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Effect of a neurodevelopmental treatment based on physical therapy protocol for infant with Russel-Silver syndrome: A case report

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ABSTRACT

The Russell-Silver syndrome is a rare genetic disease, with various clinical presentations. Little is known about its developmental delay and methods of treatment in the literature. The aim of this study was to evaluate the effect of physical therapy to treat a child with developmental delay caused by Russell-Silver syndrome. The intervention was performed over a period of one year, during 1-hour, twice a week, at the University Hospital of the Federal University of Sergipe (UFS). Motor performance and functional capacity were assessed by using the Inventory of the Pediatric Evaluation of Disability (PEDI). Data were analyzed quantitatively, comparing pre-and post-treatment measures. After the intervention, there was a significant increase in scores for analysis in the areas of self-care and social function. In the area of mobility, the scores also increased, showing an improvement of the engine and consequently higher degree of functionality of the child, but the infant kept showing a lower level of development when compared with typically developing children at the same age. Physical therapy was an efficacious method of intervention, enhancing and acelerating the motor and functional development in a infant with Russell-Silver syndrome.

Introduction

The Russell-Silver syndrome (RSS) is described as an intrauterine and postnatal features, whose lateral asymmetry is associated with dysmorphisms as prominent forehead and triangular face, clinodactyly of the fifth finger and low set ears. Besides these characteristics, dental malformation, hypoplastic mandible, precocious sexual development, underdeveloped muscles, discrepancy of upper and lower limbs, and disturbance of weight and height are present in RSS[1,2,3,4,5,6,7,8].

Historically, Henry Silver observed the first characteristics of RSS in 1953, which included intrauterine growth retardation, short small for gestational age (SGA), body asymmetry, high levels of urinary gonadotropins and normal motor development. In parallel, Russell noted other characteristics, such as both pre and postnatal growth retardation, clinodactyly in fifth finger, triangular face, prominent forehead and mandibular hypoplasia in 1954. Initially, it was not clear that Silver and Russel studies were considering and describing the characteristics of the same disease. However, in 1961, there was a consensus that, despite some differences, the descriptions by Silver and Russell had common evidences for RSS[4,6,7].

Its etiology is still unknown, and the most cases are attributed to genetic mutations[6]. Incidence occurs in about 1:50,000 to 1:100,000 live births and children present severe intrauterine growth retardation. Some cases of this syndrome are not recognized because the features are presented so insignificant, and these children are considered as having nonsyndromic short stature only[1,4,6,7].

A validated method for obtaining the diagnosis is unknown. It depends of the investigator's experience associated to detailed physical examination. According to Azcona and Stanhope[2] and Beserra and Guimarães [7], the diagnosis can be established when the child presents, at least, three of the following characteristics: 1) small newborn for gestational age (e.g., weight and/or length at the birth with two or more standard deviations below gestational age average); 2) short stature at the time of diagnosis; 3) characteristic facial patterns (e.g., small triangular face, prominent forehead, low implantation of the ears, small jaw, thin lips with down-turned corners; 4) clinodactyly of the fifth finger; 5) body asymmetry[7].

Some features should be observed in the first years of life and taken into consideration for the diagnosis. The low weight is due to the a low caloric intake caused by food aversion, which may be associated with hypoglycemia, constipation, gastroesophageal reflux, esophagitis or gastrointestinal complications. Renal, heart and genitals abnormalities may also be present in a minority of cases, requiring further investigation. The incidence of growth hormone deficiency in these patients is controversial, as several studies have reported normal serum concentration of growth hormone in children with RSS[1,2,4,6,7,9].

Treatment should be focused on major identified anomalies in the clinical evaluation, since not all children have the same features and can often involve the work of a multidisciplinary team, containing physiotherapists, psychologists, neurologists, endocrinologists, among others[6,10].

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Motor development is an evolutionary, sequential and continuous process of movement on the chronological age, where the individual progresses from one simple movement to a more elaborate and coordinated movement. The delayed psychomotor development is defined as a significant delay in several areas of the development, such as language, fine and/or gross motor coordination, cognition, and in activities of daily living, and each of these areas can be compromised to a greater or lesser grade. This delay directly affects the child's life as well as your relationship with environment[11,12].

