

# Noncompaction with dysmorphism, mental retardation, general wasting, and hypogonadism requires neurologic and sophisticated cytogenetic investigations

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## Noncompaction with dysmorphism, mental retardation, general wasting, and hypogonadism requires neurologic and sophisticated cytogenetic investigations

To the Editor,

We read with interest the article published entitled "Case of fatal heart failure with biventricular noncompaction, genital skeletal abnormalities and mental retardation." by Ataş et al. (1) regarding a 48-year-old female from consanguineous parents with dilated cardiomyopathy (dCMP), left ventricular hypertrabeculation/noncompaction (LVHT), primary amenorrhea, bilateral amazia, ovarian dysgenesis, uterine aplasia, hypergonadotropic hypogonadism, macrocephaly, facial acromegaly, arachnodactyly, pectus carinatum, and mental retardation who died from heart failure 4 months after being diagnosed with LVHT. We have the following comments and concerns.

We do not agree with the statement that LVHT is a genetic disorder. Although LVHT is associated with various monogenic disorders, in particular neuromuscular disorders (NMDs) and cardiomyopathies, and chromosomal defects (2), a causal relation between these genetic defects and LVHT has not yet been proven. The strongest argument against a causal relation is that only a small number of patients with

NMDs, cardiomyopathies, and chromosomal defects present with LVHT (2). An argument in favor of a causal relation, however, is that LVHT also occurs familial (3).

The patient underwent cytogenetic investigation; however, it is not mentioned which technique was applied (1). Did the authors investigate complex chromosomal re-arrangements and micro-aberrations by means of fluorescence in-situ-hybridization (FISH) or microarray assays? In particular, did they apply multi-color FISH, telomere/subtelomere FISH, reverse painting, fiber FISH, quantitative FISH, or cobra-FISH?

According to Figure 1, the patient presented with generalized muscle wasting (1). Was this due to being bedridden prior to admission or was this due to involvement of the peripheral nerves or the skeletal muscles? Did the patient ever undergo a clinical neurologic investigation, nerve conduction studies, or needle electromyography? This is of particular importance because LVHT is associated with NMDs in more than half of the cases.

Concerning mental retardation and macrocephaly, it would be interesting to know cerebral imaging results. Was there cerebral atrophy, calcification, demyelination, or hydrocephalus? Did she ever develop seizures? Was an electroencephalogram ever recorded?

Because LVHT can be complicated by stroke embolism, it is important to understand whether the individual or family history was positive for stroke/embolism. Did cerebral imaging reveal previous embolic stroke? Furthermore, patients with LVHT and dCMP require oral anticoagulation with vitamin-K antagonists for primary prophylaxis of stroke/embolism (4). Did the patient receive phenprocoumon or warfarin in addition to heart failure therapy on dismissal?

Furthermore, because LVHT is complicated by arrhythmias, it would be worthwhile to know the results of long-term electrocardiography recordings. Did the two sisters and brother who deceased in childhood die suddenly? Was the family history positive for falls, syncope, fainting, or sudden cardiac death? Was an autopsy conducted in the three deceased children?

Because LVHT may be acquired in some cases (5), it would be interesting to know whether the patient had undergone previous echocardiographies and if these were revised for LVHT?

Overall, this interesting case merits further evaluation with regard to genetic background and possible neuromuscular or cerebral comorbidities. Only if LVHT patients are comprehensively investigated, the pathogenetic background of this enigmatic cardiac abnormality may be elucidated.

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