Letter

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Management of Dystrophinopathies Should Include Non-Compaction and Cerebral Involvement

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Letter to the Editor

Duchenne muscular dystrophy (DMD) is one of the most common neuromuscular disorders in childhood, also affecting the respiratory muscles and the heart. Birnkrant et al., summarized the status of diagnosing and treating.

Critical response to a review article in Lancet Neurology

An issue which has not been addressed by Birnkrant et al., is left ventricular hyper trabeculation/non-compaction (LVHT). Recognition of LVHT in dystrophinopathies is crucial since it is complicated by thrombo-embolism originating from the intertrabecular spaces, heart failure, and ventricular arrhythmias.

When reviewing cardiac disease in DMD it is essential to include LVHT and brain abnormalities. Since LVHT is complicated by thrombus formation, heart failure, or ventricular arrhythmias, it is important for the outcome to prevent these complications by appropriate therapy.

Recently, Birnkrant et al., nicely reviewed the respiratory, cardiac, osseous, and orthopedic management in patients with Duchenne muscular dystrophy (DMD) [1]. The study raises several comments and concerns.

An issue which has not been addressed by Birnkrant et al., is left ventricular hyper trabeculation/noncompaction (LVHT). LVHT has been first described in a DMD patient by Finsterer et al., in 2005 [2]. Subsequently, LVHT has been reported in other studies. In a study of 186 Japanese DMD/BMD patients, aged 4-64 y, 19% presented with LVHT (DMD/BMD+) [3]. DMD/BMD+ patients had a lower left ventricular systolic function than DMD/BMD- patients [3]. Over a mean follow-up of 46m, left ventricular function deteriorated more rapidly in DMD/BMD+ than in DMD/BMD- patients [4]. Additionally, mortality was 37% in DMD/BMD+ compared to 14.6% in DMD/BMD- patients [3]. In a study of 151 Italian DMD patients, LVHT was detected in 10%. LVHT was first reported in a DMD-carrier by Finsterer et al., in 2012 and confirmed in 2013. In a study of 15 genetically confirmed DMD-carriers, LVHT was found in 40% on cardiac MRI (cMRI) upon application of the Peterson-criteria and 13% upon application of the Grothoff-criteria [4]. One third of the females had systolic dysfunction and 60% had late gadolinium enhancement [4].

Recognition of LVHT in dystrophinopathies is crucial since it is complicated by several abnormalities, which have a strong impact on the outcome and prognosis of DMD patients or carriers. Complications of LVHT include thrombo-embolism originating from the intertrabecular spaces, heart failure, and ventricular arrhythmias. Thrombus formation between the intertrabecular spaces may be complicated by thromboembolism into any territory, most frequently, however, in the cerebral vessels leading to ischemic stroke, the peripheral limb arteries, or the gastrointestinal arteries. Ventricular arrhythmias may be complicated by cardiac arrest, by systolic dysfunction, or by sudden cardiac death (SCD). Heart failure may be complicated by thromboembolism.

Heart failure requires appropriate heart failure therapy. If heart failure is associated with left bundle branch block and drug-resistant, implantation of a cardiac resynchronization therapy (CRT) system should be considered. If end-stage heart failure occurs, heart transplantation (HTX) is not only a theoretical option, as mentioned by the authors, but realized practice [5]. However, when considering HTX, it should be taken in mind, that immunosuppressants, required life-long after HTX, may be myotoxic and thus may worsen muscular manifestations of the disease. Ventricular arrhythmias may respond to antiarrhythmic therapy. To recognize ventricular arrhythmias in time, implantation of a reveal-recorder can be useful. If malignant ventricular arrhythmias are recorded, implantation of an ICD is

recommended to prevent SCD. If LVHT is associated with atrial fibrillation or severe heart failure, oral anticoagulation is indicated for primary prevention of stroke/embolism.

The authors also did not discuss cerebral involvement and cognitive impairment in DMD patients or female carriers.

The stimulating review by Birnkrant et al., could be more meaningful if LVHT and brain abnormalities in DMD would have been addressed. Since LVHT is complicated by thrombus formation, heart failure, or ventricular arrhythmias, it is important for the outcome to prevent these complications by appropriate therapy.

Author Contributions

JF: Design, literature search, discussion and the first draft.

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