ACTA MYOL 2024;XLIII:48-56
doi: 10.36185/2532-1900-553

ORIGINAL ARTICLES

Indications and techniques of non-invasive ventilation (NIV) in neuromuscular diseases

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The neuromuscular patients may experience the need for respiratory support due to the onset of respiratory failure. Some skills are essential to achieve therapeutic success. In addition to technical knowledge, it is essential to have knowledge relating to individual neuromuscular diseases. The availability of alternative respiratory supports and various interfaces can be a valuable weapon at different moments in the course of the neuromuscular disease. Furthermore, the doctor-patient relationship plays a key role as does taking care of the patient's psychological sphere.

Key words: neuromuscolar diseases, non-invasive ventilation, respiratory failure

Introduction

Patients with neuromuscular disorders (NMDs) develop respiratory impairment as muscles weaken.

The decision to initiate assisted ventilation is multifactorial, and may include consideration of patient symptoms, spirometry, pulmonary pressures, sleep studies or blood gas values [1-5]. Non-invasive ventilation is considered a standard of care in respiratory failure due to neuromuscular disease [6,7]. The evolution of technology and the definition of ultra-specialist professional figures has increased the possibilities for treating some pathological conditions. In the field of ventilation, this progress has made it necessary to acquire specific skills for patient care and the continuous updating of doctors who implement non-invasive mechanical ventilation, as well as knowledge of the new technological supports available on the market.

In this article, we describe the indications and techniques of non-invasive ventilation (NIV) in neuromuscular diseases that can guarantee greater success in their application.

Overview of available respiratory supports

In the history of non-invasive mechanical ventilation, we have moved from negative ventilation to positive abdominal pressure with mouthpiece, to positive pressure ventilation through a nasal or oro-nasal mask to return in selected cases to previous methods.

The demand for mechanical ventilation developed during the outbreak of the poliomyelitis epidemic in the early-to mid-twentieth century. The development of a negative pressure ventilator[8] with the use of the iron lung, decreased mortality by about 50%[8].

Received: June 2, 2024
Accepted: June 27, 2024

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How to cite this article: Annunziata A, Coppola A, Simioli F, et al. Indications and techniques of non-invasive ventilation (NIV) in neuromuscular diseases.. Acta Myol 2024;43:48-56. https://doi.org/10.36185/2532-1900-553

Negative pressure ventilation (NPV)

Negative pressure ventilation (NPV) [9-15] has played a crucial role in the past; however, today, it is still used only in a few hospital departments. This technique has some strengths and is able to guarantee breathing that is completely similar to the physiological one, consisting of a mechanical inspiratory phase followed by a passive expiratory phase. Both phases are applied using a negative pressure fan and some accessories attached to it, such as a curia or poncho.
NPV preserves physiological functions such as speech, coughing, swallowing, and feeding, and its main benefit is the prevention of endotracheal intubation and related problems [10-14]. The limitations of the method are the lack of protection of the upper airways, particularly in unconscious patients, which can cause aspiration, considering the described impact of NPV on the lower oesophageal sphincter [15]. Upper airway obstruction may occur in unconscious patients, in patients with neurological disorders with bulbar dysfunction, and in those with sleep apnoea syndrome [15]. NPV has been used successfully in acute and post-acute situations [16] (Fig 1). Its use has been described in a

Figure 1. A DMD patient, intolerant to positive pressure ventilation, treated with NPV during acute respiratory failure due to pneumonia.

case of nemaline myopathy complicated by pneumothorax [17]. NPV was used, in combination with other respiratory supports to enable weaning from oro-tracheal intubation in a patient suffering from acute respiratory failure with hypoxemia, hypercapnia, and respiratory acidosis [18] There are no guidelines on the use of NPV.

