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ORIGINAL ARTICLE

Physiotherapy performance in motor and respiratory repercussions of patients with spinal muscular atrophy type I

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Muscular Atrophy, Spinal, Physical Therapy Modalities, Rehabilitation.

Abstract

Objectives: To verify in the literature the physiotherapeutic resources that have already been used in the motor and respiratory repercussions of patients with spinal muscular atrophy type I. **Methods:** This is a literature review carried out in the PubMed, LILACS, SciELO and PEDro databases, in period from June to August 2019, using the keywords "spinal muscular atrophy type I" OR "Werdnig-Hoffmann syndrome" AND "physiotherapy", as well as the terms in English. **Results:** 53 articles were found, but after the inclusion criteria, only 5 were relevant to the research. In the respiratory system, Cough Assist®, nasotracheal aspiration, acceleration of expiratory flow and postural drainage were used, both effective for bronchial hygiene. Pulmonary expansion and noninvasive ventilation improved ventilatory mechanics. In the musculoskeletal system, stretching, joint mobilization, kinesiotherapy, and use of orthoses provided gains in functionality and reduced deformities. **Conclusions:** Kinesiotherapy provided motor gains such as cervical control, stretching, mobilization and orthoses, helping to maintain functionality. In respiratory repercussions, the use of Cough Assist®, nasotracheal aspiration, acceleration of expiratory flow and postural drainage were able to reduce hospitalizations for pneumonia. The pulmonary expansion and noninvasive ventilation maneuvers reversed microatelectasis, improved hematosis and contributed to increased life expectancy.

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INTRODUCTION

Spinal Muscular Atrophy (SMA) is a neurodegenerative disease with an autosomal recessive genetic character that leads to the degeneration of alpha motor neurons located in the anterior horn of the spinal cord and brainstem[1,2]. There are two genes that codify the transcription of survival proteins of motor neurons (SMN), SMN1 and SMN2[1]. The main gene for synthesizing this protein is SMN1, but in SMA it is absent on chromosome 5q13. SMN2, in turn, is present and produces, in small amounts, the survival protein of motoneurons, determining the severity of the pathology, therefore, the higher the concentration of SMN2, the lower the functional repercussion[2].

There are four clinical variations of the pathology, modifying according to the onset of symptoms. SMA type I is known as Werdnig-Hoffmann Syndrome, it presents the first signs until 6 months of age, with absence of cervical control. They end up dying due to respiratory complications and bulbar dysfunctions. Type II starts around 6 months and manifests itself until 18 months, they can sit without support, but are not able to walk[3]. SMA type III or Kugelberg-Welander disease starts after 18 months of age, children acquire gait[4]. Finally, type IV is the latest type, its symptoms appear around the age of 20, it is the least aggressive compared to the other types[5].

The diagnosis is made through the clinical picture and complementary exams, such as electroneuromyography, muscle biopsy and genetic investigation[2,5-7].

The participation of Physiotherapy in the rehabilitation of these patients is fundamental, because it will act directly on the motor and respiratory repercussions resulting from the disease . Multidisciplinary care works in prevention, promotion and rehabilitation with the aim of improving the quality of life of patients and prolonging life expectancy[8-9].

The present study aimed to verify the physiotherapeutic resources used in the motor and respiratory repercussions of patients with Spinal Muscular Atrophy type I.

METHODS

A literature review was carried out in Pubmed (National Library of Medicine), LILACS (Latin American Literature in Health Sciences) , PEDro (Physiotherapy Evidence Database) and Scielo (Scientific Electronic Library Online) databases. The descriptors used were: Spinal Muscular Atrophy type I, Werdnig-Hoffmann Syndrome and Physiotherapy, also used in English and Spanish , relating them to the Boolean descriptors AND. The selected articles were published in the last 10 years to obtain the most up-to-date information.

The inclusion criteria defined were: articles published in the last ten years, in English, Portuguese and Spanish, study design consisting of clinical trials and case reports, which addressed the role of motor and respiratory physiotherapy, the effectiveness of the techniques and the population studied being patients with Spinal Muscular Atrophy type I.

Literature review articles on other neuromuscular diseases, those that did not report the effectiveness of physical therapy resources and studies in patients diagnosed with SMA type II, III and IV were excluded.

RESULTS

32 articles were found in PubMed, when reducing the search to articles with up to 10 years of publication, 22 articles were found. Those whose study design was clinical trials or case reports, 11 articles. These were submitted to the reading of abstracts and only 3 articles fit the central theme of study; the others addressed the role of Physiotherapy in patients with SMA type II and III or discussed the drug effects on the motor and respiratory system of children with SMA type I.

