Early physical therapy in Freeman-Sheldon Syndrome: a case report

Fisioterapia precoce na Síndrome de Freeman-Sheldon: relato de caso

Fisioterapia temprana en el síndrome de Freeman-Sheldon: reporte de un caso

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ABSTRACT

Freeman-Sheldonn Syndrome (FSS) is a rare condition that presents as autosomal dominant due to mutations in the embryonic myosin heavy chain (MYH3). Individuals with FSS develop contractures that lead to motor and respiratory impairments and difficulties in eating. Physiotherapy can act in the prevention of skeletal deformities, muscle rebalance and in the improvement of respiratory function, thus aiming to promote more functionality for the patient. This study aimed to report a case of physiotherapy in the FSS. This is the first case report of an infant with FSS seen at a teaching clinic twice a week, lasting 1 hour each session, for a period of two consecutive months. The care protocol was built based on the assessment of the infant's primary and secondary disabilities, from which the functional goals (short, medium and long term) were pre-established and the activities developed aimed at functional improvement. The performance of physiotherapy in the FSS over a period of two months had a positive effect on improving cervical extension in prone posture and improving cervical alignment in supine posture. Physiotherapy plays an important role in the prevention and maintenance of functional aspects, improving the patient's health condition, from increasing functionality and reducing disabilities.

Keywords: Freeman-Sheldon Syndrome, physiotherapy, precocious, quality of life.

RESUMO
Síndrome de Freeman-Sheldon (FSS) é uma condição rara que se apresenta como autossômica dominante devido a mutações na cadeia pesada de miosina embrionária (MYH3). Indivíduos com FSS desenvolvem contraturas que levam a deficiências motoras e respiratórias e dificuldades alimentares. A fisioterapia pode atuar na prevenção de deformidades esqueléticas, reequilíbrio muscular e na melhoria da função respiratória, visando assim promover mais funcionalidade para o paciente. Este estudo teve como objetivo relatar um caso de fisioterapia na FSS. Este é o primeiro relato de caso de um bebê com SFS visto em uma clínica de ensino duas vezes por semana, com duração de 1 hora em cada sessão, por um período de dois meses consecutivos. O protocolo de atendimento foi construído com base na avaliação das deficiências primárias e secundárias do lactente, a partir das quais os objetivos funcionais (curto, médio e longo prazo) foram pré-estabelecidos e as atividades desenvolvidas visando a melhoria funcional. A realização de fisioterapia na FSS ao longo de um período de dois meses teve um efeito positivo na melhoria da extensão cervical na postura prona e melhoria do alinhamento cervical na postura supina. A fisioterapia desempenha um papel importante na prevenção e manutenção dos aspectos funcionais, melhorando o estado de saúde do paciente, aumentando a funcionalidade e reduzindo a deficiência.

Keywords: Síndrome de Freeman-Sheldon, fisioterapia, precoce, qualidade de vida.

RESUMEN
El síndrome de Freeman-Sheldonn (SFS) es una afección poco frecuente que se presenta como autosómica dominante debido a mutaciones en la cadena pesada de la miosina embrionaria (MYH3). Las personas con SFS desarrollan contracturas que conducen a deficiencias motoras y respiratorias y dificultades para comer. La fisioterapia puede actuar en la prevención de deformidades esqueléticas, reequilibrio muscular y en la mejora de la función respiratoria, con el objetivo de promover más funcionalidad para el paciente. Este estudio tuvo como objetivo reportar un caso de fisioterapia en el SFS. Este es el primer informe de caso de un bebé con SFS visto en una clínica de enseñanza dos veces por semana, que dura 1 hora cada sesión, durante un período de dos meses consecutivos. El protocolo de atención se construyó a partir de la evaluación de las discapacidades primarias y secundarias del lactante, a partir de las cuales se preestablecieron los objetivos funcionales (corto, mediano y largo plazo) y se desarrollaron las actividades destinadas a la mejora funcional. La realización de fisioterapia en el SFS durante un periodo de dos meses tuvo un efecto positivo en la mejora de la extensión cervical en la postura propensa y en la mejora de la alineación cervical en la postura supina. La fisioterapia juega un papel importante en la prevención y mantenimiento de los aspectos funcionales, mejorando el estado de salud del paciente, desde el aumento de la funcionalidad y la reducción de las discapacidades.

