

Artigo de Relato de Experiência

Pediatric physical therapy intervention in leigh's syndrome: a clinical case report

Intervenção fisioterapêutica pediátrica na Síndrome de Leigh: relato de caso



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Keywords: Leigh Disease; Motor Skills Disorders; Child; Physical Therapy, Motor Development.

ABSTRACT

Objective: The aim of this single case study was to describe a physical therapy intervention in a 5-year-old boy with Leigh Syndrome (LS). **Method:** The balance was tested through the Pediatric Balance Scale (PBS) and the function in daily life activities was measured with the Pediatric evaluation of disability inventory (PEDI). Furthermore, the plantar pressure distribution (PPD) was measured during a self-selected speed, walking on a capacitive sensing platform. The intervention was based on the use of kinesiotherapy, sensory and proprioceptive resources, in 50-minute sessions, twice a week, for 10 weeks. **Results:** The present study demonstrated gains for the child in relation to balance during gait. The same way, the improvement of postural control, task planning and other skills was observed. **Conclusion:** The physical therapy intervention in this case report for Leigh Syndrome was effective and could serve as a basis for forthcoming studies or clinical therapy intervention that follow this kind of treatment.

RESUMO

Objetivo: o objetivo deste estudo de caso foi descrever uma intervenção fisioterapêutica em um menino de 5 anos com Síndrome de Leigh (LS). **Método:** o equilíbrio foi testado por meio da Escala de Equilíbrio Pediátrico (PBS) e a função nas atividades da vida diária foi medida com a avaliação Pediátrica do Inventário de Incapacidade (PEDI). Além disso, a distribuição da pressão plantar (PPD) foi medida durante uma velocidade auto selecionada, caminhando em uma plataforma de detecção capacitiva. A intervenção baseou-se no uso de cinesioterapia, recursos sensoriais e proprioceptivos, em sessões de 50 minutos, duas vezes por semana, durante 10 semanas. **Resultados:** O presente estudo demonstrou ganhos para a criança em relação ao equilíbrio durante a marcha. Da mesma forma, foi observada a melhora do controle postural, planejamento de tarefas e outras habilidades. **Conclusão:** A intervenção fisioterapêutica neste relato de caso para a Síndrome de Leigh foi eficaz e poderia servir de base para estudos futuros ou intervenção em terapia clínica que segue esse tipo de tratamento.

Palavras-chave: Doença de Leigh; Alterações das habilidades motoras; Criança; Fisioterapia, Desenvolvimento Motor.

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INTRODUCTION

Described by Denis Archibald Leigh in 1951, the Leigh Syndrome (LS), also known as

subacute necrotizing encephalopathy, is defined as a neurodegenerative disease with variable symptoms, caused by genetic and hereditary mitochondrial dysfunction accompanied by bilateral damage in the central nervous system (CNS).^{1,3} The syndrome typically affects children in the first year of life, with rapid deterioration. In most cases, the child does not present dysmorphic characteristics at birth.^{1,3}

The diagnosis involves the presentation of clinical signs, which may include: hypotonia, delayed psychomotor development, ataxia, spasticity, dystonia, convulsions, abnormal eye movements such as nystagmus, and respiratory impairment.^{1,4} In addition, image exams may present damage in the basal ganglia and brainstem, and laboratory alterations with probable lactic acidemia and increased levels of lactate and pyruvate in the blood.^{1,5}

Due to early mortality, which is around three years of life, and the low incidence of LS, estimated at 1:40,000 live births, the performance of controlled studies with a large sample becomes infeasible.^{1,3,6}

It is recognized that the physical therapy minimizes or prevents health complications, aiming at early intervention in the neuromuscular disorders, as well as in the treatment of clinical signs such as hypotonia, spasticity, and delays in motor development.⁷ (2 The principles of rehabilitation for most neuromuscular disorders are similar, however, because there is no known progress and little information in the literature, a study that addresses the physiotherapeutic intervention in LS is considered valuable in the assistance of the presented clinical signs. Therefore, the objective of this single case study was to describe the effects of a ten-week physical therapy intervention for a 5-year-old boy with LS. After the evaluations were performed, was elaborate a program combined balance training, endurance and strength though daily life activities.

CASE DESCRIPTION

This single case study was conducted at the Physical Therapy Clinic (PTC), which was approved by the Ethics and Research Committee on Humans under the standpoint CAAE: 07739919.6.0000.0118.

Subject

The subject M. S. is a boy whose non-consanguineous parents reported cesarean delivery with a gestational age of 39.4 weeks after preeclampsia was diagnosed, but the newborn was not impaired. The Apgar score was 9 in the 1st and 5th minutes, birth weight of 2270 g, length of 47.5 cm and a head circumference of 33 cm.

