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Letter to the Editor

Letter of response to "Myotonic dystrophy type 1, individualised respiratory care rather than standart prognostication"

Dear Editor,

We read with interest the letter "Myotonic Dystrophy type 1, Individualized Respiratory Care Rather Than Standard Prognostication" by Dr. Yetimakman and colleagues [1] refering to our paper recently published on your Journal [2] and carefully revised the comments and issues raised in this letter.

Indeed, we fully agree with them regarding the complexity of the pathophysiology of respiratory impairment in DM1 involving different aspects related to the multisystem involvement of the disease, and indeed we underscored this topic in the introduction section of our paper [2].

They argue about the significant association found in our study between restrictive pattern and indication to Non Invasive Ventilation (NIV) in Myotonic Dystrophy type 1 (DM1) patients; our findings indicate that pulmonary restrictive syndrome would be a major determinant for NIV indication, but this does not mean that indication to NIV should be accounted only by this parameter. We clearly stated in the Materials and Methods section [2] that indication to NIV was addressed in the presence of symptoms suggestive of chronic respiratory insufficiency plus the presence of diurnal hypercapnia, evidence of nocturnal hypoventilation at nocturnal pulse-oxymetry, FVC $<$ 50% of predicted or AHI $\geq$ 10 events/h, in agreement with the DM1 Expert Panel Consensus Statement about the respiratory management of DM1 patients [3].

Moreover, Yetimakman and colleagues remind that FVC could be biased by other specific features in DM1 patients; again, we are fully aware that either the facial weakness or the poor compliance during the execution of the PFTs could lead to technically inaccurate data: therefore (see again ourMaterials and Methods), we did not include for the analysis PFT results likely biased by the above mentioned issues according to the pneumologist or the technician assisting DM1 patients during the PFT.

So, data from our work clearly state that not only FCV value but also hypercapnic failure and functional status should be taken into consideration to start NIV in DM1 patients. However we do agree with Yetimakman and colleagues that the benefits of NIV would be uncertain in DM1, supported in their opinion by the results by O'Donoghue et al. [4]: in fact, in our opinion that study shows important limitations, in particular a very small cohort of study, including only 12 DM1 patients, all of whom had also mild respiratory failure, which do not reflect the clinical spectrum of the DM1 population, and a time span of evaluation (2-months) that is too short to assess any changes in the lung capacity function in such patients.

Instead, we again emphasize that even if adherence to NIV therapy can often be a challenge in DM1 patients for well known factors, mainly the contribution of the CNS involvement, researchers clinicians dealing with DM1 will have to increase their efforts to overcome this critical issue[2,3,5]., as increasing DM1 patients' compliance to NIV could allow to reduce morbidity and mortality due to respiratory causes.

Gabriella Silvestri, MD, PhD and Salvatore Rossi, MD, on behalf of all the co-authors of the paper.

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