ORIGINAL RESEARCH

Proposal for a physical therapy program in a child with Angelman syndrome, emphasizing postural balance: a case study

Proposta de atuação fisioterapêutica em uma criança com síndrome de Angelman, enfatizando o equilíbrio postural: estudo de caso

Propuesta de actuación fisioterapéutica en un niño con Síndrome de Angelman, enfatizando el equilibrio postural: estudio de caso.

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ABSTRACT | The Angelman syndrome is characterized by neuromotor difficulties, such as ataxic gait and delayed acquisition of motor skills. However, there are few studies investigating the effect of interventions directed to this population. This study aimed to investigate the effect of a balance training in a child with Angelman syndrome. The participant was a nine-year-old girl. The training protocol was implemented during an eight-week period, twice a week, and consisted of activities involving static balance under various difficulty levels. After the training, the postural sway measured by biophotogrammetry changed from 38° to 13.78°. The scores in the Berg scale changed from 27 points to 37 points. In the Timed Up & Go test, the child's time to complete the task changed from 15 to 12 seconds. Taken together, the results suggest that the training led to improved static and dynamic balance, as well as functional mobility.

Keywords | Angelman syndrome; rehabilitation; postural balance.

RESUMO | A síndrome de Angelman (SA) é caracterizada por alterações neuromotoras como marcha atáxica e atraso na aquisição de habilidades motoras, porém são escassos os estudos investigando o efeito de intervenções aplicadas a essa população. O objetivo do estudo foi verificar o efeito de um treino de equilíbrio em uma criança com SA. Participou do estudo uma criança de nove anos de idade com diagnóstico de SA, sexo feminino. Foi aplicado um protocolo para treino de equilíbrio por oito semanas, com frequência de duas vezes por semana. O treino consistiu em atividades envolvendo equilíbrio estático sob diversas condições de dificuldade. Após o treino, a análise de biofotogrametria computadorizada do equilíbrio estático revelou redução do grau de oscilação, que passou de 38° para 13,78°. A pontuação na escala de Berg passou de 27 pontos, na avaliação, para 37 pontos na reavaliação. No teste *Timed Up & Go*, a criança realizou a tarefa em 15 segundos, na avaliação, e, na reavaliação, em 12 segundos. Em conjunto, os resultados sugerem que o treino favoreceu melhora no equilíbrio estático e dinâmico, bem como na mobilidade funcional.

Descritores | síndrome de Angelman; reabilitação; equilíbrio postural.

RESUMEN | El síndrome de Angelman (SA) es caracterizado por alteraciones neuromotoras como marcha atáxica y atraso en la adquisición de habilidades motoras, pero son escasos los estudios investigando el efecto de intervenciones aplicadas a esta población. El objetivo de este estudio fue verificar el efecto de un entrenamiento del equilibrio en un niño con SA. Participó del estudio un niño de 9 años de edad con diagnóstico de SA, sexo femenino. Fue aplicado un protocolo de entrenamiento para el equilibrio por 8 semanas, con frecuencia de 2 veces por semana. El entrenamiento consistió en actividades involucrando el equilibrio estático sobre diversas condiciones de dificultad. Después del entrenamiento el análisis de biofotogrametría computarizada del equilibrio estático reveló reducción de los grados de oscilación, que pasó de 38° para 13,78°. La puntuación en la escala de Berg pasó de 27 puntos en la evaluación para 37 puntos en la reevaluación. En el test Timed up and go la niña realizó la tarea en 15 segundos en la evaluación y 12 segundos en la reevaluación. En conjunto, los resultados sugieren que el entrenamiento favoreció la mejora en el equilibrio estático y dinámico, así como de la movilidad funcional.

Palabras clave | Síndrome de Angelman; rehabilitación; equilibrio postural.