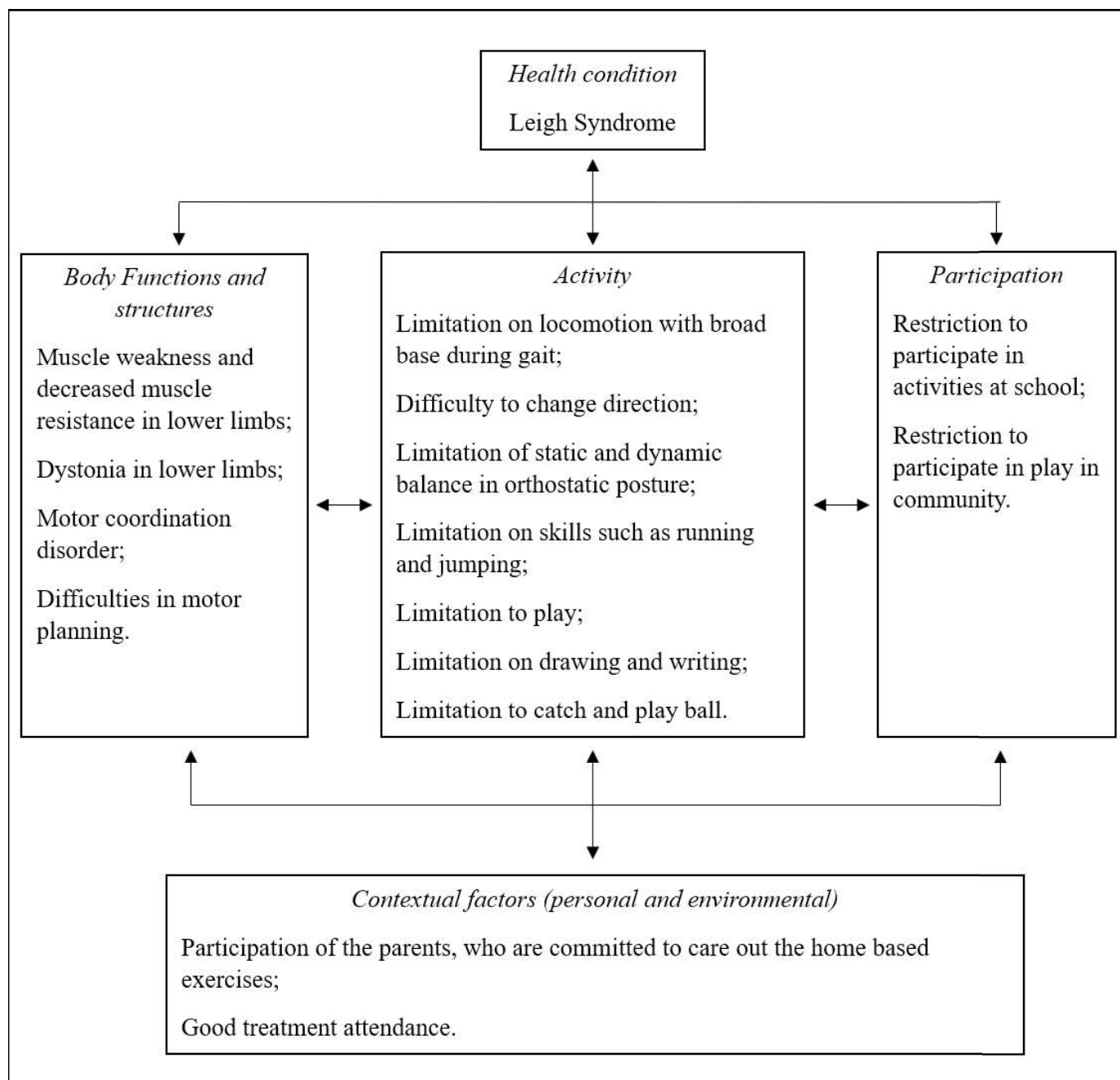
Since M.S. was two years old, he had performed neurological clinical follow-up due to neurodevelopmental delays. At 3 years old, after many episodes of lower limbs fatigue and involuntary gesture with right leg, magnetic resonance imaging was performed. Bilateral and symmetric pallidal involvements were observed due to lesions with restriction to the diffusion of water molecules, as a result of lactate peak measured through spectroscopy. An exome sequencing (variant ChrX19.373.591 A>G) confirmed the diagnosis of Leigh Syndrome, associated with deficiency of pyruvate dehydrogenase. Since then, episodes strength loss from upper limbs and trunk had been observed by the parents, mainly in situations of effort. These episodes lasted from 15 minutes to 1 hour and occurring more than 3 times a day.