The early physical therapy is instituted, the better psychomotor development, that contributes to a better prognosis. The physical therapy is planned according to the degree of functionality of the child correlated with chronological age; procedures involving stimulation of neurodevelopmental postures, balance training and gait, strengthening and cognitive stimulation[13].

Although the motor retardation can be considered as one of the complications in the development of children with RSS, few studies have reported this aspect as well as the means of specific treatments for disorders arising out of it[11,12,14,15,16,17]. Thus, this appears to be the first study to analyze the evolution of kinetic-functional child with RSS undergoing physical therapy intervention.

This study aimed to analyze the effects of a neurodevelopmental treatment-based physical therapy intervention in the motor function in a child with RSS.

Methods

The clinical history of the child, main motor dysfunction occurred in the course of child`s development, physical therapy performed and the patient's response to this treatment were described in this study. The intervention and evaluation were performed by a single therapist, during 1-hour, twice a week, along 12 months. This study was approved by the Ethics Committee on Human Research at the Federal University of Sergipe. His progenitor signed a consent form authorizing the child's participation in the study.

Participant

This study was conducted in a physical therapy outpatient at the University Hospital of Federal University of Sergipe. Only one male child, aging 18 months, diagnosed as RSS was involved in the study.

Case Report

Participant was born at term labor, weighting 2475 grams, lenghing 45 cm, with head circumference of 36.5 cm. He was the first and only child of young, healthy, non-consanguineous parents, and no similar cases in their families were reported; the child has small malformations in face, asymmetric upper and lower limbs, with normal karyotype (46 XY) and state.

The geneticist assessed the infant after birth and detected neuromotor developmental delay, deficits in height and weight, standard head circumference, prominent forehead, asymmetry in implementation and size of the ears, bilateral epicanthus, high arched palate, micrognathia, short neck, bilateral clinodactyly of the fifth fingers and toes, single palmar crease, slight asymmetry in size and diameter of the upper and lower limbs. Considering the skeletal asymmetry, short stature, normal head circumference and small changes in face and limbs, the infant received a diagnoses of RSS.

Physical Therapy Assessment

After assessed by the the physical therapist, those functional manifestations were found at 18-month age: deficit in balance for anteroposterior and lateral-lateral sitting position; low hip control; weakness in muscles of abdominal, upper and lower limbs; deficit for staying in semi puppy position; no ability for performing puppy, half kneeling, kneeling and bipedalism positions, and roll efficiently. Also, there was no straightening reaction of the body and difficulty to manipulate objects (fine and gross motor skills deficit) was observed.

Aim of Physical Therapy Treatment

Considering those functional deficits found after physical therapy evaluation, the aims of the study were: (i) to stimulate different stages of neuromotor development, (ii) to contribute to the improvement of static (semi-puppy, puppy, sitting position, quadrupedalism, half kneeling, kneeling and bipedalism) and dynamic (crawling, anterior, posterior and lateral walking) neuromuscular motor activities.

Treatment Protocol

The protocol was developed based on the identified dysfunctions in the infant at the first evaluation and then adjusted according to the motor function improvement. The physical therapy intervention consisted of:

- Rollover stimulus bilaterally with contralateral placement of toys; lower limbs alternating; anterior superior iliac crest stimullation; upper limbs positioning.
- Neurosensomotor strategies to the development and maturation of primary motor reflex patterns in following positions and transfers: lying down, sitting and standing.
- Strenghtening exercises using isometric and isotonic exercises.
- Education of basic movement patterns of trunk, pelvis and limbs to facilitate selective control of movement.
- Gait training.

Measurements

Pediatric Evaluation of Disability Inventory (PEDI) is an instrument used to document functional performance in activities of daily living of children between six months and seven years old[18].

PEDI covers three dimensions: self-care (73 items), mobility (59 items), and social interaction (65 items). Self-care measures actions such as eating, grooming, toileting, dressing, and sphincter control. Mobility assesses functional activities like transfers, indoor and outdoor locomotion, up and down stairs. The social interaction score covers issues about communication, problem-solving, relationship, etc[18].

Each item receives score "0" (if the child is unable to perform the activity) or "1" (if the activity is already part of the functional repertoire of the child). The sum of the scores of these three areas results in raw score[18].

