Noncompaction in Duchenne Muscular Dystrophy

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To the Editor With interest we read the article by Misumi et al. about a teenage boy with Duchenne muscular dystrophy (DMD) who had developed dilated cardiomyopathy, isolated left-ventricular hypertrabeculation/noncompaction (LVHT), systolic dysfunction, and fatal ventricular tachycardias (1). We have the following comments and concerns.

In Fig. 2, the authors attempt to present LVHT on echocardiography (1). However, the figure shown is not convincing since the structure marked with an arrow as LVHT could be also a papillary muscle. Why did the patient not undergo cardiac magnetic resonance imaging (MRI), cardiac computed tomography (CT), or ventriculography to confirm the echocardiographic diagnosis? Did the patient undergo autopsy and was LVHT confirmed on a post-mortem examination?

DMD patients frequently receive steroids for muscle weakness, however, there are indications that steroids, in particular deflazacort, have a beneficial effect also on the heart, particularly on systolic dysfunction (2). Did the patient receive steroids when admitted for ventricular tachycardias? Were steroids discontinued shortly before cardiac compromise?

DMD is typically due to deletions or duplications in the DMD-gene. Which mutation was found in the presented patient? Did the mother also carry the mutation? Were other relatives tested for dystrophin mutations?

The life expectancy of DMD patients can be significantly prolonged to 30-40 years. Ventilatory support and cardiac treatment are the main causes for prolonged survival of these patients, and LVHT is known to be associated with ventricular arrhythmias and sudden cardiac death (3). Why was implantation of an implantable cardioverter defibrillator (ICD) or administration of a LifeVest not considered? Did the patient also develop other known complications of LVHT, such as embolism or heart failure? Did the patient receive intermittent positive pressure ventilation at the time when ventricular arrhythmias occurred?

LVHT is known to be familial in 15-30% of the cases (4). Were any first degree relatives screened for LVHT? Of particular interest is the mother of the patient who could be a carrier of the mutation manifesting clinically in the heart as well as in the muscles. DMD-carriers may even present with LVHT (5).

Overall, this interesting case merits confirmation of LVHT by means of imaging techniques other than echocardiography, confirmation of the neurological diagnosis by genetic techniques, and intense family screening for dystrophin mutations and for LVHT. An ICD should be considered in DMD cases with ventricular tachycardias. Patients should be involved in the decision-making concerning the degree of invasiveness of cardiac or respiratory treatment.

The authors state that they have no Conflict of Interest (COI).

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References


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