

# Detection of the mutation may guide treatment of heart and muscle in Duchenne muscular dystrophy

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## Dear editor

We read with great interest the article, by Kono et al, about a 32-year-old male with Duchenne muscular dystrophy (DMD), who was admitted for dilated cardiomyopathy manifesting as heart failure, left bundle branch block, Mobitz-II block, bradycardia, and arterial hypotension. He profited from implantation of a cardiac resynchronization therapy-D system with a defibrillator and beta-blocker treatment.<sup>1</sup> We have the following comments and concerns.

Diagnosing DMD only by muscle biopsy is insufficient. The diagnosis needs to be confirmed by genetic studies.<sup>2</sup> Was the patient investigated for deletions, duplications, or point mutations in the *DMD* gene? Which were the results of the Western blot for *dystrophin*? Was there complete absence of *dystrophin* or were there indications for a truncated protein, compatible with the diagnosis of Becker muscular dystrophy (BMD)? Since the patient was quite old for DMD, it has to be convincingly shown that BMD was definitively excluded. Diagnosing DMD on a molecular genetic level is important as genetic therapy in form of exon skipping is available for a certain type of mutations.<sup>3</sup>

It has been shown that patients with DMD profit from the administration of steroids, usually deflazacort.<sup>4</sup> There are indications that steroids not only improve the performance of the skeletal muscle but also improve cardiac functions.<sup>5</sup> Furthermore, steroids reduce the myocardial fibrosis burden in these patients as has been shown by cardiac magnetic resonance imaging (MRI).<sup>6</sup> Did the presented patient receive steroids in addition to heart failure therapy? Did he profit from steroids with regard to cardiac and muscle function? Which side effects did he develop from steroids?

Dilated cardiomyopathy in DMD is characterized by replacement of the myocardium with connective tissue.<sup>6</sup> Myocardial fibrosis in DMD can be best documented by application of cardiac MRI and administration of gadolinium to assess the amount of late gadolinium enhancement (LGE).<sup>6</sup> Did the patient undergo cardiac MRI and was gadolinium administered to assess the amount and location of myocardial fibrosis? Assessment of LGE in DMD is important since the amount of LGE strongly correlates with left ventricular systolic dysfunction.<sup>6</sup>

Approximately two-thirds of the DMD patients inherit the mutation from their mothers. DMD carriers may manifest clinically or biochemically. Was the mother of the described patient investigated for her carrier status? Did she manifest clinically or were there other indications for being a DMD carrier? Did she undergo electrocardiography

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(ECG) recording or echocardiographic investigations? Was a cardiac MRI carried out and did it show LGE as has been previously reported.<sup>7</sup>

Overall, this interesting case presentation would profit from more extensive genetic and cardiologic investigations to assess the degree of myocardial fibrosis and from evaluation of the genetic background in this particular patient and his relatives.

## Disclosure

The authors report no conflicts of interest in this work.

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