Palabras clave: Síndrome de Freeman-Sheldon, fisioterapia, precoz, calidad de vida.

1 INTRODUCTION

Freeman-Sheldon Syndrome (FSS) is a rare disease, with an unknown prevalence [1] . It is also known as whistling face syndrome, craniocarpotarsal dysplasia syndrome, craniocarpotarsal dystrophy, and Type 2A distal arthrogryposis [2]. It is caused by a mutation in the embryonic heavy chain myosin, more precisely in the MYH3 gene that is present on the

short arm of chromosome 17 [3]. The MYH3 protein plays an important role in the production of skeletal muscle cells and, when altered, impairs the myophysiology of early development, thus causing residual defects and muscle contractures [4].

The clinical diagnosis of FFS is based on the physical characteristics present, such as: craniofacial - whistling of the face, microstomia, presence of supraorbital edema, prominence of the supraciliary arches, sunken eyes, narrow and high palate, hypoplasia of the nose, low-set ears, prominent nasolabial folds and H- or V-shaped chin dimple-, malformations - camptodactyly with ulnar deviation of fingers and bilateral clubfeet - or other changes that may be present, such as scoliosis, joint stiffness, thumb contracture on adduction, spina bifida occulta and congenital hip dysplasia or dislocation [3; 4; 5]. Molecular tests can be performed on FSS, however they are not conclusive for diagnosis [4].

As it is a syndrome that affects the craniofacial region and is usually accompanied by orthopedic problems, due to musculoskeletal malformations, FSS leads to a decrease in functionality and physical dependence in patients, especially for walking, requiring a lot of assistance from the caregiver/family member. Disability generates a negative impact on the patient's life, who often presents psychosocial difficulties and develops depression, anxiety and low self-esteem[1].

Treatment for FSS is not yet specific. In most cases, it is aimed at improving the patient's functionality and reducing the impact of skeletal deformities, changes in respiratory function, and feeding difficulties [6]. In the presence of some physical alterations, surgical interventions are necessary to provide a better quality of life for these patients [2].

In view of the characteristics of the disease, physical therapy can act in FSS early in childhood, minimizing delay in neuropsychomotor development, improving limb function, preventing deformities and contractures, rebalancing the muscles, and improving respiratory function.[6; 7]. To date, few studies on FSS have been identified, and none of these reports the role of physical therapy in infants with FSS.Therefore, this study aimed to report a case of physiotherapy performance in FSS.

2 BRIEF CASE REPORT

This is a case report of a patient diagnosed with Freeman–Sheldon Syndrome, admitted to the Clinical School of Physiotherapy at the Federal University of Sergipe – Lagarto, in November 2018. The mother was invited and granted her child's participation in the research, signing the consent form. This research was submitted to the ethics and research committee of the Federal University of Sergipe and was approved with CAEE: 62995022.4.0000.0217.

The physiotherapeutic evaluation was performed based on a standard pediatric form adopted at the clinic, which contained identification data, clinical diagnosis provided by the patient's own physician, clinical history, functional evaluation, and physical examination - ADLs, postures and active movement, coordination sensorimotor, tone, primitive reflexes (present or persistent), range of motion (passive vs. active), muscle strength, other systems (sensory, vision, hearing, cognition, language), use of auxiliary equipment, social participation/limitation and contextual factors (barriers vs. facilitators).

The patient was 11 months old on the evaluation day, male, and a resident of the city of Boquim-SE. The mother's main complaint was that the patient “has a closed hand, his mouth is whistling, he's all tough, he never cried and doesn't complain about anything”. In the history of the current illness, the prenatal, delivery, and postnatal period, it was reported to have had an uneventful pregnancy until the moment of birth, in which the newborn presented dyspnea and needed to be removed for surgery. intensive care unit (ICN). After 15 days in the ICN, he was discharged from the hospital, as his condition improved. The infant was being followed up by a multidisciplinary team composed of a physical therapist, an occupational therapist, a speech therapist, and a neurologist. The family's desire and expectation regarding physical therapy was that one day the infant would be able to walk.