Despite PEDI has three areas, the scales are independent and just the area of greatest functional deficit can be used. For this study, we used only the parts I and II, whereas part III describes the modifications used by the child in performing their activities, and it does not constitute quantitative scales.

Data Collection

The method of evaluation using the PEDI protocol was conducted through interviews with patient`s mother. The interviews were scheduled for the first and last day of treatment being carried out before and after treatment, respectively.

Data Analysis

The values found with the application of the PEDI, the raw, normative and continuous scores were analyzed, separately, comparing the values presented in the pre and post-treatment.

Results

It was observed that the pre-treatment, within the areas that constitute the functional skills, the results of raw score, in both areas (self-care and mobility), showed a very low score (2 and 2, respectively) having a slightly better performance in the area of social function (6) (Figure 1).

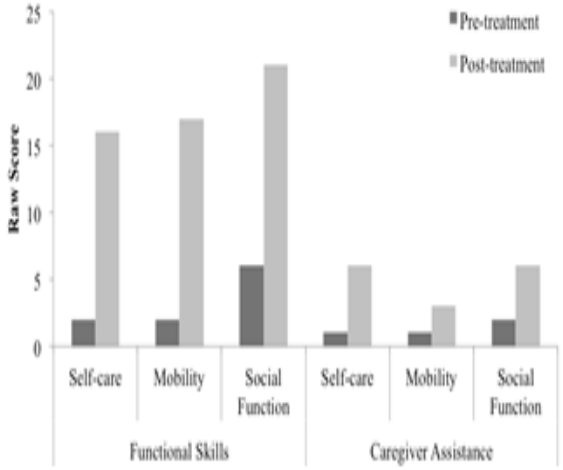

Figure 1. Raw score according to the Pediatric Evaluation of Disability Inventory (PEDI), for the areas of self-care, mobility and social function in the pre and post-treatment periods considering functional skills and caregiver assistance.

At the post-treatment phase, there was an increase in all raw scores, especially for the self-care and mobility areas. Considering the caregiver assistance, it can be noticed that the child needed a high level of assistance before treatment, and then decreased after treatment. However, but the assistance for mobility was still necessary after treatment.

Through analysis of the normative score (Figure 2), at the functional skills, it was noted that the child received scores below 10-points in the three areas assessed before treatment, meaning a lower development than that expected in children with typical development.

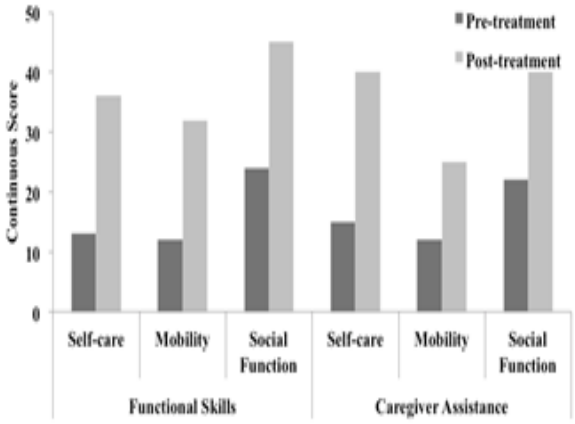

Figure 2. Continuous Score according to the Pediatric Evaluation of Disability Inventory (PEDI) for the areas self-care, mobility, and social function, in the pre and post-treatment periods considering functional skills and caregiver assistance.

After treatment, the normative score of the area of self-care increased, but it still showed that the child has developmental disorder. In the area of mobility, even with an increase in raw score, the child still scored below 10-points; in the area of social function, the child presented a significant advance in its development, scoring 36.1, showing that, for its age, the child is within normal limits.

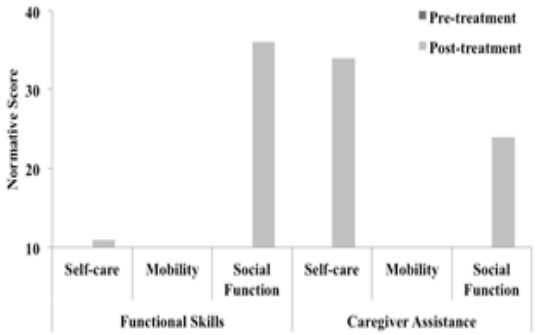

Figure 3. Normative Score according to the Pediatric Evaluation of Disability Inventory (PEDI), for the areas of self-care, mobility, and social function, in the pre and post-treatment periods considering functional skills and caregiver assistance.

