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Letter to the Editor

Myotonic Dystrophy type 1, individualised respiratory care rather than standart prognostication

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Dear Editor,

We read with interest the article “Prevalence and predictor factors of respiratory impairment in a large cohort of patients with Myotonic Dystrophy type 1 (DM1): A retrospective, cross sectional study” by Rossi et al. [1].

The authors have studied predictor factors of respiratory impairment to evaluate the risk of severe respiratory complications since the respiratory involvement is relevant to prognosis in DM1 patients. They point out that indication to NIV is associated to restrictive pattern in DM1 patients.

First, it is a fair question whether FVC is a sensitive measurement to assess progression of muscle weakness. It is feasible but challenging to obtain accurate measurements of pulmonary function in the clinic in patients with NMD. There is the risk of including technically inaccurate data from subjects unable to both inhale and exhale fully with maximal effort. This is more likely to occur in the weaker subjects, and will result in an erroneously low FVC [2].

Second, we want to underline some aspects regarding the indications and effect of NIV in this population. Progressive respiratory muscle weakness is the common indication for starting NIV treatment in many neuromuscular disorders. In patients with DM1, muscle weakness may develop slower. Respiratory failure may be related to abnormal respiratory control at medullary level rather than due to nocturnal retention of CO2 [3]. Daytime somnolence is often present in these patients without sleep disorder [4]. This is also attributed to CNS involvement and not responsive to sleep disorder treatment.

Although the authors argue that patients with more severe symptoms appreciate the benefit and tend to cooperate more, it is well established that adherence to NIV therapy is a challenge in these patients. This is because of the patient's unawareness of respiratory muscle impairment, because of the disease affecting the frontal lobes.

In a study by O'Donoghue et al. [5] evaluating the effects of withdrawal of NIV in DM1 patients with chronic hypercapnic respiratory failure; patient reported parameters, sleep structure and fragmentation was unchanged and ventilation parameters such as hypercapnic ventilator response (HCVR) were only changed slightly. This implies treatment might not be needed in these patients from the aspect of functional status. They claim that lack of symptomatic improvement may be one reason why DM1 patients have been reported to tolerate NIV poorly. In the article by Rossi et al., there was only 50% compliance with NIV, even in their subgroup with restrictive syndrome. This also justifies the suggestion that the decision to treat or not in this population is different to that in other forms of neuromuscular disease and delaying initiation of NIV should be considered.

Not only pulmonary function demonstrated by FVC but also hypercapnic failure and functional status of the patient should be taken into consideration.

Conflicts of interest and source of funding

None declared.

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Abbreviations: DM1, Myotonic Dystrophy type 1; FVC, forced vital capacity; HCVR, hypercapnic ventilator response; NIV, non invasive ventilation; NMD, neuromuscular disorders

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