Non-invasive Positive Pressure Ventilation (NPPV)

Over 30 years ago the first authors described the effectiveness of Non-invasive Positive Pressure Ventilation (NPPV) in patients with NMD indicating that it could be used to chronically ventilate patients with neuromuscular diseases [19-24].
Acute respiratory failure is also a common life-threatening complication of acute onset neuromuscular diseases, and may exacerbate chronic hypoventilation in patients with neuromuscular disease or chest wall disorders [25]. Today, NPPV can be applied with a variety of interfaces, ventilators, and ventilator settings and it has become routine practice in many centres and is commonly used in patients with respiratory failure associated with neuromuscular disease.
The use of NPPV in this patient population is likely to expand, particularly with increasing evidence that this therapy is life prolonging in patients with diseases such as amyotrophic lateral sclerosis [26,27].
Appropriate selection of equipment and settings for NPPV is paramount to the success of this therapy. The selection of equipment for NPPV is based on the physiological needs of the patient, the clinician's familiarity with NPPV, the desires of the patient, and the availability of equipment [28,29].
Initially, the NPPV settings are selected based on short-term symptoms such as chest expansion, accessory muscle use, and comfort. Often, the prescribed settings are a compromise between those likely to be therapeutic and those tolerated by the patient. In some cases, the initial settings may be sub-therapeutic, but they are further adjusted as the patient becomes increasingly tolerant of the therapy, with the goal of improving symptoms such as morning headache, fatigue, and daytime sleepiness. NPPV is now widespread in many centres.
NPPV is recommended as the first-line therapeutic approach for the management of acute hypercapnic respiratory failure whether or not related to neuromuscular disorders [29]. NPPV by avoiding endotracheal intubation, can decrease the patient's exposure time to mechanical ventilation and to Intensive Cure Unit (ICU), reduce lung infections, barotrauma, tracheal stenosis, and the requirement for tracheostomy [30-38]. Furthermore, starting non-invasive ventilation and therapies to improve cough and manage secretions at the right time can preserve muscle function, alter the natural progression of the disease and reduce hospital admissions [30-38].
However, NPPV efficacy may be dramatically reduced by the presence of bulbar dysfunction and by excessive bronchial secretions [30-38].
NPPV may also be associated with sudden deterioration in respiratory function and vital signs that require immediate tracheal intubation, which can be challenging.
Although there is no evidence from randomised trials to support the routine use of NPPV instead of invasive ventilation in patients with acute neuromuscular respiratory failure, some evidence from observational studies however suggests that NPPV should be tried with caution in patients with severe bulbar dysfunction [38-40].
Greater experience in the use of the different modes of assisted ventilation is often required, based on the clinical picture, which can also vary between daytime and night-time treatment. However, for the chronic ventilation of neuromuscular patients different NPPV alternatives are available to clinicians.

Mouthpiece ventilation (MPV)

Mouthpiece ventilation (MPV) is a type of noninvasive ventilation delivered through a mouthpiece [41]. The mouthpiece allows for supportive ventilation with the possibility of detaching for talking or eating. Often,

it may be a rescue strategy for noncompliant patients in Duchenne muscular dystrophy (DMD) [42], and in neuromuscular disorders in general [43]. Best practice guidelines for management of mouthpiece ventilation in neuromuscular disorders have been developped [44].
However, desaturations are possible both during MPV and in case of mechanical ventilation with a mask due to the increase in resistance due to the presence of secretions and excessive losses of the system [45]. The low-pressure warning system of mouthpieceless ventilators in NIV mode can be de-activated while their use without backup respiratory rate can be useful in less dependent patients (frequent disconnections); on the other hand, severely ventilator-dependent patients can benefit greatly from a more responsive ventilator, with greater speed in tidal volume regulation [11].

Intermittent Abdominal Pressure Ventilator (IAPV)

Intermittent Abdominal Pressure Ventilator (IAPV) is an attracting alternative respiratory support whose mechanism of action involves intermittent abdominal compression without any interface on the patient's face [46]. The device consists of a corset with an elastic inflatable bladder that fits over the abdomen. A tube connects the bladder to a ventilator that delivers air to the bladder and abdominal wall (Fig. 2), lifting the diaphragm to cause exhalation below functional residual capacity [46].
IAPV has been used in several neuromuscular patients: traumatic tetraplegic ventilator-dependent patients, spinal cord injury, non-Duchenne myopathies, Duchenne muscular dystrophy, myelopathy, polymyositis [47-49]. There are no guidelines on the use of IAPV and on the parameters to be set. Indications are derived from case reports and experience; however, an expert consensus document on IAPV has recently been published [45].

