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Letter to the Editor

Is Left Ventricular Noncompaction A Cardiac Manifestation of Trisomy 13?

Josef Finsterer, MD, PhD[1] and Claudia Stöllberger, MD[2]

[1]Krankenanstalt Rudolfstiftung, Messerli Institute, Vienna, Austria
[2]2nd Medical Departments, Krankenanstalt Rudolfstiftung, Vienna, Austria

*Corresponding Author
Josef Finsterer, MD, PhD

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With interest we read the article by McMahon *et al.*, about an 11-year-old girl in whom left ventricular hypertrabeculation/noncompaction (LVHT) was found on transthoracic echocardiography (McMahon, C. J. *et al.*, 2005). The presented data and their interpretation raise the following concerns:

LVHT is not only a congenital cardiac abnormality but may also develop during lifetime in single cases (Finsterer, J., & Stöllberger, C. 2001; Finsterer, J. *et al.*, 2004). Thus, the interpretation of LVHT as the result of an arrest in the compaction process of the embryonic heart requires extension by other pathomechanistic explanations, like compensatory attempt of an impaired myocardium or destruction of an impaired myocardium.

Did the patient show any abnormalities on ECG or left ventricular wall thickening on echocardiography? Did the restrictive filling pattern of the transmitral flow disappear after therapy? What was the cause of heart failure? Did the authors interpret heart failure due to restrictive cardiomyopathy?

Whether LVHT is associated with an increased rate of embolism is controversially discussed. In a series of 62 LVHT patients thrombo-embolic events were recorded in only 10% of them, but in 15% of age-, sex- and left ventricular function-matched controls (Stöllberger , C., & Finsterer, J. 2004).

LVHT has been assessed by at least three different definitions (Stöllberger , C., & Finsterer, J. 2004; Chin, T.K. *et al.*, 1990; Oechslin, E.N. *et al.*, 2000). Did the echocardiographic findings match with all of them. In this respect it is noteworthy that the authors mix up Oechslin's definition of LVHT with Stölberger's definition. The presence of more that three trabeculations is part of Stöllberger's definition.

Since LVHT occurs frequently in addition with skeletal muscle disorders like Duchenne and Becker muscular dystrophy, myotonic dystrophy 1, dystrobrevinopathy, Pompe's disease, myoadenylate-deaminase deficiency, metabolic myopathy, cypher gene mutations, Friedreich ataxia, and Barth syndrome, it would be interesting to know if the described patient was also seen by a neurologist specialised in neuromuscular disorders.

Reference 14 is misinterpreted. In this study we systematically looked for Fabry's disease in 26 adult patients with LVHT but found α-galactosidase A decreased in none of them (Stollberger, C., & Finsterer, J. 2004 & Stöllberger, C. *et al.*, 2003).

Did the authors consider LVHT as a manifestation of another co-existing disease in their patient? Did they investigate the relatives of the patient for LVHT and neuromuscular disease? Though quite likely, the data supplied do not unequivocally prove that LVHT was a manifestation of trisomy 13.

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Overall, patients with LVHT require a multi-disciplinary diagnostic approach. It is also essential to investigate relatives of LVHT patients since familial cases have been reported. Since there is no consensus about the diagnostic criteria for LVHT effort should be made to uniformly characterise LVHT in pediatric and adult cases.

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