Locomotor Training Protocol for a Joubert Syndrome Child: A Single Case Study

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Abstract: Joubert Syndrome (JS) is a rare and complex genetic ciliopathy, characterised by cerebellar malformation, resulting in hypotonia, ataxia, and profound neuropsychomotor delays, leading to challenges in gait acquisition. Evidence for effective, targeted locomotor interventions in this population is scarce. This study aimed to evaluate the effectiveness of a six-week structured locomotor training protocol on independent walking (≥10 steps) and participation. A single-case experimental design (A-B-A) was implemented. The four-year-old boy underwent 4-week intervention, with sessions administered four days per week — two conducted by the researcher and two by the family members. Outcomes included motor behavior by the Infant Motor Profile (IMP), administered three times at baseline, twice weekly during intervention, and three times post-intervention. Participation was assessed through the Young Children's Participation and Environment Measure (YC-PEM), administered once at pre- and post-intervention. The child achieved the primary functional goal of independent walking for more than 10 consecutive steps. Parent-reported participation increased across home, school, and community settings, with a notable shift in parental support strategies. In contrast, the IMP changes were minimal. It is concluded that a focused, intensive, structured locomotor training protocol facilitated the gait acquisition and enhanced participation of a child with JS, supporting the role of task-specific, family-inclusive rehabilitation in individuals with this condition. Nevertheless, future research should explore strategies to integrate approaches addressing co-occurring behavioural challenges to optimise engagement and efficacy of neuromotor intervention programs.

Keywords: Joubert Syndrome, Gait, Participation, Motor outcome, Single-case report.

INTRODUCTION

Joubert syndrome (JS) is a rare, genetically heterogeneous ciliopathy neurological disorder first described by the Canadian neurologist Marie Joubert in 1969 [1]. The JS condition results from pathogenic variants in one of over 34 genes (predominantly autosomal recessive and one X-linked) [2, 3]. JS disrupts primary cilium function, pivotal for cell signaling and embryonic development [2]. Specifically, JS is classified as a sensory ciliopathy associated with mutations affecting proteins located in the distal region of the basal body, such as HYLS1, OFD1, and TALPID3 [4]. The worldwide incidence estimation ranges from 1 in 55,000 to 200,000 live births, with 284 confirmed cases reported in Italy [5].

Diagnosis is based on the association of specific phenotypic characteristics with neuroimaging findings. Neuroimaging typically reveals malformations in the brainstem - elongation and narrowing of the pontomesencephalic junction, deepening of the interpeduncular fossa - , and on cerebellar vermis - thickening and horizontalization of the superior cerebellar peduncles, hypoplasia or incomplete midline fusion – [6, 7]; visible as the pathognomonic pattern of "molar tooth sign" [7, 4]. The clinical phenotype is complex and multisystemic. Reported manifestations encompasses craniofacial disorders (macrocrania, prominent forehead, epicanthal folds, ptosis, anteverted nostrils, low-set and tilted ears, eversion of the lower lip, and retinitis pigmentosa), alongside to disorders in ocular system (nystagmus, strabismus, congenital retinal dystrophy, ocular coloboma), musculosqueletal system (syndactyly, and polydactyly), respiratory system (hyperpnea and sleep apnea), visceral involvement (hepatic fibrosis, renal dysplasia and multicystic kidneys), and behavioral issues (self-injury and self-directed biting) [8-11].

The core neurological presentation involves a triad of hypotonia, ataxia, and oculomotor apraxia, which collectively create a profound and persistent barrier to motor development [8, 9, 11]. A significant and often devastating consequence is a severe impairment in gait acquisition; a substantial proportion of children with JS restrictively achieve an impaired gait [11]. This locomotor deficit directly limits participation in daily life, social engagement, and overall quality of life, establishing a critical need for effective targeted rehabilitation strategies.

Previous studies have demonstrated physiotherapy as a cornerstone of management for children with JS. The case study of a 19-month-old infant with JS conducted by 11 reported improvements in rolling,

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sitting, crawling, walking, climbing, and descending stairs following a 13-month physiotherapy program [12]. Moreover, robust evidence of physiotherapy efficacy in analogous conditions, such as ataxic cerebral palsy, supports the efficacy of motor learning principles - specifically, high-intensity, repetitive, task-oriented training, and functional training approaches – [13] for driving neuroplastic adaptation and functional gains [13, 14] across biopsychosocial domains.

Despite the growing knowledge about the clinical characteristics of JS, there is a significant methodological limitation regarding the current literature on JS rehabilitation. Existing reports are predominantly descriptive, lacking standardized, protocol-driven interventions with rigorous outcome measurement. Additionally, no studies known have investigated the effects of a progressive, task-oriented locomotor program combining treadmill, obstacle-based, and functional gait training in children with JS.

To address this gap, a structured locomotor training protocol has been developed, grounded in contemporary motor learning bases. This study employs a single-case experimental design to provide a rigorous, within-subject evaluation of the protocol's effects. The primary aim was to evaluate the effectiveness of a six-week, structured locomotor training protocol on quantitative measures of motor performance (promote independent walking for more than 10 consecutive steps). The secondary aim was to assess its impacts on participation in home and community settings. We hypothesized that the intervention would lead to measurable improvements in locomotor skill quality and increased participation levels of daily life in a young child with Joubert Syndrome.

