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Letter to the Editor

Genetic background of left ventricular hypertrabeculation/ noncompaction

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Keywords: left-ventricular-hypertrabeculation/noncompaction (LVHT), dystrophin, laminA/C, AMPD1, mtDNA, and PMP22 genes.

In a recent article Xing *et al.*, (2006) investigated 79 patients with left-ventricular-hypertrabeculation/noncompaction (LVHT) for mutations in the G4.5 (TAZ), FK506-binding-protein-1A (FKPB1A), α1-syntrophin (SNTA1), cypher (LDB3, ZASP), and α-dystrobrevin (DTNA) genes respectively (Xing, Y. *et al.*, 2006). We have the following comments and concerns.

LVHT is not only congenital but also acquired (Finsterer, J. *et al.*, 2004).

No causal relationship between arrhythmias, heart failure, cardiac transplantation, or death and LVHT has yet been established.

LVHT is not only associated with mutations in the DTNA, TAZ, and LDB3, but also the dystrophin, laminA/C, AMPD1, mtDNA, and PMP22 genes (Finsterer, J. *et al.*, 2005).

Affection of >1 segment is a new diagnostic criterion (Oechslin, E. N. *et al.*, 2000). Which segments are meant? Reduced systolic function is also a new diagnostic criterion and contradicts isolated LVHT (Oechslin, E. N. *et al.*, 2000). A mandatory diagnostic criterion is a ratio non-compacted/compacted layer >2 in systole (Oechslin, E. N. *et al.*, 2000). According to the definition applied, all 79 patients should have had non-isolated LVHT, which contradicts the provided data, since 66 patients must have had isolated LVHT (relation isolated/non-isolated LVHT: 6:1).

Echocardiography is unable to demonstrate "myocardial insufficiency".

LVHT is more prevalent in adults than infants (Stöllberger, C., & Finsterer, J. 2004).

How was a "potentially informative family member" defined? How many family members of the 20 patients with familial LVHT were affected?

A typical feature of Barth syndrome and dystrobrevinopathy is myopathy (Bleyl, S. B. *et al.*, 1997). Why were these patients not neurologically investigated?

Why was blood drawn from 103 LVHT-patients but only 79 included? Which were the criteria to exclude 24 patients?

LVHT was also described together with the ZASP-mutations C587T, C638T, and G349A. Were these mutations also screened in the present series?

Mitochondrial inheritance and mitochondrial disorder was found in how many patients? Were patients with X-linked inheritance but absent G4.5 mutation also screened for dystrophin mutations? Recently, a Duchenne-patient with LVHT was described (Finsterer, J. *et al.*, 2006).

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LVHT appears to be rather the common endpoint of different pathogenetic processes than a distinct cardiomyopathy. Presumably, LVHT represents the myocardial effort to compensate for insufficient contractility.

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