Case Report

Motor development of a child with rubinstein-taybi syndrome submitted to physiotherapeutic intervention

Desenvolvimento motor de uma criança com síndrome de rubinstein-taybi submetida à intervenção fisioterapêutica

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ABSTRACT | OBJECTIVE: Analyze the motor behavior of a child with Rubinstein-Taybi Syndrome under physical therapy stimulation. **METHOD:** This is a case study realized with a child diagnosed with SRT, evaluated before, during, and after physiotherapy sessions, through the Alberta Infant Motor Scale. The treatment was fulfilled at the Sun Ray Integration Center located in the Santa Maria neighborhood, in Aracaju / SE in which the ducts were applied three times a week for 50 minutes and consisted in the training of neuroevolutionary postures. **RESULTS:** It was verified an increase in all postures scores in the second and third evaluations, which shows an improvement in motor development. **CONCLUSION:** The Physiotherapeutic intervention can bring benefits to treat the characteristic delay of SRT, confirming the importance of further studies investigating motor performance and the effect of early stimulation in children with this syndrome.

DESCRIPTORS: Developmental Disabilities. Physical Therapy. Rubinstein-Taybi Syndrome. Child Health. Multiple Abnormalities.

RESUMO | OBJETIVO: Analisar o comportamento motor de uma criança com Síndrome de RubinsteinTaybi sob estimulação fisioterapêutica. **MÉTODO:** Trata-se de um estudo de caso realizado com uma criança diagnosticada com SRT avaliada antes, durante e após sessões de fisioterapia através da *Alberta Infant Motor Scale*. O tratamento foi realizado no Centro de Integração Raio de Sol, em Aracaju/SE, onde as condutas foram aplicadas três vezes na semana, com duração de 50 minutos, e consistiram no treino das posturas neuroevolutivas. **RESULTADOS:** Verificou-se aumento nos escores de todas as posturas a partir da segunda avaliação, o que demonstra melhora no desenvolvimento motor. **CONCLUSÃO:** A intervenção fisioterapêutica pode trazer benefícios no tratamento do atraso característico da SRT, ratificando a importância de novos estudos que investiguem o desempenho motor e o efeito da estimulação precoce em crianças com essa síndrome.

PALAVRAS-CHAVE: Deficiências do desenvolvimento. Fisioterapia. Síndrome de Rubinstein-Taybi. Saúde da criança. Anormalidades múltiplas.

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Introduction

Rubinstein-Taybi Syndrome (RTS) is a genetic condition that can be caused by elimination or mutation of the CREBBP gene located in the chromosomal region 16p13.3 in which the CREB binding protein is encoded or by the 22q13 EP300 gene encoding the p300 protein. There are few reports attributed to the EP300 gene, equivalent to a percentage of 8% to 10% of cases, whereas mutations in the CREBBP gene commonly occur in about 60%[1].

The CREBBP gene has the function of instructing the production of the CREB protein, important in the regulation of growth and cell division, so when a copy of the CREBBP gene is eliminated or mutated, the cells produce half the normal limiting the function exerted by it interrupting the normal development. What happened is similar to what happens to the EP300 gene, which instructs the production of the specific protein that helps to control the activity of other genes. Both are important for development before and after birth[2].

Despite being a rare syndrome, affecting 1 in every 125,000 individuals, it has great clinical variability making it difficult to establish relationships between genotype and phenotype[3]. Genetic alterations give the patient-specific clinical characteristics such as microcephaly, high nasal bridge, excess hair, large thumbs, duplicate distal phalanges, hypotonia and ligament laxity, growth and language retardation, intellectual disability, and behavioral problems[4-6], in addition to the frequent presence of malignant and benign tumors with a significant prevalence of meningiomas and pilomatrixoma[7].

Several syndromes have a delay in neuropsychomotor development associated with them, but each has its own characteristics and degrees of impairment, taking into account the type of syndrome, external factors, such as family and social environment, and internal factors such as tone and visual acuity, leading physiotherapy to determine a prognosis according to a set of influential factors and not just the cause of the disability[8].