Figure 3. A LGMD patient affected by acute respiratory failure due to pneumococcal pneumonia, treated with HFNC and NPV.

It is important to stress that MPV and IAPV are possible alternatives to NIV but they request the patient's cooperation[50,51]. Both are good options in selected cases, in an acute context and can easily be used in the weaning or post-acute phase. NPV may be used in those

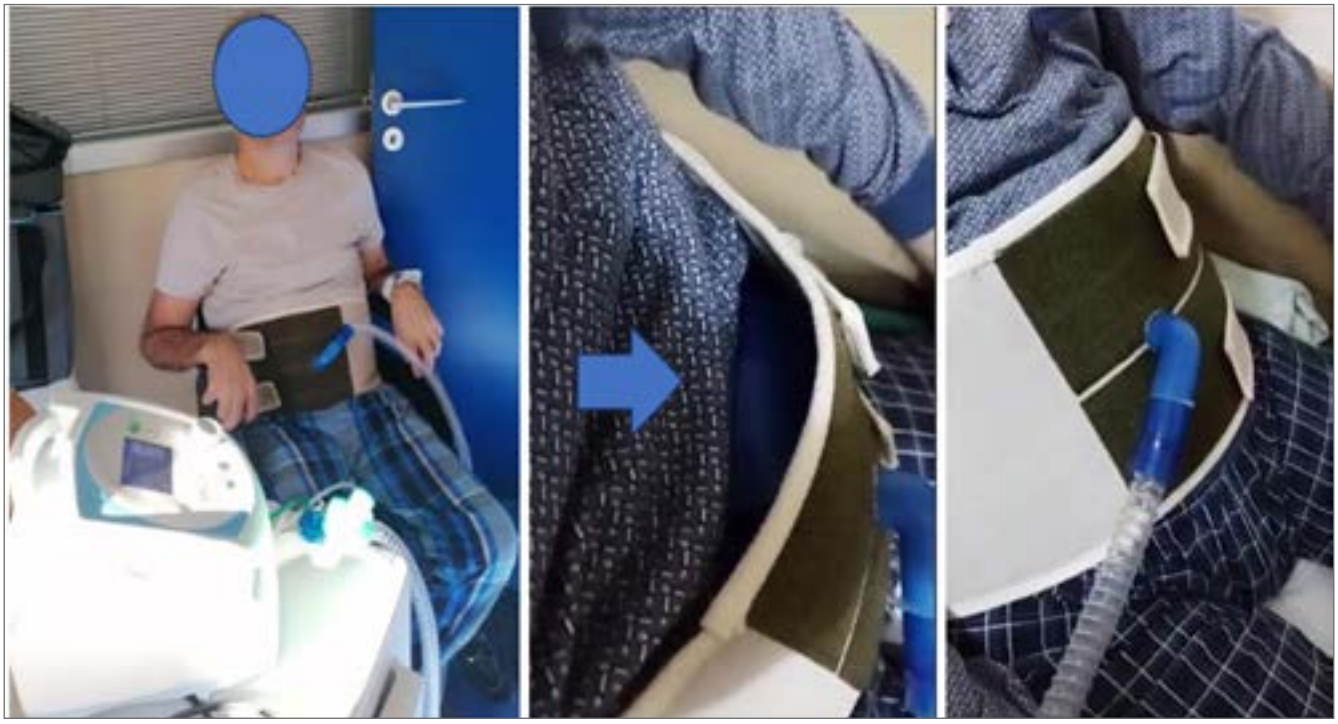

Figure 2. A LGMD patient during the IAPV study. The blue arrow indicates the elastic inflatable bladder that compresses the abdomen to cause exhalation.

patients who cannot tolerate a face mask due to facial deformities, claustrophobia, or excessive airway secretion [18]. Patients requiring 24 hours respiratory support can benefit from NPV alternate with positive-pressure ventilation, or in combination with high flow nasal cannula [18] (Fig. 3).

