM) and left ventricular noncompaction (LVNC) are both genetically determined and familial diseases. Hypertrophic cardiomyopathy (HCM) is defined as hypertrophy of the myocardium more than 1.5cm, without another identifiable cause, such as long-standing hypertension, amyloidosis, aortic stenosis, glycogen storage disease. Many of the mutations associated with HCM involve the cardiac sarcomeric proteins and include actin, myosin, or troponin component of the sarcomere and it is most frequently transmitted as an autosomal dominant trait. Left ventricular noncompaction is a rare congenital cardiomyopathy which is characterized by the presence of a thin, compacted epicardial layer and a non-compacted thicker endocardial layer of myocardium, with prominent trabeculation and deep recesses communicating with the cavity of the left ventricle. The cause of the disorder has been identified as mutations in genes associated with the mitochondrial function, like G4.5 which encodes the protein tafazzin, genes related with the cytoskeleton, like those of alpha-dystrobrevin or dystrophin, genes that code proteins of the Z line of the sarcomere, like LDB3, which codes the protein Cypher/ZASP, genes of the internal nuclear membrane proteins (LMNA, which encodes lamin A/C) and even genes that code sarcomeric proteins like cardiac alpha-actin and the beta-myosin heavy chain and cardiac troponin T. The clinical picture of both diseases, HCM and LVNC, varies from mild forms until severe forms with heart failure and complex ventricular arrhythmias. LVNC and HCM may appear as overlapping entities. Cases of patients sharing both the LVNC and HCM phenotypes have been already published, and it is speculated that mutations in sarcomere protein genes known to cause hypertrophic cardiomyopathy and dilated cardiomyopathy may be associated with left ventricular noncompaction.[1-5] In our case report, we are presenting patient with clear overlapping pheenotyp for LVNC and HCM, using the imaging method cardiac MRI.

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LITERATURE

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