Studies point out that motor delays diagnosed early tend to result in a good prognosis, highlighting the role of neuropediatric physiotherapy that aims to stimulate motor learning by inducing the nervous system to practical and dynamic changes, leading the child to functional independence[9,10]. No findings were spotted in the literature on the physical therapy intervention in the respective syndrome, even though it knows its importance in correcting motor delays. Thus, this study aimed to analyze the motor behavior of a child with RTS under physical therapy intervention.

Methodology

This is a case study approved by the Ethics and Research Committee of the Estácio of Sergipe University Center under CAAE nº 98140718.6.0000.8079 and opinion nº 2.956.639, carried out with a child diagnosed with RTS, who started the research at the corrected age of 7 months and has been followed since her insertion in the Pediatric Physiotherapy service of the Raio de Sol Integration Center, located in the Santa Maria neighborhood, in Aracaju/SE.

The Alberta Infant Motor Scale (AIMS) was used as a motor monitoring instrument, where the child was assessed before, during, and after physiotherapy sessions by the same evaluator, obeying an interval of three months between evaluations, totaling a period of nine months follow-up. The tests were performed through free observation of the child's spontaneous movement, with an average collection time of 02 hours.

The treatment consisted of motor physiotherapy sessions performed three times weekly for nine months with a daily duration of 50 minutes and consisted of training neuroevolutionary postures and stimulating balance and protection reactions, using resources such as therapeutic rolls and balls, skateboards, and surfboards balance.

Results

There was an increase in the sub-scale scores of all neuroevolutionary postures in the second assessment, which demonstrates that after three months of treatment, there was an improvement in motor development and evolving with higher scores in the third and fourth assessments, as shown in Graph 1. Then, an important gain in the acquisitions of rolling was carried out, extending postures in the prone position, trunk control in sedation, and weight support in orthostasis.

Graph 1. Graphical representation of AIMS subscale scores

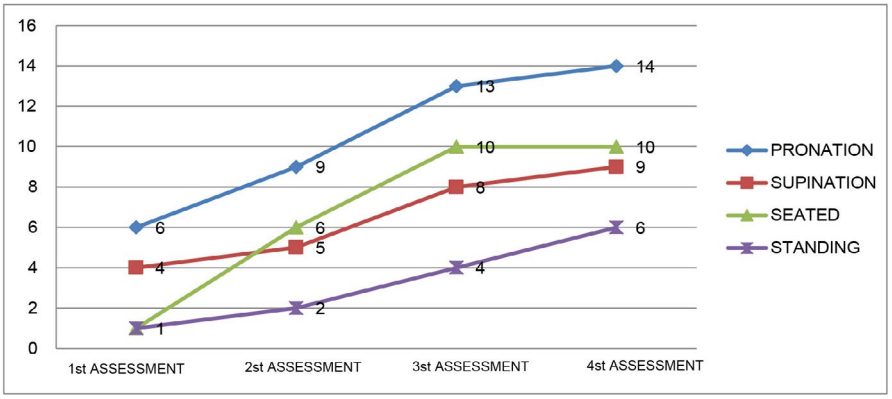

Table 1 presents the values of the raw scores of the four assessments performed, where motor behaviors showed that even with changes in the subscale values, resulting in a consequent increase in total scores, the child still presents motor delay when taking into account his corrected age.

Table 1. Representation of total AIMS scores

	CORRECTED AGE	GROSS SCORE	CLASSIFICATION
1st ASSESSMENT	7 MONTHS	12	< 5%
2st ASSESSMENT	10 MONTHS	22	< 5%
3st ASSESSMENT	12 MONTHS	35	< 5%
4st ASSESSMENT	16 MONTHS	39	< 5%

AIMS: *Alberta Infant Motor Scale*

Discussion

It was possible to perceive the importance of physical therapy intervention in minimizing motor delays resulting from RTS, noting an improvement and gains in all neuroevolutionary postures.