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INTRODUCTION

The Angelmen Syndrome (AS) is frequently a result of deletion or rearrangement of the long arm of chromosome 15 in the 15q11-q13 region[1,2]. Among the characteristics of the syndrome are optic atrophy, cerebral atrophy associated with ventricular dilatation, intellectual disability and macrostomia[3-5]. Besides, other symptoms are common: delayed motor development, hyperactivity, severe compromise in language, peculiar happy behavior with unmotivated laughter, seizure, myoclonia, stereotyped upper limb movements and ataxic gait[6,7].

The ataxic gait can really affect the functionality of subjects with AS, and it is a result of cerebellar deficits in the adjustments of motor responses by means of the feedback system, as well as the force modulation and the amplitude of movements involved in the motor learning and in balance control[8,9].

Balance is the skill that maintains body orientation in relation with the external environment, and it depends on the continuous transmission of visual, somatosensory, vestibular and proprioceptive information, besides the coordination of neuromotor recruitment patterns. When this skill is compromised, the stability could be reduced, thus increasing body oscillation or changing the strategy of movement in response to disturbances[10,11].

One of the ways to practice balance is based on the destabilization of the patient so that there can be a postural adjustment, enabling the use of movement strategies in the sagittal plane (anterior posterior) and in the frontal plane (medial-lateral), in order to maintain balance in many circumstances[12,13]. Besides, balance training can have additional challenges to encourage the use of the remaining vestibular information, or to make other sensory information gradually available, for instance, using different surfaces.

Rodrigues et al.[14] assessed patients with demyelinating disorders and balance deficits. One group performed specific balance training and the other were submitted to general physical therapy with emphasis on strengthening and stretching, without emphasizing balance. The conclusion was that the group with specific training presented greater improvement in balance than the group with global treatment.

Studies demonstrate that the balance training applied for children with cerebral palsy[15] and adults with ataxia[16] — using stable surfaces, progressing for unstable ones[15], and movements with varied speed and complexity[16] — promote significant balance improvement[15]. There is still evidence that the balance training is associated with the improvement in locomotor skills in children with cerebral palsy at school age[17]. Such studies show that the balance training is relevant to improve the functional development. However, no studies investigating the effect of balance training in patients with AS were found. Besides, there are only few studies that use an assessment instrument that is easy to employ in the clinical practice of physical therapists, who do not have access to complex systems for kinetic and biomechanical analyses.

Therefore, this study aims to verify the effect of balance traning on balance and on the functional mobility of a child with AS. The participant is expected to show improved static and dynamic balance, expressed by the increased score in the Pediatric Balance Scale, as well as a lower angle of postural oscillation in posturometry. Benefits for functional mobility are also expected, demonstrated by the reduced time to perform the Time Up & Go (TUG) test.

CASE REPORT

A nine-year-old female patient participated in the study. From the development history, the child presented with delayed development, being referred to genetic counseling when the clinical diagnosis of SA was concluded, in 2004. From this moment on, the child has been followed-up by a multiprofessional team, with a physical therapist, occupational therapist, phonoaudiologist and psychologist.

In the training period, the child remained on treatment with the multidisciplinary team, going through conventional therapy in a group twice a week, in 30-minute sessions.

The study was approved by the Human Research Ethics Committee from the University Center Unifafibe (protocol 0246/2010).

Instruments

TUG is a valid instrument that assesses the time spent by the subject to stand up from a chair, walk a 3 m distance, turn around, walk towards the chair and sit down again[18].

The Berg Balance Scale (modified), the Pediatric Balance Scale[19], is a valid instrument to assess children with motor dysfunctions and it investigates static and dynamic balance, counting on 14 items that are common to the daily life. Each item is scored according to an ordinal scale that ranges from 0 to 4 points. Points are based on how long a position can be maintained, on the distance which the upper limb can reach in front of the body and on the time to complete a task. The maximum score is 56 points[19]. The instrument has been through a recent cultural adaptation to be applied in Brazilian pediatric patients[20].

