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Letter to the Editor

Outcome of Cardiac Disease in Primary Myopathies may not only Depend on Appropriate Cardiac Therapy but also on the Neuromuscular Diagnosis

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Letter to the Editor

We read with interest the article by Andrikopoulos *et al*. about a 44-year-old man diagnosed with Becker muscular dystrophy (BMD) who developed symptoms of heart failure at the age of 35 [1]. The coronary arteries were normal. The heart failure was accompanied by moderate systolic dysfunction and was successfully treated with beta-blockers (BBs), angiotensin-converting enzyme antagonists (ACEIs), aldosterone antagonists (MRAs), and loop diuretics [1]. At the age of 39, he was diagnosed with BMD based on a muscle biopsy [1]. At age 43, his heart failure worsened to NYHA III, and at age 44, he had a left ventricular ejection fraction (LVEF) of 20%, left bundle branch block (LBBB), non-sustained tachycardia, and left ventricular hypertrabeculation (LVHT) [1]. Implantation of a cardiac resynchronization therapy (CRT-D) defibrillator resulted in clinical improvement and an increase in LVEF to 30% [1]. This study is noteworthy, but several points deserve discussion.

The first point is that we disagree with the diagnosis of BMD [1]. Since BMD was diagnosed solely on the basis of a muscle biopsy, and muscle biopsies can be misleading, it is conceivable that not BMD, but in fact a differential diagnosis of BMD, such as LGMD or metabolic myopathy, is the correct diagnosis. Immunohistochemical evidence of dystrophin deficiency may be secondary and does not necessarily indicate only a primary dystrophinopathy [1]. Generally, BMD is diagnosed based on the detection of a causative variant in the dystrophin gene on chromosome Xp21.2. Knowledge of the underlying mutation in the dystrophin gene is essential not only for the diagnosis of BMD, but also for establishing genotype-phenotype correlation, for genetic testing of other first-degree relatives, for genetic counseling, for assessing disease progression, and for evaluating whether the mutation occurred sporadically or was inherited from the patient's mother.

The second point is that it was not reported when the first clinical manifestations of BMD occurred in the index patient [1]. Knowledge of the onset of symptoms and signs of BMD is crucial because, in most cases of BMD, cardiac involvement develops after the onset of muscular manifestations. Only in rare cases have cardiac disease been reported that developed before the onset of muscular symptoms [2]. The onset of cardiac symptoms before muscular symptoms would further support the conclusion that the patient was indeed suffering from a primary myopathy alongside BMD.

The third point is that the patient did not undergo cardiac MRI prior to implantation of the CRT-D system [1]. A cardiac MRI would have been useful not only to assess the extent of myocardial fibrosis but also to determine whether late gadolinium enhancement was present and whether or not intertrabecular thrombi were present. Since LVHT can be complicated by cardiac embolism and emboli can originate not only from the left ventricle but also from the intertrabecular spaces, this information is crucial to decide whether or not the patient requires anticoagulation in addition to heart failure therapy and antiarrhythmic therapy.

The fourth point is that respiratory muscle strength was not assessed when heart failure worsened to NYHA III at age 43 [1]. Because respiratory muscle function can be compromised in patients with BMD, it is important to monitor BMD patients not only for cardiac abnormalities but also for progressive weakness of skeletal muscles, including axial and primary respiratory muscles. Distinguishing between worsening cardiac disease and muscle involvement is crucial, as treatment for both can differ significantly.

The sixth point is that cardiac medications following CRT-D system implantation were not reported [1]. Knowledge of cardiac medications following CRT-D system implantation is crucial to determine whether cardiac medications could be reduced as a result of the CRT-D system.

In summary, BMD must be diagnosed based on genetic testing, and BMD patients with LVHT require cardiac MRI to assess whether anticoagulation is indicated or not.

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