

Letter to the Editor

Muscle involvement in Duchenne muscular dystrophy progresses differently, as shown by MRI and diffusion tensor imaging

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Dear Editor,

We read with interest the article by Sane *et al.* [1] on a retrospective cross-sectional study on the characteristics of MRI pattern of muscles, the correlations between MRI T1-W grade score and muscle strength and pattern of muscle involvement, the correlation between fractional anisotropy (FA) and muscle strength, and the correlation between FA values and T1-W grade score of fat infiltration in lower limb muscles of 78 patients with Duchenne muscular dystrophy (DMD). The MRI grade score and Vignos scores increased with age [1]. Muscle strength and FA scores decreased with age [1]. There was a distinct pattern, extent, and distribution of lower limb muscle involvement [1]. The study is noteworthy, but some points should be discussed.

The first point is that the results of the genetic tests were not reported [1]. Did all included patients carry a pathogenic mutation in the dystrophin gene? We should know in how many patients DMD is due to a point mutation, deletion, or duplication in the dystrophin gene. It is important to know the type and extent of the mutation because there may be a genotype–phenotype correlation that allows not only the prediction of disease progression but also the assessment of whether the type and extent of the mutation correlates with the severity of muscle involvement on imaging.

The second point relates to the retrospective design of the study [1]. Retrospective designs have several disadvantages [2]. They allow only limited control over the sampling of the population and only limited control over the type and quality of predictor variables. In addition,

the relevant predictors may not have been recorded in the medical record, and it may be difficult or impossible to detect confounding variables and causality. Furthermore, it may be inevitable that some information is missing because the data are based on the review of medical records that were not originally intended for the collection of data for research purposes. Selection and recall errors also affect the results, and the reasons for differences in the number of lost to follow-ups often cannot be determined, which can lead to bias [2].

The third point is that the kind of treatment the included patients regularly received was not reported [1]. We should know what is meant by the “standard treatment” that the patients were receiving at the time of the MRI examination [1]. Did all patients really receive the same treatment? Since DMD is characterised by cardiac involvement, we should know how many of the included patients had dilated cardiomyopathy and required drug treatment. Cardiac disease in DMD starts between 10 and 15 years of age and can be treated with different approaches [3]. Therefore, it is quite unlikely that all included patients received the same “standard therapy”.

The fourth point is that muscle volume and architecture may strongly depend on whether a patient is ambulatory or non-ambulatory. Since 32 patients were ambulatory and 46 were non-ambulatory, this fact alone may lead to different MRI results, regardless of the stage of the disease [4]. Was there a difference in outcome parameters between these two groups?

The fifth point is that it is unclear how the slice planes for the MRI slices were standardised so that they were acquired at the same level in each patient. There is also no

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mention of whether the inter-observer and intra-observer variability was high or low and whether the examinations were repeated in some patients at some time point.

In summary, this interesting study has limitations that affect the results and their interpretation. Addressing these limitations could strengthen the conclusions and support the message of the study. All unanswered questions need to be addressed before readers uncritically accept the conclusions of the study. Muscle involvement in DMD, as assessed by MRI and DTI, varies in progression,

presumably depending on mutation type, degree of mobility, and type and dosage of current medications.

Disclosures

1. Institutional review board statement: Not applicable.
2. Assistance with the article: None.
3. Financial support and sponsorship: None.
4. Conflicts of interest: None

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