Biophotogrammetry is a method used to analyze body angles through phographic data[21,22]. In this study, the method was used in association with the Romberg test, which assesses static balance with the patient standing up on a bipodal support, with open eyes and a half-open base at the same distance from the shoulder, keeping this position for 30 seconds[23]. In order to evaluate body oscillations in the sagittal plane (anterior posterior sways) in the stage of interpreting the images for the posturometry analysis, a reflective adhesive was placed in the earlobe region to assess the deviations in the balance line related to the angle in relation to the plumb line. In order to perform the test, a plumb line attached to the ceiling was used and placed 30 cm away from the wall, while the digital camera used to capture the images was placed 1,5 m away from the wall and 1,28 high in relation to the ground.

The picture with major degree of body oscillation was selected. In this picture, a straight line was outlined perpendicular to the plumb line to determine the intersection point[24].

For the results of the computerized biophotogrammetry to be more reliable, the assessment and reassessment images were submitted to the intraobserver reliability test, in which images were assessed within a one month interval, with reliability of 0.9.

Data collection was performed before and after balance training, called, respectively, assessment and reassessment.

Intervention Protocol

Fifteen sessions of balance training were performed for an eight-week period, twice a week and lasting 50 minutes each[16].

The balance training was performed by means of eight activities, counting on different support bases and using sensory and motor strategies (Chart 1). The protocol used in this study was adapted from Alegretti et al.[15].

RESULTS

According to the Berg scale, at the initial evaluation the patient presented less than 4 points in all the tasks, with more difficulty in the items "remain supported with one foot to the front", "remain on a bipodal support", "turning to look back" and "reaching an object ahead", in which the score was 0. At reassessment, the child presented changes in 9 items, out of the 14 items, as demonstrated in Table 1.

In the TUG test, the child performed the task in 15 seconds during assessment. At reassessment, performed after the physical therapy intervention, the task was performed in 12 seconds.

According to the analyses performed through biophotogrammetry, at the assessment a 38º anterior posterior sway was observed, and at reassessment, the calculated sway was of 13.78º.

Chart 1. Activities for balance training

1	Patient standing up with bipodal support at a stable surface. The therapist performed a unidirectional and bidirectional distabilization with mild intensity.
2	Patient standing up on a stable surface with a narrow base. The therapist performed unidirectional and bidirectional distabilization.
3	Patient on unipodal support on a stable surface. Functional upper limb tasks were performed according to her functional capacity.
4	Patient standing up with bipodal support over a foam surface with average density, with parallel feet slightly separated.
5	Patient standing up with bipodal support over a foam surface with average density, with parallel feet slightly separated. The therapist conducted anteriorposterior and laterolateral distabilizations.
6	Patient standing up with bipodal support over a foam surface with average density, with parallel feet slightly separated. Upper limb tasks were performed, such as throw the ball, demanding postural adjustment from the child.
7	Patient standing with unipodal support over a foam surface with average density.
8	Patient on bipodal support over na elastic bed.

Table 1. Score obtained by the participant in the assessment and reassessment according to the Berg balance scale

Assessed items	Assessment	Reassessment
1. From standing to sitting	3	4
2. From sitting to standing	3	4
3. Transfers	2	3
4. Stay standing	3	3
5. Sitting with the back supported and feet without support on the ground	3	4
6. Stay standing with eyes closed	3	3
7. Staying on a bipodal support with the feet together	3	3
8. Staying in a semi tandem position	0	1
9. Staying in a unipodal support	0	1
10. Turning 360 degrees	1	1
11. Turning to look back, left and right, while the shoulders remain still	0	2
12. Picking na object on the ground from an erect position	3	4
13. Putting the feet alternately on a step without support	3	4
14. Reaching ahead standing up with the extended arm	0	0
Total	27	37

DISCUSSION

The study aimed to analyze the effect of the physical therapy treatment with emphasis on the balance training of a child with AS during a two-month period.