Timing of the respiratory support in neuromuscular patients

There is no unique answer. It depends on the neuromuscular disease and its clinical evolution: weakness of the respiratory muscle leading to respiratory failure, swallowing difficulties, loss of ambulation, with wheelchair-confinement [1-5] are all factors that influence the choice.

When choosing the right time to start ventilation, it is necessary to distinguish neuromuscular diseases (NMD) into three categories: i) rapidly progressive disorders such as amyotrophic lateral sclerosis (ALS) and spinal muscular atrophies (SMA), which worsen over months leading to death within a few years; ii) relatively rapid progressive diseases such as DMD in which death occurs within young adulthood and iii) slowly progressive diseases such as Becker muscular dystrophy (BMD), FacioScapuloHumeral Dystrophy (FSHD), Limb-Girdle Muscular Dystrophies (LGMDs) and Myotonic Dystrophy type 1 (DM1), that experience a slowly progressive reduction in muscle function and a slight reduction in life expectancy [1-5].

NIV is a current and effective treatment for respiratory failure that can reduce diurnal and nocturnal symptoms with improvement of quality of life and survival. It should be early used when there is an evident respiratory involvement. The great benefits related to the early use of NIV in NMDs have supported the anticipation of the timing and setting for the initiation of NIV [52] early.

Current international consensus guidelines recognize some indicators of when to initiate long-term NIV in patients with NMDs, but do not make clinical differences among the different underlying disorders[5]. All patients with a diagnosis of rapidly progressive NMD should perform respiratory functional tests every 2-3 months. In particular, the progressive and significant decline in Forced Vital Capacity (FVC) is an indisputable indicator of the need for NIV.

In ALS, NIV should be started when FVC is less than 80% of the predicted value if associated with respiratory symptoms such as dyspnea, use of accessory respiratory muscles, tachypnea, excessive fatigue, excessive daytime sleepiness [27,29,53]. Another indicator for starting NIV is a sniff nasal inspiratory pressure (SNIP) < 40 cm H2O or a maximum inspiratory pressure (MIP) < 60 mmH2O. It is important to say that the decline in respiratory function over three months associated with respiratory symptoms, is more useful to assess respiratory involvement in clinical practice rather than absolute values [29].

A further important assessment is the detection of nocturnal hypoventilation (NH). NH is a complication of respiratory system in NMDs that can lead to symptomatic daytime hypercapnia. Various definitions of NH have been currently proposed but which of these best relates to prognosis remains unclear [30,37]. In patients with NMDs, a correlation between sleep disorders and reduction in FVC has been shown [54]. In a recent study, Boentert et al. [29] showed that oximetry alone and even polygraphy are not sufficient to reveal the presence of nocturnal hypoventilation; furthermore, the combination of bicarbonate excess in the early morning and desaturation time (t < 90%) are also independent predictors of NH.

Table I. Indications to start NIV in NMDs.

• FVC < 80% associated with symptoms such as tachypnea and use of accessory muscles, tachypnea, excessive fatigue, excessive daytime sleepiness
• SNIP < 40 mmHg
• MIP < 60 mmHg
• Daytime hypercapnia PCO2 > 45 mmHg
• Nocturnal saturation < 88% for 5 consecutive minutes

Transcutaneous capnography is strongly recommended for an accurate detection of NH in patients with ALS [37]. Furthermore, residual hypoventilation is significantly associated with adverse outcomes in ventilated adult patients with NMDs [37]. The presence of daytime hypercapnia with PCO2 > 45 mmHg and nocturnal oximetry showing an oxygen saturation ≤88% for 5 consecutive minutes, are considered an expression of NH and are indicators to start NIV in NMDs [26-28] (Tab. I).

There are no validated criteria to start long term NIV in children. In clinical practice, NIV may be initiated in an acute setting after invasive or NIV weaning failure in pediatric intensive care unit, in a subacute or chronic setting, due to the presence of abnormal nocturnal gas exchange alone or in association with respiratory events in a polysomnography [41,48,49].

