



Letter to the Editor

Reply to the letter entitled “Predictors of respiratory impairment in patients with myotonic dystrophy type 1”



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Dear Editor,

We read with interest the letter of comments “Predictors of respiratory impairment in patients with myotonic dystrophy type 1” by Prof. Kawada [1] about our work concerning the assessment of the prevalence and predictors of respiratory impairment by a cross-sectional study on a large Italian cohort, recently published on Your Journal [2].

His main comment regards the argument that our results would be biased by the fact that they have been obtained through a cross-sectional study [1], as we also specified this point as one of the limitations of our paper [2]. It is definitely recognized, and Prof. Kawada underlined it as well, that the worsening of the respiratory functions, in terms of decline of Forced Vital Capacity (FVC), is slowly progressive in Myotonic Dystrophy type 1 (DM1) patients [3]; this means that conducting a prospective study would require many years to be performed on a large cohort as we did. Moreover, a longitudinal assessment, besides being long lasting, should also take into account the chance of a significant drop in the follow up of DM1 patients related to their cognitive and behavioural symptoms [4].

A cross-sectional study as we conducted it, on the other hand, is an approach also adopted by other groups with recognized expertise in DM1 clinical research [5,6]. Although with limitations, it is in our opinion nevertheless reliable, allowing to collect extensive data from a significant cohort of DM1, being a rare disease, that also covered the full disease severity spectrum; moreover, all data used for the statistical analysis concerned standard diagnostic procedures routinely applied in all the Participating Referral Centers for DM1, and accurately collected from patients' medical records by the co-authors.

In his letter, Prof. Kawada also pointed out that the mechanisms, the natural history and time-course of respiratory functional decline have been poorly understood, being complex and multifactorial [1], and we also agree with this observation, and indeed we specified this particular issue in the introduction of our paper [2]. However, our results and previous literature data strongly support the major role for the skeletal muscle weakness in the pathophysiology of respiratory insufficiency in DM1 [7–9].

Regarding the last point, the primary aim of the present study was

to evaluate the occurrence of respiratory impairment. We indicated “respiratory impairment” as the presence of restrictive lung disease, and we defined it as FVC < 80% of the predicted. Therefore, we applied the logistic approach to define the predictors. The multivariate regression analysis uses FVC as a continuous variable, and identifies parameters that are correlated with the values of FVC, rather than with the condition which we defined “respiratory impairment”. For these reasons, we preferred to use a cut-off value for FVC, and, consequently, a multivariate logistic approach.

Conflicts of interest

All the authors declare that they have no conflict of interest.

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