Following caregiver assistance analysis before the treatment, despite the assistance to be great in all areas, there was the assistance given to the child's mobility, which reaffirms the motor disturbance in the functional skills. At post-treatment, the assistance decreased in the area of self-care, even if the child was still showing a low score (10.9) in the same area at the functional skills. In the area of mobility, the assistance still remained high. In the area of social function, it was observed that the child had a higher score before than after treatment.

All of values increased, both functional abilities of infant and offered assistance by the caregiver in post-treatment, which shows development of the infant within a constant time.

Discussion

Studies have shown that PEDI test is valid and reliable in applications for children in Brazil19,20,21,22. The present study focused only on the dimensions of self-care and mobility in functional activities.

The results of this study demonstrated that there was an evident increase in both raw and continuous scores in the areas of self-care, mobility and social function. Importantly, the raw score is the sum of the scores obtained in the requested tasks by the PEDI, and the continuous score shows infant`s development within a period of time, without comparing him with other children.

However, when analyzing the normative score, it was observed that there were positive changes in the area of mobility. The normative score compares the child with delayed motor development in children with normal motor development at the same age group. This leads us to assume that, although the child had presented an increase of motor development, determined by raw and continuous scores, according to the normative score, he showed a development lower than that was expected.

In this study, the infant presented, in both raw and continuous scores, higher score in the social function when compared with areas of mobility and self-care before the treatment, despite his social development is considered lower than expected, according to the normative score.

It is assumed that this has occurred because the infant presents a preserved cognitive state, although no tools for this diagnosis were used. In a study by Monteiro et al19, which evaluated through the PEDI functional ability and need for

caregiver assistance in 64 patients with Rett syndrome, the area of social function showed the greatest deficiency, which can be explained by serious cognitive impairment in patients with Rett syndrome. This fact shows the importance of preserving the cognitive state for social and family inclusion.

It is supposed that when the infant was 18 month-year old there was a delay in the development of his social functions when compared with normal children of the same age. However, with 30 month-year old, it was expected that the progress of development would have been larger when compared with the degree of progress that infant obtained at 18 month-year old.

One factor that may have influenced this score is the vocabulary-related language delay, once that the infant showed little more than four words in the post-treatment period. It may be that the motor delay is related to the delayed acquisition of understanding language21.

The delay in motor development in RSS is a disorder that may significantly influence the performance of activities of daily living, having a negative impact on the functionality and independence of the infant.

It also observed an association of motor development with optimization of the self-care tasks, as well as the increase in the scores related to social function. With this, it can be stated that the lower the score in the area of mobility, lower will be motor independence and functional performance of the infant20.

Before the treatment, this infant showed that the area of self-care proved quite deficient in functional abilities, which meant that the infant needed a very significant help to carry out him tasks. After treatment, there was an improvement of self-care, considering all the scores, although the increase of normative score was not satisfactory.

During treatment, guidelines were given to the child's mother, so she encouraged her son to try to do some simple activities related to self-care. However, in the course of the sessions, it was noticed that the guidelines were not followed, because even the infant being able to perform some of these tasks, the progenitor has promoted aid in large-scale.

It is assumed that poor stimulation to perform self-care tasks at home was a factor that contributed to the infant had not gotten a major evolution in this area. According Brianeze22, a physiotherapy program is crucial for the improvement of functional abilities, as well as the importance of encouraging the activities of daily life at home.

Perform the taught tasks in physiotherapy sessions at home is an optimizer and substantial factor for the significant, continuous and permanent increase in children with developmental delay. Moreover, there is a view to the fundamental role of the family to the achievement of satisfactory results.

Conclusions

Based on the review conducted for yield this paper, it can be seen that the literature is scarce regarding the description of cases involving developmental delays in children with RSS.

The planned intervention protocol proved to be very effective in the identified abnormalities during the evaluation. In conclusion, physical therapy evidenced a key role in the stimulation and acceleration of motor development and, consequently, functional improvement and social functions of this infant with RSS.

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