Identification of the best interface for the patient

Oro-nasal masks are used by at least 25% of neuromuscular patients, mainly to counteract persistent and involuntary oral losses, which would be more frequent if nasal masks were used [54]. Furthermore, they can cause persistent obstructive breathing episodes of the upper airways due to the pressure of the chin area which can push the jaw back during sleep favored by the tension of the straps which maintains the position of the interface [48,52].

Nasal mask can improve upper airway stability and effectiveness of non-invasive ventilation by reducing side effects and allowing lower applied pressures; in the meantime, there could be an increase in involuntary losses with an increase in desaturations and asynchronies, and a worsening of the patient's prognosis [53,54]. The choice of the interface should also include the ability to modify it during the day, particularly for ventilator-dependent patients. It is necessary to meet the patient's needs and try to resolve any issues related to leakage and non-tolerance to prevent interruption of the NIV. There are currently no randomized studies available on the use of nasal and/or oro-nasal masks in neuromuscular patients.

The main suggestions of an optimal interface are:

- optimal sealing;
- comfort;

- easy to apply and remove;
- test and prescribe at least two different interfaces resting on different areas, allowing for alternative use to minimize pressure sores;
- evaluate the packaging of masks tailored to the patient's face.

Prolonged ventilation is associated with the progressive deterioration of physical conditions and skin trophism favoring facial pressure ulcers, especially on the nasal root and frontal skin. Prevention of these complications is essential and it is better to tolerate minimal air leakage from the mask rather than risk injury to the skin. In the case of multiple hours of ventilation, it is necessary to have multiple interfaces to vary the skin pressure points, with the use of hydrocolloid material placed on the anti-face. A potential alternative is the full-face interface, which is easy to apply to the patient's, reducing pressure ulcers (Fig. 4).

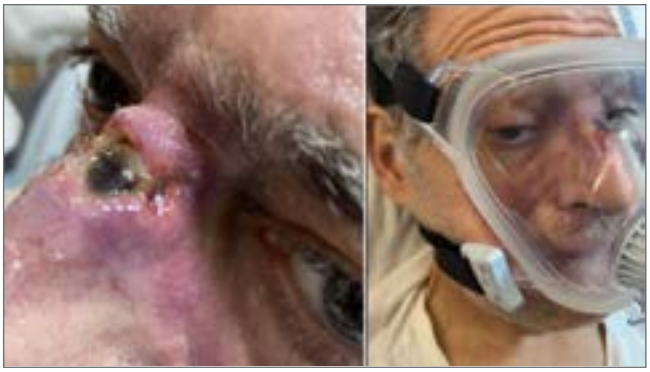

Figure 4. Ulcer of the bridge of the nose in an ALS patient in treatment with NIV 24 hours a day. The same patient in a full-face mask.

Strategies to prevent or reduce facial ulcers or erythema include the use of a polyethylene oxide/water hydrogel dressing, a silicone foam dressing, and fabric NIV interfaces. Prevention or treatment of skin ulcers can also be achieved by opting for a fabric mask or a custom-molded mask.

Research indicates that midface hypoplasia significantly complicates respiratory support via a nasal mask especially in children as up to 30% of them have malocclusion problems. Using different masks or preferring a cloth mask or a custom-molded mask can help mitigate the development of midface hypoplasia or craniofacial malformation. Bockstedte et al. [55] described a study on customized interfaces for NIV with three-dimensional scans acquired using an intraoral scanner and a facial scanner (3dMd Flex System). The goal was to create a quick and affordable custom NIV solution manufactured in-house, that improves both fit and comfort for the patient. Despite the higher costs of customized masks compared to standard ones, it has been shown that in pediatric patients, it is possible to improve their effectiveness with a better seal, simultaneously obtaining an improvement in general comfort[55]. In recent years, the new interfaces with minimal support on the face that leave the bridge of the nose completely free have offered clinicians an additional weapon for NIV; the interface is often comfortable and appreciated by patients also for the possibility