Concerning supine and pronated postures, the study by Rodrigues et al. is similar to the findings of this report when they verified an improvement in motor performance in a premature child after physical therapy intervention, which consisted of psychomotor stimulation, techniques to facilitate motor skills and stimulate sensorimotor skills and postural tone. Also, assessed through the AIMS, there were gains in the prone posture evidenced by the control of the cervical, support in the forearms and hands in an attempt to explore the space that surrounded it, as well as the improvement in the supine position in which the child experiences the rotation of the head towards some stimulus and thus activates your perception and laterality[11]. Other studies[12,13] reinforce that, as the prone position is improved, the child develops skills in weight support against gravity, favoring learning in supine and sitting postures.

This study also demonstrates that as the patient increased the score in the prone posture, the score of the sitting posture also increased, starting to acquire ten postures in the last application. Pin et al. affirm that the sitting position is related to other motor acquisitions, mainly the prone posture, due to the recruitment of the muscles of the head and upper trunk to perform extension, thus facilitating the acquisition of trunk control in sedation[14]. Vala also mentions that the average age of the individual with SRT learning to sit is between 1 and 2 years old, which happened to the child in this study. At the end of the treatment with a corrected age of 12 months, she maintained the position of sedestation without the support and performed manipulation of objects in this posture[15].

The stimuli aimed at manipulating objects activate muscles working joints such as the shoulder, elbow, and wrist intervening in the kinematic change of the hand, inducing the child to reach, midline, and dissociate these joints[16,17], in addition, trunk control in sedation. it generates axial stability that physiologically facilitates appendicular activities such as the functions of the upper limbs in exploration, eye-hand coordination, and object manipulation[18,19]. These results coincide with the findings in this study, as the child acquired the ability to manipulate objects and bring the midline, in addition to acquiring postural control in the sitting and standing positions with support.

In the standing posture, the child showed an evolution, performing weight unloading and maintaining orthostasis with support, however, there was no gait acquisition. It is known that walking is a later skill and, according to AIMS, it develops between 10 to 15 months. Thus, at the end of the intervention, the child under this study had 12 months of corrected age and was therefore in the physiological period of gait development. However, it is essential to highlight that the influence of the genetic condition in the face of delays in important neuroevolutionary stages, such as sitting and crawling, contributes to the delay in the walking process, and in individuals, with RTS this acquisition occurs around the age of 4[15].

According to the Association of Rubinstein-Taybi Syndrome (ARTS), individuals with RTS have the characteristics of adapting to schedules and routine tasks, and this aspect facilitates the maturation of their activities. Thus, taking into account that the intervention process in the acquisition of motor learning requires repetition, it is important to include training in the child's routine until the creation of engram, development of functional skills, and improvement of acquired functions[20].

Conclusion

In this context, it is concluded that the physical therapy intervention in Rubinstein-Taybi Syndrome may bring benefits in the development of neuroevolutionary motor skills. Despite the relevance of this study, which proposes to disseminate scientific knowledge on this little explored and well-known theme, there are limitations such as the scarce literary collection about the motor performance of children with RTS, mainly regarding early stimulation. It is therefore suggested that new studies be carried out to accompany the global development until the acquisition of gait and that propose new therapeutic interventions, in order to minimize the delays inherent to the syndrome.

Author contributions

Santos AP, Silva NC and Nascimento LKA were responsible for the execution of the project, data collection, tabulation, data analysis and creation of tables and figures. Santos AP, Silva NC were responsible for writing the text and standardizing the rules. Santos AP and Nascimento LKA are responsible for revising the text and adding significant parts. Nascimento LKA guided the project.

Competing interests

No financial, legal or political conflicts involving third parties (government, companies and private foundations, etc.) have been declared for any aspect of the submitted work (including, but not limited to, grants and funding, participation in advisory council, study design, preparation of the manuscript, statistical analysis, etc.).

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