MATERIALS AND METHODS

This study employed a longitudinal single-subject experimental study. The study was conducted and reported in accordance with the CARE (CAse REport) guidelines for case reports, available at:https://www.care-statement.org/. The child recruitment was based on convenience.

The study was reviewed and approved by the Human Research Ethics Committee of the Federal University of São Carlos (CAAE: 71592723.3.0000.5504) (Annex A). All procedures conformed to the Brazilian National Guidelines and Regulatory Standards for Research Involving Human Subjects (Resolution No. 466/2012, National Health Council, Brazil). Written informed consent was obtained from the child's parents through the Free and Informed Consent Form (Annex B).

CASE PRESENTATION

A four-year-old male with a confirmed diagnosis of Joubert syndrome, 20 kg, 1,07 m, caucasian, firstborn, born in Osasco-SP, Brazil, and currently living with his parents in Porto Feliz - SP, Brazil. The child was assessed qualitatively and quantitatively during a 6-week structured locomotor training protocol.

The child had a normal gestational history; only in the third morphofunctional ultrasound were alterations in fetal formation observed. Born at term, weighing 3.275 kg, 0.52 m in height, 0.359 m head circumference, and Apgar scores of 8 and 9 at 1^0 and 5^o minutes, respectively. The child was diagnosed with Joubert Syndrome at 2 years and 7 months of age. Brain magnetic resonance imaging, dated November 7th, 2020, revealed the "molar tooth" sign - a cerebellar peduncle abnormality consistent with Joubert syndrome (Annex C).

Clinically, the child presents with generalized hypotonia, ataxia, persistence of primitive reflexes, macrocephaly (0,53m), ocular hypertelorism, oculomotor apraxia, delayed neuropsychomotor development, and non-ambulatory status. Additionally, the child presents behavioral manifestations, including stereotyped hand movements, behavioral dysregulation, including self-injurious behavior (Table **1**). The child receives follow-up care from a multidisciplinary team, including a pediatrician, ophthalmologist, nutritionist, occupational therapist, and physiotherapist.

Aiming to promote independent walking, the child participated in a six-week structured locomotor training protocol. The study followed an A-B-A single-case design. It comprises a 1-week baseline phase (A), in which the child was assessed once by YC-PEM, and three times by IMP assessments with three-day intervals. Followed by a 4-week intervention phase (B), in which the researcher performed two additional IMP assessments per week, always before the intervention sessions. Finally, the last section was a 1-week post-intervention phase (A), which repeated the first phase assessments design (Figure **1**). All intervention sessions and assessments were conducted in environments familiar to the child to maximise ecological validity and minimise distress. Sessions took place either in the child's home or at the Rehabilitation Center of Santa Casa de Misericórdia de Porto Feliz (Porto Feliz, São Paulo, Brazil). Formal institutional authorization for the use of the centre's facilities was obtained prior to the study (Annex D). To ensure optimal participant state and data consistency, all the assessments were strictly scheduled according to the child's daily routine, ensuring at least one hour

Table 1: Participant Characterization

Characteristic	Information
Age	4 years, 6 months, 19 days
Sex	Male
Diagnosis	Joubert Syndrome
Age at diagnosis	2 years 7 months
Main clinical features	Hypotonia, macrocephaly, ocular hypertelorism, oculomotor apraxia, motor developmental delay, delayed acquisition of independent walking
Observed behaviors	Hand stereotypies, self-regulation difficulties, self-injurious behavior; signs indicative of Autism Spectrum Disorder (ASD), under diagnostic investigation
Weight	20 kg
Height	1.07 m
Head circumference	0.53 m
Chest circumference	0.58,5 m

Legend: Phenotypic characteristics of the participant during the structured locomotor training protocol. Note that the data were collected through parent/caregiver interviews, clinical records, and anthropometric measurements.

Source: Authors.

