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Myopathic background of non-compaction in children

With interest, we read the article by Lilje et al. on the clinical findings and outcome of 66 paediatric patients with left-ventricular hypertrabeculation (LVHT), also known as left-ventricular non-compaction. The study raises concerns.

Though repeatedly mentioned, the non-compaction hypothesis is insufficient to explain the pathogenesis of LVHT, since it does not explain acquired LVHT developing during life-time. To explain acquired LVHT, pathogenetic concepts other than the non-compaction hypothesis have been proposed. Acquired LVHT is also the reason why 'non-compaction' should be replaced by a more descriptive term.

Concerning the mortality of LVHT, only few data are available. Among 86 adult LVHT patients, we calculated a mortality of 5.3% per year. Predictors for mortality in this study were advanced age, the presence of a neuromuscular disorder, exertional dyspnoea, oedema, heart failure, left anterior hemiblock, and reduced systolic function. The genetic background of LVHT is far more heterogeneous than reported. LVHT was not only associated with mutations in G4.5, cypher/ZASP, DTNA (dystrobrevin), and lamin A/C genes, but also with mutations in GAA, AMPK, AMPD1, mitochondrial, frataxin, CSX, and PMP22 genes. LVHT has been also found in Turner syndrome, Ohtahara syndrome, Roffman syndrome, Noonan syndrome, nail-patella syndrome, Melnick-needles syndrome, MIDAS syndrome, DiGeorge syndrome, congenital adrenal hyperplasia, distal 4q-trisomy/1q-monosomy, distal 5q-deletion, trisomy-11, and trisomy-13. Because of the heterogeneous genetic background and the familial occurrence of LVHT, it should be mentioned how many patients were related to each other and whether a family screening had been carried out.

There is no consensus on the definition of LVHT. The authors contribute to this confusion by introducing a further echocardiographic definition, which additionally measures the X/Y-ratio at the level of papillary muscles and the mitral valve, locations where LVHT is usually difficult to distinguish from normal cardiac structures. Was the ratio measured at end systole or end dia-stole? How often was LVHT located at the septum? How were false tendons and aberrant bands discriminated from LVHT?

Also, the distinction between isolated and non-isolated LVHT does not contribute to the understanding of LVHT. Is LVHT with ECG abnormalities or relative tricuspid or mitral insufficiency isolated or non-isolated? Which cardiac abnormalities were found among the non-isolated cases?

Basic information is not provided. Were any abnormalities found on blood tests? Which ECG abnormalities were found? Which were the indications for echocardiography? How many patients had a reduced systolic function? How many had dilated or wall-thickened left ventricles? Were coronary abnormalities found? Which cardiac therapy was given? How many patients were lost to follow-up? Did LVHT localization or morphology change during follow-up?

It is mentioned that the echocardiograms were evaluated by two independent investigators. How often did they disagree on the diagnosis? What happened if they disagreed?

In up to 80% of patients, LVHT is associated with neuromuscular disorders such as dystrophinopathies, dystrobreinopathies, myotonic dystrophy, zaspatophisies, myoendylate-deaminase deficiency, Charcot-Marie-Tooth disease, mitochondrial disorder, Barth syndrome, Friedrich ataxia, or Pompe's disease. Were patients seen by a neurologist and how many had neuromuscular disorders?

Overall, a common definition of LVHT for paediatric and adult LVHT is warranted. Furthermore, more clinical information about paediatric LVHT patients is required to compare them with adult cases.

References


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Myopathic background of non-compaction in children: reply

We appreciate the interest of Finsterer et al. in our article. Unfortunately, not all of their numerous concerns can be addressed here, some of which have been raised and replied to previously.

(i) Chin et al. were among the first to describe NCVM. It was considered to result from an arrest in the process of embryonic myocardial compaction. There is much evidence, including excellent work on animal models, to support this pathogenic concept and the term NCVM. Both have been widely accepted. The evidence for 'acquired' NCVM is scarce. A few reports, mostly by Finsterer et al., have been published on single patients with 'acquired' NCVM. Although there is a variety of reasons for secondary left ventricular hypertrobrabeculation