3 THERAPEUTIC INTERVENTION

Functional assessment was performed on the first day of treatment by two trained physiotherapy trainees. Functional changes and changes in body structures and functions were identified, as detailed in Table 1. From the second appointment, specific procedures were carried out for these changes. Physiotherapy sessions were performed twice a week, lasting 1 hour each, during a period of two consecutive months (November to December 2018). The consultations were supervised by a physical therapist and a teacher responsible for the sector.

Table 1. Treatment plan including secondary, primary and objective deficiencies

Secondary deficiencies (how you perform the task)	Primary disabilities (body structure sand functions)	Objetives
Patient hás difficulty keeping the head in the supine midline	Weakness in head and neck extensor muscles	- Perform cervical extension to track objects; - Roll from supine to prono following stimuli; - Set with head control for midline; - Carry out weight bearing for the lower limbs in a standing position with support from the therapist
Performs the extent of the cervical to look at objects in front of it when the prono position is passively only	Compactodactilia and muscle stiffness that limits ADM	**Immediate objective**
It has difficulty holding objects and performing range	Does not have muscle control due to stiffness and base disease	- Perform cervical extension for 5 seconds; - React to the stimulus to roll
Performs the rolling with total dependence	Has scoliosis in “C” to the left	**Medium term objective**
Sit with medium trunk support and adopt inclined posture due to scoliosis	Muscle weakness of MMII and trunk	- Perform cervical extension for 30 seconds; - perform the lateral decubitus to assisted; - Sit with support on a lower trunk;
Stand with support in medium trunk and total dependence, make side flexion to the left of trunk, flex the hip and knee and do not discharge MMI weight discharge	Typical face of SFS	**Long -term objective** - Make cervical extension for 1 minute; - perform the lateral decubitus to prono independently; - Sit without support for 5 seconds; - Stand for 10 seconds with trunk support and weight discharge in MMII
Does not present facial expressions		

Fonte: Autoral

The proposed treatment, following the functional objectives, consisted of activities such a: passive stretching of the upper trapezius muscles; a stimulus for manual reach, in supine, from the presentation of toys in the midline and shoulder height; a stimulus for rolling, from the facilitation of movement by the pelvis or shoulder girdle on the opposite side when rolling; stimulation for cervical extension in the prone position, from the presentation of toys in the midline, on the platform and in horizontal and vertical movements for visual tracking, and with the therapist's facilitation through tactile stimuli on the forehead; stimulus for trunk and cervical control in the sitting position, with support from the therapist and bandage (Neuro Nustim Fabrifoam® neoprene bands) on the trunk and tactile stimuli for contraction of trunk and cervical extensor muscles; and stimulation in the standing position, performing weight bearing on the lower limbs, as shown in Figure 1.

Figure 1. Patient with Freeman-Sheldon Syndrome, during the physical therapy intervention, illustrating the following activities: a) stretching of the trapezius muscle; b) stimulus for manual reach; c) stimulus to roll to both sides; d) stimulation of cervical extension with the patient in the prone position; e) stimulus for trunk and neck control with the infant sitting on the therapist's leg; f) stimulation of lower limb extension and weight bearing, in the standing posture.

Fonte: Autoral

4 RESULTS AND DISCUSSION

After two months of physical therapy intervention in the patient with FSS, an improvement was observed in the postural pattern and in the cervical and trunk control of the infant in the supine, prone, sitting and standing postures. This evolution was evidenced by the acquisition of some of the proposed objectives.

The motor development of an infant is characterized by the acquisition of skills, which involve several areas (motor, sensory, cognitive, behavioral, and language), related to chronological age [8]. It is expected that, at 11 months of age, the infant has head control in all positions, sits without support, can grasp and reach objects, can roll from side to side and from supine to prone and from prone to supine, and stands with hand support [9]. In the present report, the infant was 11 months old and had a neuropsychomotor developmental delay (NDD) in relation to head and trunk control, language delay, and inability to roll between postures, reach and hold objects, and stand without support. of the therapist. Delay in motor development is expected in infants and children with FSS [3].