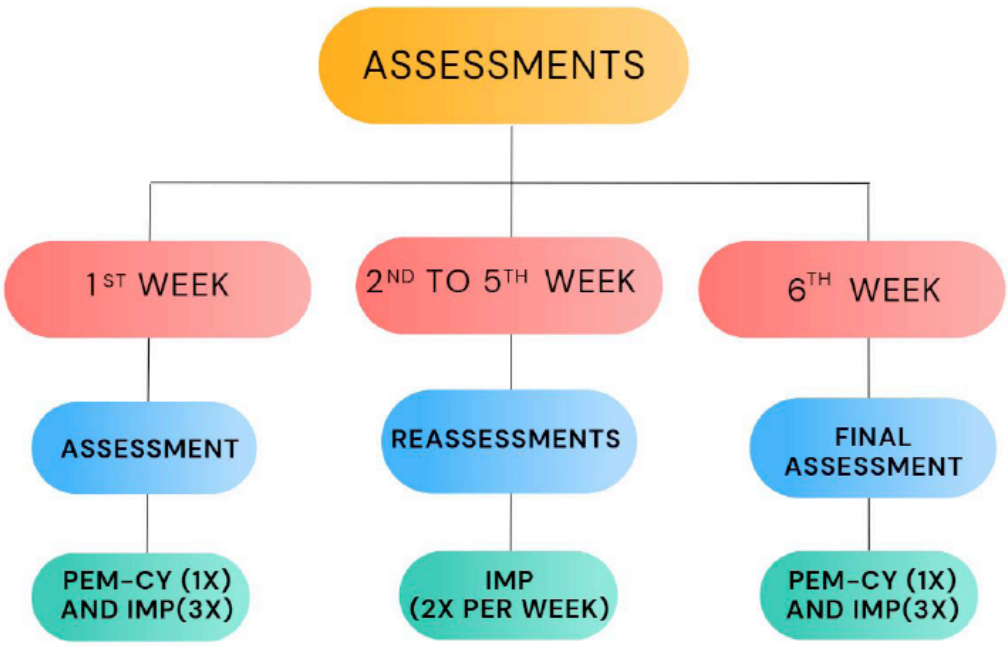

Figure 1: Flowchart of assessment procedures.

Legend = The flowchart explains the assessment procedures. Baseline assessments were conducted during the week preceding the intervention. The Young Children's Participation and Environment Measure (YC-PEM) was administered once. The Infant Motor Profile was administered on three separate days to establish a stable pre-intervention performance mean. Throughout the structured locomotor training protocol, the Infant Motor Profile was administered twice per week to monitor the progression of motor competence. In the week immediately following the intervention, post-intervention assessments were repeated using the same schedule as the baseline assessments. Once by the Young Children's Participation and Environment Measure and three times with the Infant Motor Profile. 1st: First; 2nd: Second; 5th: Fifth; 6th: Sixth; YC-PEM: Young Children's Participation and Environment Measure; IMP: Infant Motor Profile; X: times.

Source = Authors.

post-prandial, during the child's periods of highest alertness, and avoiding vaccination days.

Outcomes were selected to align with the International Classification of Functioning, Disability and Health (ICF) [15] framework, targeting body function and structure (motor performance) and participation. Motor performance was assessed using the Infant Motor Profile (IMP) [16]. The IMP is a validated, video-based observational assessment designed to evaluate the quality of gross and fine motor behaviour in infants and young children (up to 18 months old or the acquisition of mature gait). It comprises 80 items, scored across five domains –

variation, adaptability, fluency, symmetry, and performance – generating the final total score [16, 17]. The patient is evaluated in supine, prone, seated (on the floor and in the caregiver's lap), and standing positions, as well as locomotion and transfers within them [16, 17]. All the assessments were entirely video-recorded with a standardised camera setup. To ensure reliability and minimise bias, all the videos were subsequently scored by a trained and certified assessor of IMP, who was blinded to the study phase. The participation in daily life was assessed using the Young Children's Participation and Environment Measure (YC-PEM) [18]. The YC-PEM is a parent-reported questionnaire that measures a child's (ages 0 to 5) involvement in activities across three settings: home, daycare/preschool, and community. The assessment is conducted using 73 items divided into two domains: child participation (28 items) and environmental impacts (45 items). The measure provides scores for participation frequency, involvement, and environmental support. The YC-PEM was chosen to evaluate the real-world, ecological impact of the locomotor intervention beyond laboratory-based motor tasks.

The structured locomotor training protocol was administered over four consecutive weeks, four days per week (Monday, Tuesday, Thursday, and Friday), with 40 minutes per day. This schedule provided a total direct intervention dose of 16 sessions and 640 minutes of targeted training. The protocol comprised a core set of functional, task-oriented exercises designed to facilitate the independent gait acquisition. For instance, it included weight-bearing, postural control, balance, obstacle negotiation, and the specific training of repetitive locomotion exercises in a progressively challenging way. To mitigate habituation, promote motor learning, and maintain engagement, the exercise sequence was intentionally randomized (Table **2**) between sessions conducted by the researcher and parents or caregivers (Figure **2**).

In advance of the intervention phase, parents and/or caregivers underwent a standardised training session conducted by the researcher. This included

Table 2: Gait Training Protocol Implemented by the Researcher and Parents or Caregivers

	Monday	Tuesday	Thursday	Friday	Duration
2nd week	a) treadmill gait training	a) gait training with obstacles	a) task-oriented training	a) task-oriented training	40 min
	b) task-oriented training	b) task-oriented training	b) gait training with obstacles	b) gait training with obstacles	
	c) gait training with obstacles				
3rd week	a) gait training with obstacles	a) task-oriented training	a) gait training with obstacles	a) gait training with obstacles	40 min
	b) treadmill gait training	b) gait training with obstacles	b) task-oriented training	b) task-oriented training	
	c) task-oriented training				
4th week	a) task-oriented training	a) gait training with obstacles	a) task-oriented training	a) task-oriented training	40 min
	b) gait training with obstacles	b) task-oriented training	b) gait training with obstacles	b) gait training with obstacles	
	c) treadmill gait training				
5th week	a) task-oriented training	a) task-oriented training	a) gait training with obstacles	a) gait training with obstacles	40 min
	b) treadmill gait training	b) gait training with obstacles	b) task-oriented training	b) task-oriented training	
	c) gait training with obstacles				

Legend: The table presents the exercises executed in each session of the structured locomotor training protocol. It was applied by the researcher on Mondays and Thursdays, and applied by the parents or caregivers on Tuesdays and Fridays. In the table, the abbreviations indicate the respective weeks of the protocol: 2nd: Second; 3rd: Third; 4th: Fourth; 5th: Fifth; min: minutes.