Data Collection

Instruments

At the initial physical therapy evaluation, MS was having episodes of fatigue in right limb every day, weakness and dystonia on both lower limbs, motor coordination disorder, difficulties in motor planning and preserved range of motion (Figure 1). Furthermore, he walked independently with a broad base, difficulties in changing directions and also presented difficulties to run and jump. He had difficulty in carrying out daily activities, as described in Figure 1.

Figure 1. Context of the components of the International Classification of Functioning Disability and Health (ICF) related to the case reported in Leigh Syndrome.



After the first evaluation, assessment instruments were selected and applied according to the need verified. The balance was tested using the Pediatric Balance Scale (PBS), the functionality in the daily life activities was measured with the Pediatric Evaluation of Disability Inventory (PEDI) and the plantar pressure distribution during walking was registered on a capacitive sensing platform.

PBS is an instrument used in children from 5 to 15 years and measure the body balance and its functional capacity.^{8,9} The scale is composed of 14 items in 3 domains of sitting, standing, and

posture change, and is capable of measuring functional balance without the need for specialized equipment. Scores in the range of 0–4 points are allocated to each of the items in accordance with the extent of independent execution, with the best among 3 scores from repetition of each item 3 times being chosen. If the subject receives the highest possible score of 4 points in the first trial, no further assessment is made. The total score is 56 points, being that, the higher the score, the better the child's rating.^{8,9} PEDI aims to evaluate the functional abilities and typical performance

of children with functional limitations. The scale consists of an interview with the caregivers or through observation of the therapist. It reports three aspects of functional development: Part I: Functional Skills – including 59 dichotomous (zero if the child is unable to perform the activity and score one if he/she is capable) items grouped in 13 clusters: chair/toilet, car, and tub transfers, bed mobility, speed and method of indoor/outdoor locomotion, and stair mobility; Part II: The Caregiver Assistance Scale of the Mobility domain - including⁸ functional activities that are similar to tasks in the Functional Skills section: transfers, bed mobility, locomotion, and stair mobility (scores are defined as: independent (five), supervision (four), minimum (three), moderate (two) and maximum (one)); Part III: Modifications (the score is categorical: no modification (N), modifications related to child (C), modifications related to rehabilitation (R) and extensive modifications (E)).^{10,11}

The evaluation of the plantar pressure distribution (PPD) was performed during walking on a capacitive system platform (EMED AT®, Novel, Germany) with frequency of acquisition of 50 Hz and a resolution of 4 sensors per cm² (2,736 sensors). The evaluation of the PPD was performed in an 8 m catwalk, on a flat and straight walkway, with the child barefoot walking on a self-selected speed. Three valid records were made for each foot in the dynamic interaction of the foot and ground contact. For extraction of the PPD data, the Novel Database Pro M® (Novel, Germany) was used. The PPD variables selected were the Contact Area (which represents the total contact area of the foot with the ground), the Peak of Plantar Pressure (or the maximum pressure registered in the areas of measurement of the plantar surface) and the Integral force-time (or cumulative force produced within a period of time). It is often expressed as the area of force below the force vs. time curve.

Intervention

Based on the physical therapy assessment, goals were set to improve dynamic balance, daily life activities and lower limb-function through strengthening.

The physical therapy intervention was based on the use of kinesiotherapy, sensory and proprioceptive resources, in 50-minute sessions, twice a week, for 10 consecutive weeks.

Kinesiotherapy consisted of 15 minutes of muscle strengthening for lower limbs with adapted to daily life activities (as: sit-stand in a bench, up-down in stairs, running, playing soccer, jumping); 2 minutes of lower-limb stretching; 15 minutes of dynamic balance training in playful activities (MS had to walk, to jump obstacles on grass, benches and wedge; furthermore, he had to walk in line, to kick, throw and catch a ball, to jump on one foot each time); 3 minutes of slow and fast gait training with/without obstacles on different types of ground; 15 minutes of control training and multiple-activity planning (MS had to sit on the floor and stand up, staying on his knees and half-kneeling; he had to practice standing and squatting, taking an object from the floor and placing it in a shelf; MS had to practice some manual tasks on his knees and half-kneeling). For the intervention, stairs, mats, benches, wedge, rollers, Swiss ball, grass, tapes for demarcation lines, ball and toys were used.