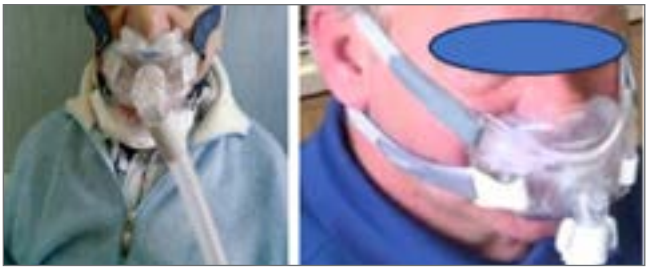

Figure 5. Interface without pressure on the nose bridge.

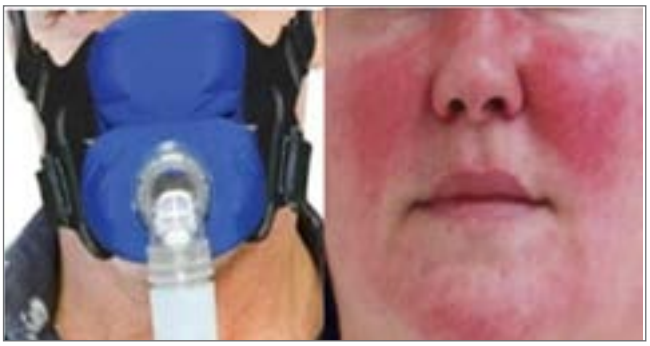

Figure 6. Cloth nose-mouth interface.

of wearing glasses or in any case having the bridge of the nose free (Fig. 5). Cloth nasal or oro-nasal interfaces are available for patients with skin allergies to silicone (Fig. 6). It is useful to have a wide range of interfaces to find the most comfortable and least leaky interface for the individual patient.

Psychological aspects and doctor-patient interaction

Psychological aspects

The doctor-patient relationship plays a key role when communicating the diagnosis and sharing the therapeutic process which is often only palliative. The patient's basic psychological structures are altered with an initial refusal of ventilatory treatment in the majority of cases [56-61]. Fear of death has been described as a factor favoring the acceptation of NIV, with a higher rate of initial adherence.

It is important to make the patient understand that the gradual acceptance of NIV leads to beneficial effects on the course of the disease and on management by the caregiver with a positive lifestyle.

Though the psychological aspects are significant and can be different in different pathologies, however there are many gaps in the literature on this aspect. Most research on the psychological determinants of NIV outcomes has been conducted in patients with ALS [58] while a small number of evaluations have been conducted in children and elderly patients with NMDs and acute respiratory failure. The psychological profile and disability are perceived differently by the patient suffering from ALS or chronic obstructive pulmonary disease compared to the patient with early onset muscular dystrophy. Different diseases have different times of onset of disability and very different