Early stimulation aims to accompany, monitor, and intervene in the child development of babies at risk or with developmental delays, by multiprofessional teams, composed of speech therapists, physiotherapists, occupational therapists, and psychologists [10]. In this team, physical therapy has shown promising results [11; 12]. In the study by Marques [13], they addressed early physiotherapy in a two-month-old infant who had characteristics compatible with SFS. Cunha [14] reported a child aged twenty-three months who presented the same face and skull aspects present in SFS. In the present report, the patient underwent early stimulation. According to the mother's report, after starting physical therapy, it can be observed that the infant began to emit sounds, such as crying, frequently - a case that was not seen before treatment -, demonstrating an evolution in the ability to communicate and react to stimuli environmental. This fact may be positively related to the stimuli offered in physical therapy. It is known that developmental neuroplasticity is a complex genetically encoded, time-dependent, maturational process that is closely regulated by intrinsic homeostatic mechanisms and is influenced by extrinsic environmental factors [15].

The experiences and stimuli that are offered to individuals should be intensified during “sensitive periods”, that is, periods in which there are plasticity peaks, which is when neuronal tissue is subject to change [16]. These changes occur in prenatal and postnatal brain development and continue, albeit to a lesser extent, throughout adolescence and adulthood [17]. Studies involving abnormal developmental plasticity in genetic disorders in some syndromes such as Rett syndrome [18], fragile X [19] and Neurofibromatosis [20] have already been reported. In most

neurological disorders in children, the normal developmental homeostasis of neuroplasticity is affected, which contributes to neurophysiological problems and behavioral phenotype [15]. In this context, the earlier the stimulus through interventions in babies with neurological problems, the greater the possibilities for changes.

In the care protocol of the present report, postural control was emphasized through the infant's prone, supine, sitting, and standing postures, which corroborates the study by [21], in which a physical therapy program was developed to improve motor development through postural control in prone, supine and sitting postures in preterm babies. The improvement in postural control, in the present study, was observed when measuring the time that the infant could maintain the cervical extension in the prone position and the maintenance of the head in the midline in the supine position. An increase in sustain times was observed from 0 to 17 seconds in prone and from 0 to 12 seconds in bench press. These gains may be related to the stimuli that were offered to the infant in the two positions during the physical therapy sessions. Research involving physical therapy in early stimulation of syndromes in short periods of treatment is still scarce.

The etiology of FSS has already been reported as a mutation in the gene responsible for encoding a protein that participates in the contractile physiology of the muscle, which may result in changes in muscle physiology, including muscle weakness, alteration in muscle tone, and development of fibrosis [22]. In this context, the role of physiotherapy as early as possible in the life of this patient becomes important, since it can act in muscle strengthening and in the prevention of contractures, thus avoiding the formation of fibrosis and deformities [15]. McCormick, Poling, Portillo and Chamberlain [23] reported the experience with therapy in multiple hand and wrist contractures in an adult FSS patient without surgical intervention and observed a moderate improvement in functional correction for movements involving the hand.

In the present report, the infant already had shortening of the cervical muscles on the left and upper trapezius muscle, weakness of the cervical extensor muscles, trunk, and lower limbs, compact dactyly in the fingers, and scoliosis in "C" to the left. Therefore, conducts were carried out to improve the flexibility and strength of the cervical, trunk, upper, and lower limb muscles, thus preventing the installation of muscle contractures.

Improvements in neck musculature flexibility and neck extensor musculature strength were observed in the infant, as previously shown in increases in infant head support times, both for midline permanence and for neck extension.

5 CONCLUSION

This study reported the experience of physical therapy care in a patient with SFS, showing positive effects. Physiotherapy as early as possible can provide an opportunity for the infant with FSS to acquire a better quality postural and motor pattern, avoiding atypical movement patterns and promoting greater functional independence, and, consequently, better health conditions. It is desirable that more cases of physical therapy interventions in infants with FSS are reported, to prove their benefits in the short and long term.

DISCLOSURE OF INTEREST

The author declares that there are no conflicts of interest associated with this manuscript, The patient's family has provided permission to publish their reports, including the patient's image and identity has been protected

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