In the LILACS database, 3 articles were found, but one of them was published more than 10 years ago and the other addressed neuromuscular diseases in general, and 1 article was included.

In the Scielo electronic library, only 1 article was found and this was the same one selected from the LILACS database. PEDro did not have any studies related to the descriptors.

A free search was performed, finding 17 more articles, but only 1 article was related to the inclusion criteria.

Of the selected articles, 3 articles discussed Respiratory Physiotherapy, 1 associated motor and respiratory conducts and the last one, only about Motor Physiotherapy.

DISCUSSION

Due to the vast number of physiotherapeutic techniques existing in pediatrics, only the most cited, effective and frequent ones in this population of patients were studied, according to the selected articles.

The article by Lima[10] is a case report of a 2-year-old girl diagnosed with Werdnig-Hoffmann Disease. The objective of the study was to verify the performance of Physiotherapy and its benefits. The patient was hospitalized with Inspiratory Pressure of 23 cmH2O, Positive End-Expiratory Pressure of 6 cmH2O, respiratory rate of 30 irpm. The physiotherapeutic conducts were performed daily, twice a day, for a period of 8 months. Passive stretching,

joint and trunk mobilization, pelvic and scapular girdle dissociation, passive kinesiotherapy, weight bearing on upper and lower limbs, tibio-tarsal pumping, stimulation of non-acquired motor milestones, such as: cervical control, midline and sedestation; bandaging in 8, use of positioning orthosis, proprioceptive stimulation and orthostatism training. Positioning in bed was corrected and caregivers were instructed on changes in position. The bronchial hygiene techniques used were aspiration, clearing maneuvers and postural drainage. Pulmonary reexpansion therapy was performed by increasing PEEP to 10 cmH2O for 15 minutes. After 4 months, she obtained functional gains in the hip, being evaluated with grade 2 of the Modified Ashworth Scale. At 1 year and 10 months, he was already able to maintain the position of flexion of the hips and knees in a closed chain and sustained the contraction for 1 minute. The authors concluded that Physiotherapy helped in motor development, but there is still a lack of scientific studies that prove the effectiveness of physiotherapeutic treatment.

The article by Magalhães[11] is a case study of a boy with SMA type I who at 11 months progressed to respiratory failure, requiring orotracheal intubation. Connected to NIV, with nasal interface in Assisted Controlled mode, the parameters were: Positive Inspiratory Pressure of 20 cmH2O, Positive End-Expiratory Pressure of 6 cmH2O, Inspired Oxygen Fraction of 21% and RR of 18 ipm. The objective of the study was to publicize the importance of respiratory maintenance and Physiotherapy in the quality of life of patients with SMA type I. The techniques used for bronchial hygiene were the use of *Cough Assist®* to aid coughing, performing 5 cycles of insufflation and deflation mechanics with pressures of +40 cmH2O and -40 cmH2O, associated with acceleration of expiratory flow and nasotracheal aspiration. Lung reexpansion therapy was performed with elevation and traction of the upper limbs in conjunction with NIV, increasing PIP by 5 cmH2O for 10 minutes, followed by a 1 cmH2O decrease every 5 respiratory cycles. Finally, prone positioning and use of NIV during sleep with the parameters already mentioned. The daytime use of NIV was not mentioned in this article, as well as patient adherence to the device. The results were satisfactory, it was observed that the use of non-invasive auxiliary respiratory devices are effective in the care of the respiratory system, as well as the physiotherapeutic techniques used, preventing hospitalizations and complications of the respiratory system.

The study by Saquetto[12] was designed to report two cases of patients with Werdnig-Hoffmann Syndrome. Both were admitted to the Pediatric Intensive Care Unit of a public hospital. The objective was to investigate the effects and safety of functional mobilization aimed at gaining flexibility in these children who are chronically ventilated. The intervention lasted 2 months, the protocol was performed 5 times a week, lasting 30 minutes each session. Patient A, male, 5 years and 8 months old, had been hospitalized in the pediatric ICU for 2 months and was on invasive ventilatory support. Severe hypotonia was observed in the trunk, upper and lower limbs, with absence of muscle contractions, making active movement of the joints impossible. The flexibility of the elbows, knees and ankles was reduced. Patient B, male, 3 years and 6 months old, admitted to the pediatric ICU for 3 years, required invasive ventilatory support since his admission. In the initial evaluation, he presented slight contractions without active movement in the lower limbs, but the upper limbs had active movement in the elbows and wrists, but with reduced muscle strength. The range of motion was evaluated and flexibility restrictions were observed in the left elbow, knee and ankle joints. Both patients used Synchronized Intermittent Mandatory Ventilation mode. The functional mobilization protocol consisted of passive transfer from dorsal decubitus to sitting postures, carrying weight on the lower limbs, transfer from sitting posture to 4 support positions on the Swiss ball and evolution from 4 support positions to kneeling, semi-kneeling and orthostatism. Bronchial hygiene maneuvers were performed before the protocol and after the motor conducts, if necessary. Weight bearing on upper and lower limbs, as well as joint mobility, was recommended during postures. The authors observed that after the intervention period, the 2 patients had increased range of motion in all segments evaluated. Patient B, being younger, obtained greater gains compared to patient A, demonstrating that the earlier the intervention, the greater the gains obtained.