During the treatment, the parents participated actively in physical therapy, where they observed and assisted in some exercises for perform at home; they had to do the activities at home and encourage the child to perform the daily life activities independently, according to the orientation of the physiotherapist, with the aim of improving performance and independence in activities that the child had difficulty.

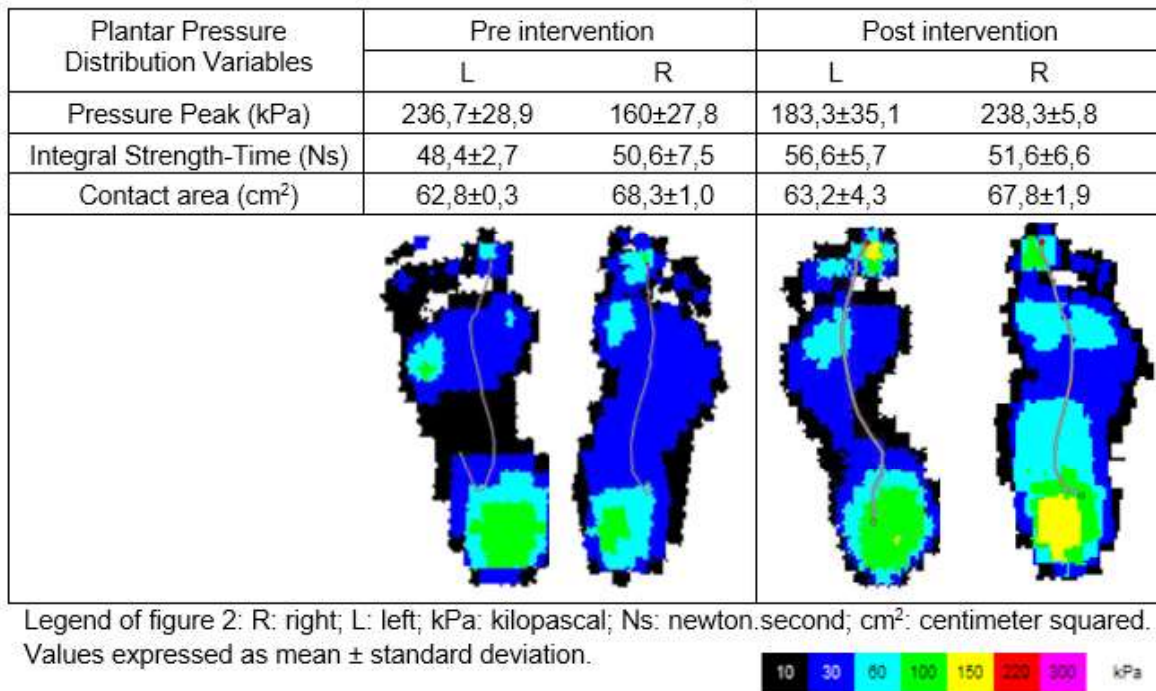
RESULTS

According PBS post-intervention evaluation, MS had improvement of one point in the score of the following items: transfers (from 3 to 4), standing without support (from 3 to 4), standing with unipodal support (from 0 to 1), rotating 360 degrees (from 0 to 1); two points in standing with eyes closed (from 2 to 4); and three points in standing with feet together (from 0 to 3), according described in the instruments. The PBS total score was 34 (before physical therapy intervention) to 43 points (after physical therapy intervention).

The pre and post-intervention scores of PEDI are shown in Table 1.

The data from PPD variables are presented in Figure 2. The self-selected pre and post treatment speed were, respectively, 4.7 (± 0.5) and 5.6 (± 0.2) km/h.

Figure 2. Representative plantigrames and variables of the Plantar Pressure Distribution (kPa) and Pressure Center Trajectory (CoP). Pictures are the means of the three attempts made by the child, pre and post physical therapy intervention for the L and R feet and self-selected speed.



After 10 weeks of treatment, totaling 22 physical therapy appointments (1 evaluation, 20 treatment sessions, 1 reevaluation), it was verified in relation to the motor behavior that M.S. improved the transfers (sitting to kneeling and orthostatic posture, four supports to standing) and acquired movements, as squats, jumps, gait pattern, running. Better performance was also observed during standing and dynamic balance, activities of jumping without space displacement and jumping through obstacles, running without falling, planning and controlling activities guided by the physical therapist, handling objects together

to walk without imbalances, as it was observed by the physical therapist during the reevaluation and through the parent's report.

The child presented three episodes of muscle fatigue in the right lower limb during the 20 sessions, which were observed through behavior and child reporting; passive stretching was then applied and the PT sessions were interrupted. However, the results obtained were positive (Table 1); there were improvements when compared to pre and post intervention in relation to balance and functional capacity.

