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Letter to the Editor

Left Ventricular Hypertrabeculation / Noncompaction as a Manifestation of Myopathy

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With interest we read the article by Chung *et al.,*. on a series of 9 patients from 2 families with left ventricular hypertrabeculation/noncompaction (LVHT) (Chung, T. *et al.,* 2004). We have the following comments and concerns.

Obviously, Oechslin's definition of LVHT was applied. Were other definitions of LVHT also considered? (Chin, T. K. *et al.,* 1990; Stöllberger, C., & Finsterer, J. 2004). Did the morphological abnormalities in the 9 affected patients also fulfill the two other LVHT definitions?

The authors talk about "phenotypic variation" which implies that they believe that LVHT is a genetic congenital disorder. However, there are several reports, which clearly demonstrate that LVHT can be also acquired (Finsterer, J., & Stöllberger, C. 2001; Finsterer, J. *et al.,* 2004). In case of acquired LVHT it is assumed that LVHT represents either a compensatory mechanism of an impaired myocardium or results from continuous destruction of the affected myocardium. The discussion about improper visualization techniques as the cause of initially overlooking LVHT is justified, since poor image quality easily results in misdiagnosing LVHT. However, in those cases with acquired LVHT mentioned above the initial examinations were properly carried out and the initial data thoroughly reviewed.

Whether LVHT is an indication for oral anti-coagulation or not is controversially discussed. In a series of 62 patients we found thrombo-embolic events during lifetime in only 10% of them and in 15% of age-, sex-, and left ventricular function-matched controls. What was the indication for oral anticoagulation in patient 4 and 6? Did they develop dilated cardiomyopathy or atrial fibrillation?

We agree that transthoracic echocardiography is the method of choice to detect and assess LVHT. In a series of 19 patients cardiac MRI confirmed LVHT in echocardiographically diagnosed LVHT patients in only 47% of echocardiographically diagnosed LVHT cases [personal communication].

Cardiac computed tomography represents a high radiation burden. Additionally 150ml of contrast medium is a high amount and cost expensive. Why not applying cardiac MRI, which most likely carries a much lower risk of side effects?

ECG and 24h-ECG data are lacking. Which was the cause of cardiac arrest in patient 3 and 7?

Patient 8 underwent bypass surgery. Was a myocardial biopsy taken during the procedure to look for any myocardial abnormality? Was LVHT in patient 7 confirmed at autopsy?

We don't completely agree that LVHT patients "commonly have diffuse left ventricular dysfunction". In a series of 77 LVHT patients fractional shortening <25% was found in only 56% of them (Stöllberger, C., & Finsterer, J. 2004).

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LVHT is associated with neuromuscular disorders in up to 82% (Stöllberger, C. et al., 2002) and 87% [personal communication] of the cases respectively. So far LVHT has been associated with Duchenne and Becker muscular dystrophy, myotonic dystrophy type 1, dystrobrevinopathy, Pompe's disease, myoadenylate-deaminasedeficiency,mitochondriopathy, cypher gene mutations, Friedreich ataxia, Barth-syndrome and various other rare genetic disorders (Stöllberger, C., & Finsterer, J. 2004; Alper, G., & Narayanan, V. 2003; Finsterer, J. et al., 2004; Ichida, F. et al., 2001; Pipo, J. R. et al., 2003; Vatta, M. et al., 2003). Did any of the nine affected patients undergo neurologic investigations?

Facial dysmorphism has been reported in 14 patients with LVHT before (Chin, T. K. *et al.*, 1990; Digilio, M. C. *et al.*, 1999; Ichida, F. *et al.*, 1999; Allenby, P. A. *et al.*, 1988). What was the reason for facial dysmorphism in family B? Did this family present with any other dysontogenetic features? Unexplained also remains the developmental delay of patient 6. What was her neurological diagnosis? Are there any cerebral imaging data available?

Overall, it needs to be stressed that despite the primary impression of an exclusively cardiac abnormality, LVHT requires more extensive investigations for extra-cardiac, including neurological, manifestations in these patients. It also should be emphasized that all family members require search for LVHT if diagnosed in one of them. Any additional cardiac abnormality requires appropriate cardiac therapy. Oral anticoagulation is indicated only if there is decreased systolic function or atrial fibrillation. Regular follow-ups are essential in LVHT patients.

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