life expectancies, and this certainly interferes significantly with the treatment process that also depends on the presence or absence of a caregiver [59,60]. Doctors must take all these indicators into account when approaching the neuromuscular patient with respiratory failure.
The physical deterioration and reduction in the quality of life produces a direct impact on the psychological sphere which, with the worsening of endogenous depression [60,61] and anxiety, leads to an increase in the number of hospitalizations for exacerbations and episodes of dyspnea [61], and a worsening of the quality of life, with increased risk of death.
Patients suffering from chronic respiratory disease must face a progressive modification of their lifestyle with the need to quickly adapt to new and sudden changes that cause a continuous imbalance in the patient's already fragile psychological structure. Very often patients are forced due to the disease to abandon their habits and give up hobbies and passions that would allow an improvement in their psychological harmony.
The mask used for NIV is often perceived by patients as a wall that limits their autonomy; in fact, it interferes with social life and is seen as a source of vulnerability due to the impossibility of repositioning or removing it easily and immediately as they are unable to move their upper limbs.
It is no coincidence, in fact, that greater adherence to ventilation is correlated to patients with greater motor autonomy or with a highly compliant family environment [61-63].
What drives some patients to refuse an essential therapy such as NIV despite the perception of dyspnea, fatigue and pain? They probably perceive themselves as a burden on the family or do not accept total dependence on others. These areas still remain unexplored.
Martin et al. [63] analyzed the physical, cognitive and psychological characteristics of 32 patients affected by ALS who had received the diagnosis at least 6 months previously and had made one or more decisions regarding NIV management and/or gastrostomy tube placement during the previous three months. They demonstrated that the main determinant of patients' physical complaints was related to the decisions made during the observational period [63]. Patients who believed they had a good understanding of the clinical course of their disease and had a proactive and participatory attitude towards decision-making, and those with fewer depressive symptoms, were more likely to refuse both interventions. Patients who refused NIV said that it undermined their sense of identity, dignity and/or autonomy. All these aspects are probably also closely related to the patient's age, the presence of a caregiver in the family unit, the individual's level of education or religious belief. ALS patients who had a more passive approach to interventions and fewer years of education could benefit from psychological assistance to make informed decisions compared to those who were better informed about the disease and had clear opinions about interventions [64].
The help of a close family member promotes the patient's acceptance of non-invasive support and specialized clinical interventions, producing positive effects on the patient care and survival. Although NIV increases caregiver burden and may negatively affect caregiver physical function (manifested by signs of exhaustion such as insomnia, anxiety disorders, or loss of attention), it does not significantly increase caregiver-related stress [64,65].

Doctor-patient interaction

Patients have sometimes reported that healthcare personnel considered ventilation as an imposition, an aggression on their sick person. Tolerance to the management of NIV improves when patients understand the therapeutic bond with their doctors and how these will be attentive to their emotional and physical needs, and when continuity of care is proposed and maintained by suggesting the best path to follow which respects life, the quality of life and, above all, the dignity of life [66-68].
However, further studies are needed to evaluate the optimal path that clinicians should promote to best help patients and their families cope with NIV. The presence of a caregiver during the course of the disease represents, in most cases, an element of positive support, especially when he/she also fully shares the treatment process. Sometimes, however, it is possible that it negatively influences the patient's ability to make choices.

Practical considerations

Patients with NMDs are extremely fragile people with considerable complexity due to the possible acute complications caused by the underlying disease, in which technical or technological knowledge alone does not allow a correct approach. The recently published practical approach to respiratory emergencies in neurological diseases can be useful for managing these situations [69]. The skills necessary for the best outcome should include knowledge of the right timing to start NIV and the potential clinical evolution of the different NMDs.
Patients should be offered major respiratory support options. Furthermore, and especially in the adaptation phase, alternative methods that potentially favor a better approach and greater adherence should be taken into consideration. The choice of the ideal interface remains a crucial point to promote adherence to treatment. Therapy failure may depend on the choice of an uncomfortable, incorrectly sized or poorly tolerated interface. Knowledge of the psychological dynamics that occur during the treatment of patients with NMDs is also necessary. The extent to which clinicians are compassionate, receptive, and cautious in their counselling about NIV can significantly influence the patient's perspective on the therapy. This perception is often associated with a generalised contempt for hospitals and a sense of threatened autonomy.
The doctor-patient relationship is one of the most complex human interactions as it puts two people who are strangers into physical contact and to share of personal information often in situations that involve varying degrees of physical suffering, anguish and anxiety.
Training a team to manage the patient on non-invasive mechanical ventilation is necessary for the initiation of NIV therapy and for the early identification of problems that could interfere with adherence and effectiveness of therapy. The team will have to make use of the support of various specialists who share the basic skills necessary to achieve the therapeutic objective both in the short term during patient monitoring and in the long term after discharge, and at home.

Conflict of interest statement

The authors declare no conflict of interest

Funding

None

Author contributions

Conceptualization, methodology, original draft writing, editing and supervision A.A.; investigation and data collection A.C, F.S., A.M., G.F., A.E, writing review A.A. and A.C, supervision G.F.

Ethical considerations

This research was conducted ethically, with all study being performed in accordance with requirements of the world medical association's declaration of Helsinki.

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