Source: Authors.

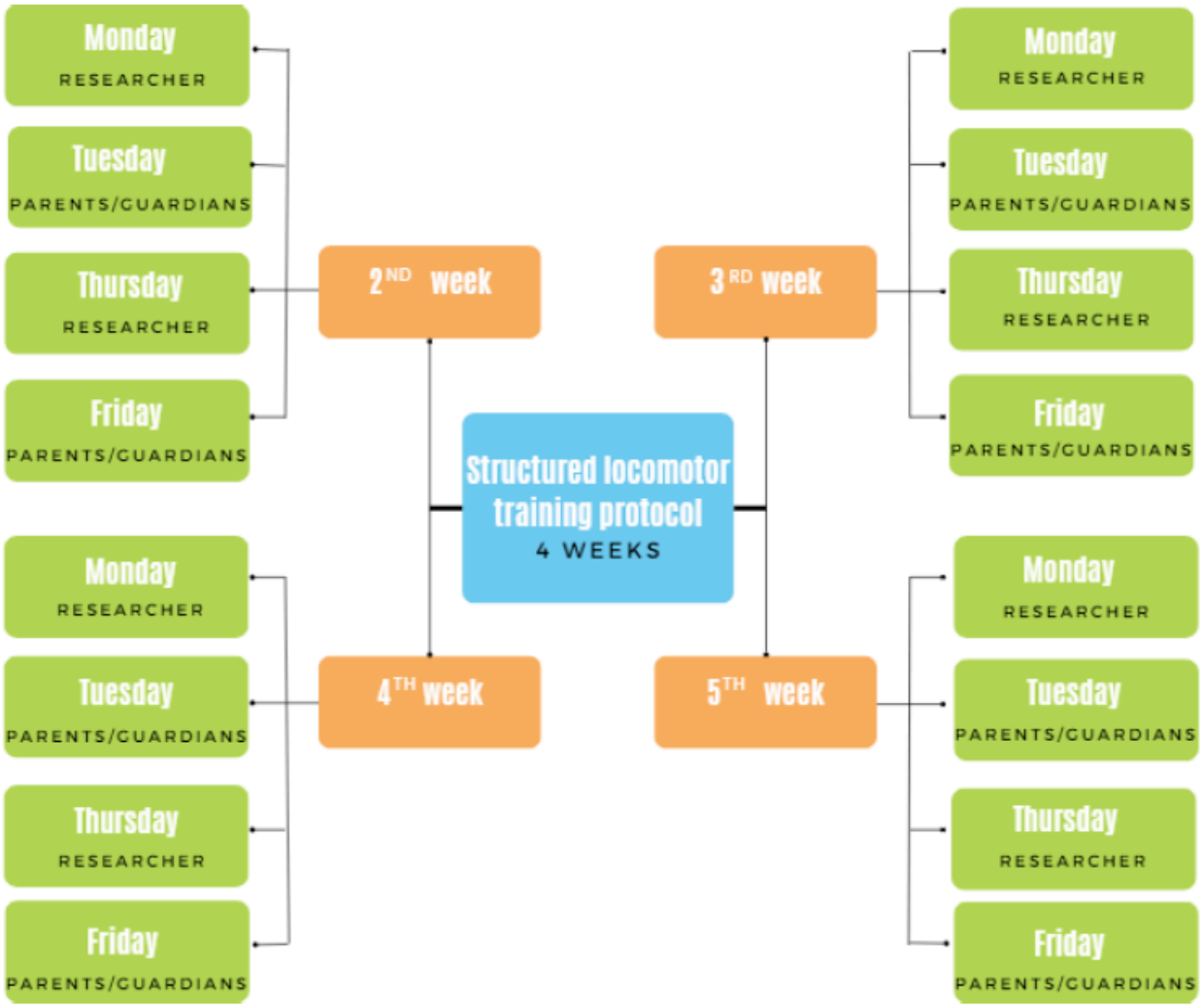

Figure 2: Flowchart of the schedule and administration of the structured locomotor training protocol.

Legend = The structured locomotor training protocol was delivered four days per week (Monday, Tuesday, Thursday, and Friday) for four weeks. The structured locomotor training protocol was administered by the research physiotherapist and the child's parents/caregivers. The researcher delivered the protocol on Mondays and Thursdays; parents/caregivers delivered it on Tuesdays and Fridays under continued researcher supervision. Each session lasted approximately 40 minutes. 2nd: Second; 3rd: Third; 4th: Fourth; 5th: Fifth;

Source = Authors.

direct instruction, hands-on practice, clarifications, and provision of a detailed printed manual with photographic illustrations of all exercises (Appendix A). Additionally, the researcher supplied the materials for home-based sessions (*e.g.*, mats, low obstacles). Following each caregiver-led session, parents should complete a standardised fidelity and feedback form. It documented the day, session duration, child's behavioural state, and any challenges encountered, enabling remote monitoring of adherence.

