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Editorial

Characterizing the Sleep Disordered Breathing in Neuromuscular Diseases - Beyond the Polysomnography

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Sleep Disordered Breathing in Neuromuscular Diseases

The development of chronic respiratory failure is the major problem in Neuromuscular Diseases (NMD), leading to increased morbidity and mortality, and their first signals are exhibit during the sleep. Around 80% of patients with NMD exhibits perturbations in sleep architecture and in gas exchange [1]. The method considered the gold standard to evaluate these perturbations is the Polysomnography (PSG). Nevertheless, it is expensive, not accessible, and it is even difficult to perform NMD patients. Even home PSG presents limitations [2] majorly for complex patients or diseases.

The development of chronic respiratory failure in NMD has as starting point a nocturnal respiratory insufficiency. Therefore, it is important to characterize and treat respiratory failure earlier, due to positive effects [3]. On the other hand, signals and symptoms seen during the day, although they appear later, may indicate the presence of Sleep Disordered Breathing (SDB) in NMD. Therefore, it is important to consider pulmonary function tests and respiratory muscle tests to better characterize and, hereafter, aim a treatment for the respiratory failure.

SDB in NMD

Normally, during the sleep there is an increase at the $PaCO_2$ with a concomitant decrease of SaO_2 levels, caused by a decrease in tidal volume (Vt) [4], at the respiratory rate [5], and an increase in the resistance of upper airways [6]. The non REM (NREM) phase presents an increase at the activity of intercostal muscles without changes at activity of the diaphragm. During the REM phase the diaphragm is the only muscle to present activity [7]. Then, there is clearly an increase in the work of breathing during sleep.

At the NMD there is a progression at the impairment of the activity of skeletal muscles, leading to thoracic deformations and reduction of global mobility. These perturbations will lead to reduction at mobilisable lung volumes associated with an increase in residual volume [8]. The aforementioned alterations in SaO_2 and CO_2 appear initially during the REM phase. However, with the maintenance of these perturbations, the chemoreceptors will become desensitized with a depression of central drive, culminating in an augmentation of CO_2 and HCO_3 levels.

Concerning the respiratory muscles, the diaphragm [9], the intercostal and the dilatators of upper airways may be affected [10]. Due to restrictive pattern achieved there is a development of reduced elastic distension, leading to a more pronounced instability in upper airways, arising a Obstructive Sleep Apnea (OSA), as seen earlier in Duchenne Muscular Dystrophy (DMD) [11] and in Pompe disease [12].

Another SDB, the hypoventilation leads to abnormalities in gas exchange and may be caused by a more pronounced impairment at the respiratory muscles during the REM phase. The hypoventilation is commonly seen in Myotonic Dystrophy [13], or in sequelae post-polio [14].

Daytime Predictors and/or Correlations with SDB in NMD

Due to the thoracic deformation and reduction at lung volumes, there have been exhaustively described relations between pulmonary function tests and SDB. Chronologically, in 1989, Heckmatt et al observed that majority of the patients with DMD who presented diurnal hypercapnia also had increased values for HCO_3. The majority of the patients presented inspiratory muscle weakness measured by Maximal Inspiratory Pressure (MIP) and this was associated with diurnal hypercapnia [15]. Bye et al described a correlation between Vital Capacity (VC) and SaO_2 during the REM phase [16]. Barbé et al presented correlations between Apnea/Hypopnea Index (AHI) with daytime PaO_2, and in a multivariate analysis, between AHI with PaO_2 and Forced Expiratory Volume at the first second (FEV1) [17]. White et al [18] found that nocturnal oxygenation correlated inversely with postural fall in VC, and then with diaphragmatic strength.

Hukins et al demonstrated, in a population of DMD patients, that those who already have diurnal hypoventilation presented also a nocturnal hypoventilation, and also that values of FEV1 lower than 40% of the predicted was a sensible, nevertheless not a specific, measure to detect SDB in NMD [19]. Ragette et al also established cut-off values for Inspiratory Vital Capacity (IVC) and MIP for the occurrence of hypoventilation during the REM phase, and for whole night hypoventilation and diurnal respiratory insufficiency [20]. Similarly, Mellies et al demonstrated a correlation between MIP and IVC with SDB, concluding that SDB can be perfectly predictable by daytime respiratory function tests [21]. Weinberg et al observed a correlation between daytime base excess with the nadir oxygen saturation, and a correlation between VC with obstructive sleep apnea index [22]. Toussaint et al established a cut-off values for VC, MIP and for the relation Vt/VC to predict the occurrence

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of nocturnal and diurnal hypercapnia studying DMD patients [23]. Romei et al showed that the abdominal contribution to inspiratory capacity was a good measure to discriminate the DMD patients who presents desaturation [24].

Recently, Quaranta et al demonstrated a correlation between Sniff Nasal Inspiratory test with (SNIP) AHI, with the time during the sleep that presents a SpO_2 lower than 90%, and with the index of desaturation. The authors affirmed that SNIP values lower than 60cmH_2O was associated with lower SaO_2 [25].

Conclusion

The PSG remains the most accurate measure to identify SDB in NMD, and it still necessary to follow up patients with NMD and it evenly allow to better manage the parameters of noninvasive ventilation [26]. Nevertheless, preliminary evaluation of daytime respiratory function may not only indicate the presence of SDB, but also act as complementary measures to better characterize respiratory failure and SDB in NMD.

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