

Pay attention to the skeletal muscles in left ventricular hypertrabeculation / noncompaction

To the Editor With interest we read the article by Hellmann et al.¹ about a 44-year-old man with dilated cardiomyopathy and left ventricular hypertrabeculation / noncompaction (LVHT).¹ We have the following comments and concerns.

The concept of LVHT to be a congenital abnormality has recently been challenged by 3 findings. First, the analysis of previous echocardiograms in LVHT patients with neuromuscular disorders (NMDs) has shown that, in some of them, LVHT was not present on those echocardiograms but occurred only during one of the follow-up tests.² This phenomenon was called “acquired LVHT” and was initially considered as not very plausible. Acquired LVHT has been observed particularly in patients with NMDs and is currently accepted as a rare phenomenon. Second, a study on athletes performing extensive muscle training has shown that LVHT either increased between the start of training and the last follow-up visit or developed during the training period.³ Third, 7% of pregnant women develop LVHT during pregnancy, which resolves after the delivery.⁴

Since LVHT frequently occurs in families,⁵ it would be interesting to know whether it was additionally detected in any of the first-degree relatives of the patient? Did any of the relatives undergo echocardiography to confirm or exclude a metabolic defect affecting the skeletal muscle and heart? Was the family history positive for stroke, syncope, heart failure, or sudden cardiac death?

Since the patient had no abnormalities on coronary angiography, cardiac findings cannot only be interpreted as LVHT but must be also recognized as dilative cardiomyopathy with heart failure.¹ Since heart failure of any cause in association with LVHT requires oral anticoagulation with vitamin K antagonists, it would be interesting to know why the presented patient did not receive phenprocoumon or warfarin as primary prophylaxis against cardiac embolic events? Was the history positive for cardiac embolism or even embolic stroke?

According to Hellmann et al.,¹ the patient died despite the previous implantation of an

implantable-cardioverter defibrillator (ICD).¹ What was the cause of sudden cardiac death in this particular patient? Did the ICD ever discharged after implantation? Did the patient adhere sufficiently to the medication prescribed? Were the histological findings interpreted as myocardial infarction? Did the histological workup of the myocardium indicate histiocytoid cardiomyopathy, which is particularly frequent in patients with metabolic myopathy?

Since LVHT is frequently associated with NMDs, the authors should mention if there was any clinical evidence that the patient suffered from a subclinical muscle disease. Did the patient report muscle cramps, easy fatigability, muscle stiffness, or muscle weakness? Did the patient complain about double vision or hearing impairment? Was the history positive for creatine kinase elevation, episodes of rhabdomyolysis, or malignant hyperthermia? Was lactate ever measured and was it elevated at rest or during mild exercise? Were any of the family members suffering from clinical or subclinical NMD?

Overall, this interesting presentation could benefit from providing more clinical details and from more thorough investigations of a possible myopathic background. Additionally, the discrepancy between left ventricular hypertrophy on autopsy and normal myocardial thickness on echocardiography needs to be resolved.

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Authors' reply With interest we read the letter by Drs. Finsterer and Zarrouk-Mahjoub, discussing the necessity of differentiation between "pure" left ventricular hypertrabeculation / non-compaction (LVHT), "acquired" LVHT, and neuromuscular disorder (NMD)-related LVHT. Certainly, their valuable comments add to the discussion; however, we cannot accept all the arguments without adding some remarks.

In their article published in 2010, Finsterer et al.¹ stated that "LVHT per se does not require a specific treatment. Only in case of complications, such as ventricular arrhythmias, cardioembolism, or systolic dysfunction, adequate therapy is indicated". In fact, the patient, previously undiagnosed and untreated, presented to the hospital with de novo heart failure (HF) just about 3 weeks before his death, and he received standard HF treatment including in-hospital thromboembolism prophylaxis. He sustained in-hospital ventricular fibrillation, followed by an implantable cardioverter defibrillator (ICD) implantation.

In another article, Finsterer et al.² presented 22 patients, of whom 4 were diagnosed with "acquired LVHT". However, only 1 of the previous 4 echocardiograms was entirely normal. It is conceivable that myopathy- or dystrophinopathy-related LVHT may evolve with time just as the skeletal muscle involvement does. Hence, in some cases, LVHT may not meet the whole range of the accepted criteria, and possibly remain unnoticed on echocardiography. We also agree that some patients may develop reversible LVHT morphology in specific physiological circumstances (such as pregnancy or high exercise workload). However, 1 of 22 patients is not a frequent occurrence but rather an exception. It is certainly vital to pay attention to earlier echocardiographic images of the patient with LVHT. We also agree that ultrastructural investigation would have been helpful in our case.³

The patient's family history of NMDs was negative, and his 2 children, aged 9 and 11 years, as well as his sister were all healthy and had normal echocardiographic results. The family history was also negative for stroke, syncope, HF, and sudden cardiac death.

Dilative cardiomyopathy usually presents as an enlarged left ventricle with low ejection fraction (<35%), but without prominent trabeculations, so its diagnosis in this case seemed unwarranted.

The cause of death of the 44-year-old patient was acutely decompensated HF, despite treatment with an angiotensin-converting enzyme inhibitor, β -blocker, and diuretic (although the patient's overall compliance was judged as poor). At the end stage of the disease, optimal treatment of acute biventricular HF, including catecholamines and furosemide infusion, proved unsuccessful. Several ICD discharges were noted on the day of acute decompensation.

The difference between the assessment of left ventricular thickness on echocardiography and on autopsy might have resulted from the fact that left ventricular hypertrophy on autopsy was assessed by weighing the left ventricle. A thick layer of trabeculation clearly affected the overall myocardial mass. The early ischemic injury found on histopathology was not compatible with an acute coronary syndrome.

Even though the prevalence of noncompaction in the population is low (0.014% to 1.3%),⁴ we agree that potential links between LVHT and NMDs should be kept in mind when dealing with LVHT patients. In conclusion, we hope that we provided adequate answers to all the questions, and we would be happy to continue the discussion in the future.

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