Acute heart failure from noncompaction requiring emergency heart transplantation

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LETTER TO THE EDITOR

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Insuficiência cardíaca aguda associada a não compactação requerendo transplantação cardíaca emergente

It was with interest that we read the article by Meneguz-Moreno et al. about a 14-year-old Caucasian girl with left ventricular hypertrabeculation/noncompaction (LVHT/NC) who developed sudden onset heart failure and had to undergo emergency heart transplantation (HTX) because of intractable heart failure.[1] We have the following comments and concerns.

Since the father of the index patient had cardiomyopathy, it is quite likely that he had undergone echocardiography.[1] Were these echocardiograms reviewed and was LVHT/NC detected in the father as well? Was the father's history positive for ischemic stroke, atrial fibrillation, heart failure, ventricular arrhythmias, or even epilepsy, as in his daughter? What type of cardiomyopathy was diagnosed and what was the cause? Did any other family member also develop cardiomyopathy, heart failure, arrhythmias, or cardioembolism?

The index patient is reported to have developed seizures.[1] What type of seizures did she develop? What was the frequency of these seizures? What were the results of the EEGs? What were the findings on cerebral MRI? What was the cause of seizures? Were they attributed to cerebral hypoxia because of cerebral hypoperfusion due to acute heart failure? Did the girl ever experience a juvenile stroke from cardioembolism? Was atrial fibrillation ever recorded on the ECG? Were intracardiac thrombi detected? What measures were taken to control seizures? What type of antiepileptic drugs (AEDs) were prescribed? Were thrombocytopenia and hepatopathy attributable to side-effects of the AED medication?

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LVHT/NC is frequently associated with genetic disorders but a causal relation has never been proven. Furthermore, LVHT/NC is associated not only with disorders due to mutations in genes such as *G4.5*, *PRDM16*, *TNNT2*, *LDB3*, *MYBPC3*, *MYH7*, *ACTC1*, *TPM1*, *MIB1*, or *DTNA*, but also with mutations in a number of other genes.[2] There are also reports of an association of LVHT/NC with chromosomal defects.[3] Concerning the *G4.5* mutation, it should be noted that this gene is the same as the taffazin gene (*TAZ*). Mutations in *TAZ* (*G4.5*) cause Barth syndrome, an X-linked disorder, in which up to half of patients present with LVHT/NC.[4]

The authors regard LVHT/NC as a congenital abnormality.[1] Though this is presumably true in the majority of cases, there are also reports showing that LVHT/NC can develop after birth (acquired LVHT/NC).[5] This is particularly the case for patients with neuromuscular disorders (NMDs),[5] pregnant women,[6] and professional athletes.[7] Acquired LVHT/NC is regarded as a mechanism for compensating systolic dysfunction or for improving blood oxygenation.

LVHT/NC is associated with NMDs in up to 80% of cases.[8] Were there clinical or laboratory indications of an NMD in the index case or any of her relatives? Did she or any of her relatives complain of easy fatigability, exercise intolerance, muscle cramps, muscle wasting, muscle weakness or myotonia, or was there hyperCKemia? Did any have a history of adverse reactions to general anesthesia?

LVHT/NC is frequently associated with myocardial fibrosis, as demonstrated by the presence of late gadolinium enhancement on cardiac MRI.[9] Did investigation of the explanted heart show subendocardial, mid-myocardial, or subepicardial fibrosis? Was there any indication of subendocardial fibroelastosis?

Was the patient followed up after transplantation? Did she develop LVHT/NC in the transplanted heart also? This is an unresolved issue if there is LVHT/NC in the transplanted heart. So far, more than 50 patients with LVHT/NC have undergone HTX, but in none of them have long-term follow-up investigations been carried out to address this question.

Overall, this interesting case requires a more thorough individual and family history, work-up for NMD or a chromosomal defect in the index patient and her relatives, and screening for LVHT/NC in other family members.

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Conflicts of interest

The authors have no conflicts of interest to declare.

References

1. Meneguz-Moreno RA, Rodrigues da Costa Teixeira F, Rossi Neto JM, et al. Isolated left ventricular noncompaction causing refractory heart failure. Rev Port Cardiol. 2016;35:185.e1–4.
2. Finsterer J, Zarrouk-Mahjoub S. Considerations about the genetics of left ventricular hypertrabeculation/non-compaction. Cardiol Young. 2015;25:1435–7.
3. Finsterer J. Cardiogenetics, neurogenetics, and pathogenetics of left ventricular hypertrabeculation/noncompaction. Pediatr Cardiol. 2009;30:659–81.
4. Ronvelia D, Greenwood J, Platt J, et al. Intrafamilial variability for novel TAZ gene mutation: Barth syndrome with dilated cardiomyopathy and heart failure in an infant and left ventricular noncompaction in his great-uncle. Mol Genet Metab. 2012;107:428–32.
5. Finsterer J, Stöllberger C, Schubert B. Acquired left ventricular noncompaction as a cardiac manifestation of neuromuscular disorders. Scand Cardiovasc J. 2008;42:25–30.
6. Gati S, Papadakis M, Papamichael ND, et al. Reversible de novo left ventricular trabeculations in pregnant women: implications for the diagnosis of left ventricular noncompaction in low-risk populations. Circulation. 2014;130:475–83.
7. Martinoli R, Papetti F, Dofcaci A, et al. Isolated left ventricular non compaction as possible cause of athletic training suspension: a preliminary study on screened athletes. J Sports Med Phys Fitness. 2013;53:240–7.
8. Stöllberger C, Finsterer J, Blazek G. Left ventricular hypertrabeculation/noncompaction and association with additional cardiac abnormalities and neuromuscular disorders. Am J Cardiol. 2002;90:899–902.
9. Alter P, Rupp H. Myocardial fibrosis in left ventricular noncompaction: is late gadolinium enhancement indeed indicative of fibrosis? Eur J Heart Fail. 2011;13:577–8.

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