Table 1. Pediatric Evaluation of Disability Inventory (PEDI) Score Scale Pre and Post Physiotherapeutic Intervention in Leigh Syndrome.

Area		Pre intervention	Post intervention
		Gross Score*	Gross Score*
Self care	Functional Skills	55	64
	Caregiver assistance	28	36
Mobility	Functional Skills	41	47
	Caregiver assistance	20	30
Social	Functional Skills	48	51
Function	Caregiver assistance	22	21

DISCUSSION

The physical therapy intervention was beneficial to the patient. Although the principles of rehabilitation for most childhood neuromuscular disorders are similar, LS has been poorly addressed in scientific studies, probably because of the low incidence of cases. In addition, low life expectancy makes the performance of studies difficult.¹² Investigations are fundamental in showing the improvement of the health condition of these children.

Hypotonia and/or dystonia in LS impairs static and dynamic balance, affecting daily life activities, as well as activities in the community.^{1,2} In a positive way, the present study observed, in the described case, an improvement of the dynamic body balance through the PBS. The decrease in the body weight discharge in the right lower limb, due to more frequent fatigue in this limb and altered muscle strength, generated changes in gait and in high postures and, consequently, in static and dynamic balance.¹² Despite the lack of literature regarding treatment protocols in LS, exercises and multiple task activities were proposed, with the focus being on muscle strengthening adapted to daily life activities, stretching, balance training and reactions, according to the scientific evidence described.^{13,14}

LS is responsible for affecting motor skills and negatively interfering with patients' cognitive function.^{1,2} The patient described to have difficulties for scheduling sequential tasks often required verbal feedback until the task was completed and without assistance. According to the literature, the LS patient may have an impairment in performing tasks and skills acquisition, because as described, the disease affects central nervous system structures. In summation, the individual may present cognitive alterations, which promotes issues for task-planning, including motor alterations, such as ataxia and dystonia, which interfere with movement.^{1,2} According to the PEDI reevaluation, the patient presented improvement in the areas related to self-care, mobility and social function, as well as the reduction of adult care in self-care and mobility tasks.

Skills such as running and jumping, in which the patient presented difficulties, demonstrate the delay of motor development, at the time of the evaluation. Due to this delay in motor development, the objectives of the intervention were traced,

according to principles in the literature where the rehabilitation team should seek to minimize or prevent musculoskeletal complications aiming to improve the motor condition. Thus, the patient presented gains in the jumping skills without space displacement and jumping through obstacles and running without falling.⁷

In addition, changes in plantar pressure distribution variables were observed in both lower limbs. In relation to the right lower limb, in which the child presented fatigue, there was an increase in peak pressure after physical therapy assistance, as well as a better distribution of pressure in all regions of the foot, reflecting a better body weight discharge in this limb.¹⁵ The change in plantar distribution reflects the maturation of motor skills, as well as better balance and better body adjustments, demonstrating the effectiveness of physical therapy assistance.^{16,17} The self-selected speed also increased in the pre and post-intervention comparison, which demonstrates more confidence in performing independent gait ability.

It was essential that rehabilitation was centered on a family participation and environment-focused approach. The M.S. parents were on all physiotherapy sessions to stimulate the child and learn how to do the exercise at home. Furthermore, the physical therapist provided knowledge on how parents could facilitate their child's participation at home and in the community. These was important to the success of treatment as shown in other study.¹⁸

The child received multi-professional treatment; however, the professionals did not have contact with each other, which is a limitation of the present study. Within the possibilities of treatment for LS, it is necessary to emphasize that the patient needs a multi-professional approach as well as the commitment of the parents to perform the home based exercises.¹ In addition to multi-professional care that addresses all aspects involved in LS, it is recommended that the care should be provided continuously and without interruptions, due to the low life expectancy of these patients and the unfavorable prognosis.¹

It is known that physical therapy proves to be effective and essential in the approach of patients with LS who suffer motor disorders.^{1,19} However, besides the few studies in the literature about LS, this case is atypical for this condition, which makes it more difficult to discuss and literature

that supports an adequate treatment. Despite this, the present study demonstrated the benefits of physical therapy intervention in children with LS in relation to dynamic balance during gait, as well as improvement of postural control and task planning and the acquisition of other skills.

CONCLUSION

Therefore, it is concluded that the physical therapy intervention in this single case report was effective and could serve as a basis for future studies and clinical approach for LS.

Conflict of interest statement: The authors declare no conflict of interest.

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