The structured locomotor training protocol consisted of three core, evidence-based exercises designed to target specific biomechanical and functional aspects of gait. First, the treadmill gait training aimed to provide high repetitions of stereotypic stepping patterns. It was conducted on a standard paediatric treadmill for eight minutes per session. The protocol followed an interval-based structure: 1 minute of active walking followed by 1 minute of rest (total of 4 cycles). The treadmill speed was set to a constant speed of 1.0 m/s, without inclination [19]. To enhance proprioceptive input and promote muscle activation, adjustable ankle weights were applied, corresponding to one-third of the estimated lower limb mass. This load was determined based on a validated anthropometric model based on limb length, circumference, foot width, body mass, and age [20]. Age-appropriate playful stimuli (*e.g.*, toys, music, and short videos) were incorporated to maintain the child's motivation and adherence to the protocol [19]. Second, the task-oriented over-ground gait training compounds functional ambulation in a real environment [21]. It was performed in a 30 m-long by 6 m-wide corridor, using the child's personal anterior

support walker to provide necessary stability. The session considered the motivation and contextual learning component by introducing a goal-directed walking task (*e.g.*, walking to a target, person, or toy). The third exercise consisted of obstacle negotiation training. This component focused on adaptive gait, balance, and selective lower-limb joint flexion. Training occurred within parallel bars for safety. Under them, a customised textured mat was constructed from closed-cell EVA foam, onto which flexible expanded polyethylene tubes ('spaghetti') were securely attached as low-height, compliant obstacles [22] to promote hip and knee flexion. The child was encouraged to step over these obstacles with a playful design of activities and varied placement of the tubes, serving to increase motivation, attentional focus, and the functional challenge of the task.

RESULTS

Regarding the primary objective of this study, key functional and behavioural milestones were observed. Most notably, by the conclusion of the intervention period, the child achieved the functional criterion of walking further than 10 consecutive independent steps. This represented a significant advance from the pre-intervention status. Parental forms and researcher notes documented a shift in the child's engagement across the intervention phase. Initial sessions were characterised by reports of low cooperation and signs of rapid physical fatigue. By the final week, both sources reported the child to be consistently more participatory, focused, and physically tolerant of the full 40-minute session duration.

The motor behavior, as measured by the IMP, is summarized in Figure **3**. The IMP total score increased from a baseline mean of 78.4% (SD 1.3) to a post-intervention mean of 80.3% (SD 1.3), representing a 1.9 percentage point improvement. Domain-specific scores demonstrated distinct trajectories over the study. Performance scores were consistently high, ranging from a baseline mean of 84% (SD 2.0) to 88%, with a final score of 85.7% (SD 2.7). The fluency domain increased from a baseline of 70% to a mid-intervention peak of 75% prior to returning to 70% at the final assessment. For symmetry, the participant started and finished with the highest score possible, 100%. Adaptability domain presented a baseline mean of 59.6% (SD 0.6), a peak of 68%, and a post-intervention mean of 64.3% (SD 0.7). Finally, the variation domain also presented a minor fluctuation, with an initial baseline mean of 77.3% (SD 2.5), a peak of 90%, and an end with a baseline mean of 81.6% (2.66).

The performance scores for upright mobility items assessed at standing and walking postures of the IMP are detailed in Figure **4**, which provides a granular

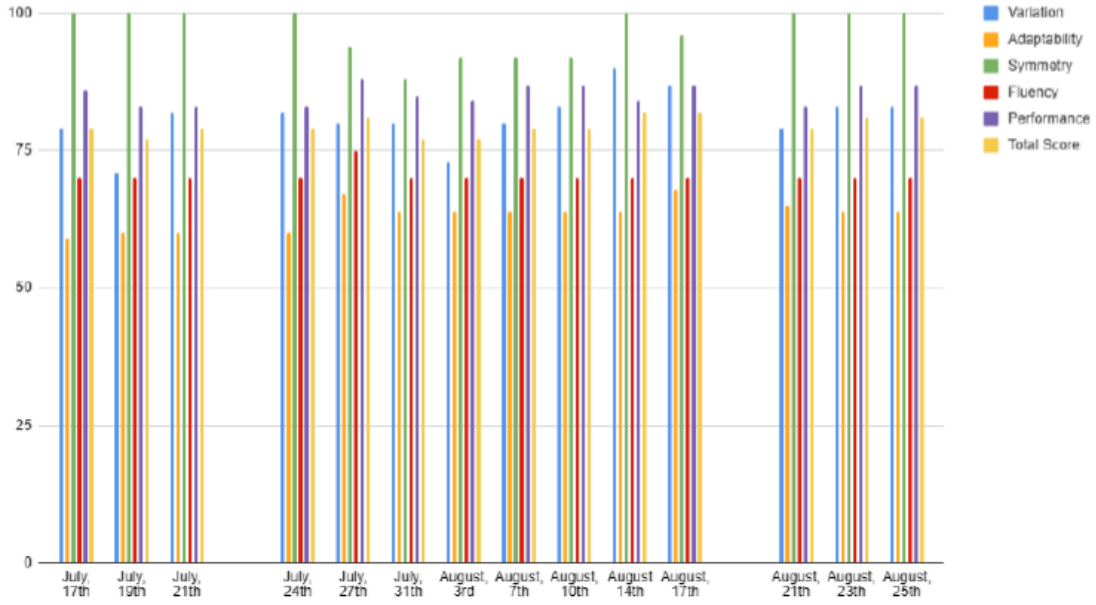

Figure 3: Infant Motor Profile (IMP) domains scores and total score.