At the initial evaluation, the child presented lower score in relation to the highest score in the Berg scale in all of the assessed items. The TUG evaluation also showed a worse than expected performance for the age group (15 seconds), since the mean expected for children aged between three and nine years is 5.9 seconds (ranging from 3 to 13 seconds)[25]. Finally, posturography showed major body oscillation, demonstrating imbalance. Such results, altogether, point to postural control and balance deficit, which justify the application of a protocol emphasizing this aspect.

After the application of the physical therapy intervention program, it was possible to observe changes in performance according to the Berg scale in items related to postural transfer and tasks of static balance. This result suggests that even though intervention was focused on static balance, it was possible to improve dynamic balance. Such improvement is relevant for the participant's performance, since the deficits in static and dynamic balance in subjects with ataxia have been related with higher risk of falling down[26].

There is evidence in literature showing that the physical therapy intervention leads to the improvement in static and dynamic balance of subjects with ataxia, demonstrated by the increased score in items performed on foot and without support in a patient with ataxia, and such improvement enables the better functional performance[27,28]. Likewise, the balance training on different support bases, employed on patients with vestibular changes, improves dynamic and static balance[29], which corroborates this study. However, the effect of the intervention in children with AS, who not only present with ataxia, but also intellectual disability and balance changes, had not been assessed in previous studies. Therefore, the results in this study suggest it is possible to improve the static and dynamic balance of subjects with AS.

The decreased time of execution of TUG demonstrates the improvement in dynamic balance, suggesting that balance training can favor functional mobility. The shorter time to execute TUG corresponds to lower risk of falls[18], which is extremely important in order for the child to participate in socially relevant activities. In a prior study by Campos et al.[30], it was observed that this test reliably measures the changes that come with intervention, with the advantage to be easily employed in the clinical practice. Alegretti et al.[15] observed that balance training benefits children with cerebral palsy, with reduced time to perform the test, improved balance and reduced frequency of falls. The findings in these studies support the findings in the child with AS, since the physical therapy protocol is similar to that proposed by Alegretti et al.[15], with similar results.

According to biophotogrammetry, the child presented reduced child oscillation after the employment of the intervention protocol, which indicated improvement in the static balance. Similarly, balance training reduced the body oscillation assessed by means of the force platform in children at school age with hemiplegic cerebral palsy[17]. Computerized biophotogrammetry was used to check static balance in children and adolescents with Down syndrome in comparison to children without disorders, and balance was assessed with eyes open and closed. This instrument proved to be reliable and easy to be employed for physical therapy evaluation, analyzing the frontal and sagittal plane displacement[24]. In this study, computerized biophotogrammetry also proved to be capable of measuring changes in the static balance of a child with AS. The only study assessing patients with ataxia by menas of biophotogrammetry demonstrated that the training in a vibrating platform reduces the patients' oscillations, but only in dynamic situations[31]. Even though this result is slightly different from this study, such difference can be due to the characteristics of the clinical picture and the different types of training.

Possible mechanisms to explain the observed changes include the improvement of directional specificity of the activated muscles facing external stimulation, as well as the improvement in spatial or temporal characteristics of muscle responses, such as faster response, lower agonist-antagonist coactivation and better modulation of amplitudes of contraction[32,33]. The integration of somatosensory, vestibular and visual stimuli seems to be adequate to minimize the insufficient balance in patients with AS. Besides, the balance disturbance generates external stimulation, facilitating the motor signals created in cortical motor centers and the brain trunk, adapting the muscle tonus during movements and enabling postural adjustment[28,34]. So, such aspects were considered for the elaboration of the training proposed for the child with AS and showed satisfactory results.

CONCLUSION

Even though it is limited to one case report, this study demonstrated the improvement in static and dynamic balance after specific training for a child with AS. Both the employed training protocol and the used instruments of assessment are easily accessible and applicable, therefore being relevant for the clinical practice of physical therapists.

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