The article by Chatwin, Bush and Simonds[13] is a descriptive cohort study of 13 children diagnosed with Spinal Muscular Atrophy type I who used Non-Invasive Mechanical Ventilation with Positive Pressure at home for ventilatory support and improved respiratory mechanics. The aim of this study was to inform medical staff and parents about the options available for respiratory management of children with SMA type I. Patients were treated with NIPPV to correct nocturnal hypoventilation, improve/stabilize the paradoxical breathing pattern, and promote clearance of secretions present in the airways. In infants, pressures were low, with Positive Inspiratory Airway Pressure of 12 cmH_2O and Positive Expiratory Airway Pressure of 4 cmH_2O, with a backup of 18 to 35 breaths per minute. When the patient needed oxygen therapy to maintain SpO_2 greater than 95%, a cough machine (*Cough Assist®*) was used to remove secretions and maintain a stable condition. Families were instructed to remove secretions from the airways daily, ensuring that there was no accumulation of secretions and to avoid respiratory infections. Respiratory physiotherapy consisted of techniques such as tapping, nasopharyngeal

aspiration, assisted coughing with *Cough Assist®* at initial pressures of +30 to -30 cmH_2O in cases of patients with difficulties in expectoration due to marked weakness of the abdominal muscles. The authors concluded that NIPPV can be used to facilitate hospital discharge and can increase life expectancy, allowing effective treatment during respiratory tract infections. *Cough Assist®* also enabled these children to receive home care and, in some cases, prevented the need for intubation and invasive ventilation.

The article by Keating[14] is a case study of a 21-month-old female child with SMA type I, 21 months old, who frequently had tracheobronchial secretion accumulation despite physical therapy intervention and use of *Cough Assist®*. Therefore, the objective was to report the effects on the patient after using the high-frequency oscillator in the rib cage coupled with NIV. Pressure was transmitted externally through a plastic jacket that formed a seal around the trunk, generating negative pressure. This device has a thixotropic function on secretions through vibrations, in addition to coughing, which prolongs the inspiratory phase with high pressure, followed by a short expiratory period. The cough mode interrupts the vibration mode, according to the frequency set by the therapist. At 21 months of age, she was hospitalized with increased volume of secretions and respiratory distress. Dependent on NIV for breathing, PIP parameters: 18 cmH2O and PEEP: 8 cmH2O, required supplemental oxygen at 4 L/min. There was atelectasis in the right lower lobe and air bronchograms on X-ray, suggesting respiratory infection. Physiotherapy was performed 4 times a day, using *Cough Assist®* , manual percussion and increased ventilatory pressure. As there was no improvement, the high-frequency oscillator was introduced before standard Physiotherapy sessions. In total, there were 4 sessions per day, for 2 weeks and after 14 days, the patient was able to breathe alone for 5 consecutive hours. A chest X-ray showed resolution of the condition and she was sent home. The authors concluded that the study is limited, since the improvement in the clinical condition cannot be attributed to the device used, because several physical therapy techniques were being performed together, which contributed to the dehospitalization.

Therefore, Physiotherapy is of fundamental importance in the multiprofessional team of care for patients with Spinal Muscular Atrophy type I, acting directly on the respiratory and motor repercussions presented by them. Due to the rarity of the disease, there are physiotherapeutic techniques that have not yet been studied, making further scientific research necessary.

The respiratory system, as it is the most affected and collaborates with the reduction of life expectancy, is the main target of scientific studies. The techniques performed such as nasotracheal and orotracheal aspiration, acceleration of expiratory flow, postural drainage, manual percussion and equipment available on the market such as *Cough Assist®* and the high frequency oscillator, aim to remove tracheobronchial secretions, through modification of the mucus rheology, making expectoration easier, to reduce respiratory infections and the number of hospitalizations. Pulmonary re-expansion maneuvers and the use of Non-Invasive Mechanical Ventilation help to reverse microatelectasis, improve oxygenation, prevent respiratory muscle fatigue, contributing to increased life expectancy.

In the musculoskeletal system, kinesiotherapy provided motor gains such as cervical and thoracic control; stretching, mobilization and orthoses helped to maintain range of motion, maintaining functionality.

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