Legend = Line graph depicting the total IMP scores (yellow), and the five domain scores - variation (blue), adaptability (orange), symmetry (green), fluency (red), performance (purple) - across all assessment timepoints. The Y-axis represents the IMP score (as a percentage for all the domains and the total score). The X-axis represents the chronological timeline of assessments, grouped into the three study phases: A - baseline pre-intervention - week 1; B - assessments performed during the intervention period - weeks 2-5; A - baseline post-intervention - week 6. Vertical shaded brands separate the phases.

Source = Authors.

Figure 4: Performance scores while standing and walking.

Legend = This graph represents the performance scores while standing and walking posture of the Infant Motor Profile (IMP).

Line graph depicting the performance scores while standing and walking posture of Infant Motor Profile (IMP). The items are presented as blue for 'stands up', orange for 'standing up', green for 'walking', red for 'balance while independently walking', purple for 'arm posture and movements while independently walking', and yellow for 'heel-to-toe gait while independent walking'. The Y-axis represents the scores on the performance domain of the IMP (as a percentage). The X-axis represents the chronological timeline of assessments, grouped into the three study phases: A - baseline pre-intervention - week 1; B - assessments performed during the intervention period - weeks 2-5; A - baseline post-intervention - week 6. Vertical shaded brands separate the phases.

Source = Authors.

analysis of standing and walking skills. At the 'standing ability', the child consistently scored 4, indicating the ability to "stand independently for more than 10 seconds, but rotates trunk to a minimal extent only". In the item 'standing up', the child consistently scored 3, demonstrating the ability to "stand up independently with the use of *e.g.*, furniture". As gait parameters, the item 'walking' was progressively scored from 3 to 4 ("walks independently") during the protocol, confirming the attainment of the primary functional goal of this study. The item 'balance while walking independently' reflects the dynamic balance of the child, who scored 2, indicating "moderate balance capacities". The item 'heel-toe gait while walking independently' was predominantly scored as 1, reflecting "no or only occasionally heel-toe gait". Finally, the item 'posture and movements of upper extremities while walking independently: presence of asymmetry', showed a clear score improvement from 1 to 2, indicating a reduction in asymmetric arm posture during gait.

Furthermore, a detailed qualitative analysis of the IMP's variation and adaptability domains for standing and walking posture revealed measurable changes. For instance, at the 'variation in standing up behaviour' item, the child presented an increased score from insufficient (score 1) to sufficient variation (score 2); as well as an upgrade from no adaptive selection (score 1) to adaptive selection (score 2). At the item 'variation in movements of arms and hands while walking independently', the child presented an improvement from score 0 (does not walk independently or walks independently with arms consistently in high or semi-high guard) to 1 (insufficient variation); as did the adaptability domain, which rose from score 0 to score 1 (no adaptive selection). This indicates that although the child did not present a variable and adapted arm movements while walking independently, he can now walk independently.

The child's participation through the YC-PEM questionnaires presented a notable progression in regard to the perceived involvement and parental desires, as shown in the pre- and post-intervention data (Table **3**). The home environment setting presented a significant shift in specific activity types and parental satisfaction. For the activity 'indoor games and play activities' and 'arts, crafts, music and hobbies',

Table 3: Results of the Young Children Participation and Environment Measure (YC-PEM), Home Participation Section

Home participation	Baseline	Post-intervention
Computer and console games	A few times a week, minimally involved. Would like to be involved in a greater variety of activities.	A few times a month, somewhat involved. Yes, would like to do it more often.
Indoor games and play	Daily, very involved. Would like to be involved in a greater variety of activities.	Daily, very involved. Do not wish any change.
Arts, crafts, music, and hobbies	A few times a week, somewhat involved. Would like to be involved in a greater variety of activities.	Daily, very involved. Do not wish for any change.
Watching TV, videos, and DVDs	Daily, very involved. Would like to do it less often.	Daily, very involved. Would like to do it less often.
Being with other people	A few times a week, somewhat involved. Would like to do it more often.	Several times a week, somewhat involved. I do not wish to change.
Socializing using technology	A few times a month, minimally involved. Would like to do it more often.	Several times a week, somewhat involved. I would like to do it more often.
Household chores	Never. Would like to be more involved.	Never. I would like to be more involved.
Personal care	A few times a week, somewhat involved. Would like to be more involved.	Daily, somewhat involved. I would like to do it more often.
Getting ready for school	A few times a week, minimally involved. Would like to be more involved.	Daily, somewhat involved. I would like to do it more often.
Homework	A few times a week, somewhat involved. Would like to be more involved.	Several times a week, highly involved. I would like to be involved more often.

Legend: The table represents the home participation topics assessed by the YC-PEM; the columns indicate the results at baseline and after the intervention protocol, based on the mother's responses.

Source: Authors.

the mothers' initial desire was "to be more involved in a greater variety of activities". At post-intervention, this desire was masked as "none," indicating satisfaction with these components. Similarly, for the topic 'the child being with other people', at baseline, the mothers' desire was "I would like to do it more often". At the end of the protocol, it has been shifted to "does not want changes", also reflecting the family's satisfaction with the child's social participation.

The results from YC-PEM showed modifications in the strategies used to help the child successfully participate in activities at home. At baseline, the mother reported employing three primary strategies, all focused on physical structuring and directive prompting within daily care routines, such as 1) providing postural support during bathing (*e.g.*, "giving a bath while seated in a chair") and requesting the child1s active assistance (*e.g.*, "asking the child to help by stretching their lower limbs to be cleaned"); 2) providing postural support during dressing (*e.g.*, "changing clothes while leaning against the wall or sitting down") and requesting motor assistance with limb movement (*e.g.* "asking the child to help with their upper and lower limbs to put on clothes"); 3) using direct verbal commands to guide and motivate task competition (*e.g.*, "using voice commands to assist and motivate in daily tasks").

Post-intervention strategies were reported involving to emphasise dialogue, explanation, and shared decision-making, for instance 1) increasing conversational dialogue (*e.g.*, "adding more dialogue to daily routines to encourage the child's participation in household conversations"); 2) providing explanatory language (*e.g.*, "adding more explanations on how to perform tasks to facilitate the child's understanding and inclusion in tasks and activities"); and 3) incorporating direct questions (*e.g.*, "adding questions to the child daily to encourage their participation in decision-making") (Table **4**).

Participation in the school setting showed clear improvements. The reported frequency of involvement in "classroom activities" increased from "sometimes" at baseline to "daily" post-intervention. Concurrently, a systematic expansion in the number and diversity of support strategies was reported. At baseline, three core strategies were applied: 1) parental assistance with school activities; 2) parental help with homework; and 3) educator professionals' support for peer communication.

Post-intervention, this support framework was enhanced through the implementation of additional strategies, such as: 1) the formal appointment of an educational professional exclusively assigned to assist and support the child's participation in classroom

Table 4: Results of the Young Children Participation and Environment Measure (YC-PEM), strategies that facilitate the child's participation at home

Strategies at Baseline	Strategies after the Intervention Protocol
Giving a bath while seated in a chair and asking the child to assist by extending the lower limbs for cleaning	Increased daily dialogue to encourage the child's participation in household conversations
Changing clothes while leaning against the wall or seated, asking the child to help with the upper and lower limbs to assist in dressing.	Provided more explanations on how to perform tasks to facilitate the child's understanding and inclusion in activities and chores
Verbal commands to assist and motivate during daily tasks	Incorporated daily questioning to encourage the child's participation in decision-making

Legend: The table represents the strategies used by parents/caregivers to facilitate the child's successful participation in home activities; the first column shows the strategies used at baseline, and the second column shows the strategies used after the intervention protocol.

Source: Authors.

activities; 2) active mediation by this professional to scaffold and promote peer interaction and communication; and 3) the establishment of structured communication channels between parents and the educational assistant to align support strategies and inform decision-making aiming to optimize developmental outcomes.

Increased participation frequency was also reported in the community setting. Engagement in unstructured outdoor physical activities and play with community peers increased from "once every 4 months" at baseline to "once a month" at post-intervention. The mother reported greater personal confidence in facilitating the child's community involvement and perceived fewer environmental barriers to participation.

Collectively, the YC-PEM data demonstrated functional gains across all the settings - home, school, and community. These changes encompassed increased activity frequency, greater parental satisfaction, an evolution towards more autonomy-supportive strategies at home, a more structured support system at school, and reduced perceived barriers in the community, modifying social and contextual dimensions of participation.

DISCUSSION

This single-case experimental study evaluated a 6-week structured, intensive locomotor training protocol for a young child with Joubert syndrome. The primary functional goal - the acquisition of independent walking for more than 10 consecutive steps - was successfully achieved, representing a meaningful clinical milestone. This functional gain was accompanied by positive changes in participation across home, school, and community settings, as reported on the YC-PEM. Notably, these functional and participatory improvements occurred without a significant change in the total quantitative score of the Infant Motor Profile (IMP). The following discussion will interpret, explore the clinical implications of the findings, and situate the results within the broader context of neurorehabilitation for children with cerebellar disorders.

Our findings support the application of intensive, task-specific locomotor training principles to children with complex cerebellar ataxia. The observed functional gain aligns with evidence for treadmill training in other paediatric populations. For instance, Ulrich and collaborators [19] reported improved gait and posture in 30 infants with Down syndrome following a high-frequency regimen of a treadmill gait training, in 8 minutes, five times a week. While our protocol employed a lower weekly frequency, once weekly, but within a multi-component program, it similarly culminated in a critical functional milestone - the child's independent locomotion. This suggests that dose distribution - integrating treadmill work within a broader regimen of varied, functional tasks - may be an effective strategy for children with JS, for whom high-frequency, single-modality training may be less tolerable.

Our results contribute to a nascent evidence base supporting the efficacy of targeted rehabilitation for JS. A prior case report documented substantial motor and functional gains in a 9-year-old with JS following a two-year, multidisciplinary program, as measured by the Gross Motor Function Measure (GMFM) and Functional Independence Measure for Children (WeeFIM) [23]. While that intervention differed significantly from ours in duration and therapeutic scope, a fundamental commonality is the application of structured, repetitive, and goal-directed therapeutic principles.

Furthermore, the over-ground, task-oriented components of our protocol on a 30-meter corridor and community with a walker support are congruent with

established best practice in paediatric neuromotor rehabilitation. Evidence strongly indicates that task-oriented training is a cornerstone of effective intervention for children with cerebral palsy [21, 24], promoting skill acquisition in ecologically valid contexts. Our protocol extended this principle by systematically progressing from a controlled corridor to community settings, thereby facilitating the generalisation of walking skills into daily life, an outcome reflected in the YC-PEM community participation data.

Therefore, the significant improvements in participation reported on the YC-PEM may be considered as a primary finding of this study. This success is likely multifactorial, with a key element being the deliberate inclusion of parents as co-therapists. Our dual-administration model empowered caregivers, increasing their competence and confidence in supporting their child's mobility in daily contexts. This aligns with evidence underscoring the efficacy of parent-mediated interventions. For instance, a prior single-subject design with two participants also utilising the YC-PEM found that parental assistance in implementing therapeutic activities over 20 weeks led to significant gains in youth participation [25]. Our results reinforce that active caregiver involvement is a potent mechanism for enhancing the ecological impact and carry-over of clinic-based therapies into home, school, and community life.

Nonetheless, the child's behavioural profile - characterised by stereotypies, self-regulation challenges, and self-injurious behaviour - likely influenced assessment performance. Engaging in the structured, demand-based tasks of the IMP requires sustained attention and compliance, which can be significantly compromised by these co-occurring behaviours. This challenge is not unique to JS; research on children with autism spectrum disorder notes that standardised motor assessments can underestimate ability due to behavioural and attentional confounding factors, rather than motor capacity per see [26]. Therefore the stable IMP scores may reflect assessment limitations and behavioural state more accurately than a true absence of neuromotor change, highlighting the critical need for functionally referenced outcomes (like step count) alongside standardised tools when evaluating children with complex neurodevelopmental profiles.

CONCLUSION

This single-case experimental study demonstrates that a time-limited, structured, and intensive locomotor training protocol can yield clinically meaningful outcomes for a young child with Joubert syndrome. The achievement of independent walking for more than 10 consecutive steps, coupled with positive changes in participation across home, school, and community settings, underscores the functional and ecological validity of this targeted intervention. The active inclusion of parents as intervention agents appears to have been a critical factor in facilitating skill generalisation and enhancing family-centered outcomes. While this study provides preliminary evidence of feasibility and benefit, its single-subject design limits generalisability. Future research should employ controlled group designs with larger samples to establish efficacy. Beyond, it should consider other approaches focusing on the behavioral alterations that may be associated with the syndrome.

ACKNOWLEDGMENTS

The authors extend their sincere gratitude to the child and family whose participation made this study possible. The authors also thank the staff and administration of the Rehabilitation Center of Santa Casa de Misericórdia de Porto Feliz - Porto Feliz, São Paulo, Brazil - for their invaluable collaboration and provision of facilities.

DECLARATION OF CONFLICT OF INTEREST

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

FUNDING STATEMENT

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

ANNEX A. HUMAN RESEARCH ETHICS COMMITTEE OF THE FEDERAL UNIVERSITY OF SÃO CARLOS APPROVAL

UNIVERSIDADE FEDERAL DE SÃO CARLOS - UFSCAR

PARECER CONSUBSTANCIADO DO CEP

DADOS DO PROJETO DE PESQUISA

Título da Pesquisa: TREINO DE MARCHA EM UMA CRIANÇA COM SÍNDROME DE JOUBERT: Estudo de caso

Pesquisador: CAROLINE TELES

Área Temática:

Versão: 2

CAAE: 71592723.3.0000.5504

Instituição Proponente: Departamento de Fisioterapia

Patrocinador Principal: Financiamento Próprio

DADOS DO PARECER

Número do Parecer: 6.454.170

Apresentação do Projeto:

As informações elencadas nos campos "Apresentação do Projeto", "Objetivo da Pesquisa" e Avaliação dos Riscos e Benefícios" foram extraídas do arquivo Informações Básicas da Pesquisa (PB_INFORMAÇÕES_BÁSICAS_DO_PROJETO_2152272.pdf, de 04/10/2023) e/ou do Projeto Detalhado (postado em 04/10/2023): RESUMO, HIPÓTESE (se houver), METODOLOGIA, CRITÉRIOS DE INCLUSÃO E EXCLUSÃO.

Objetivo da Pesquisa:

Objetivo Primário:

Verificar a efetividade de um protocolo de treino de marcha na aquisição da marcha independente por aproximadamente 10 passos, em uma criança com síndrome de Joubert.

Avaliação dos Riscos e Benefícios:

Riscos:

Risco de queda da própria altura, risco de quedas nos obstáculos do treino de marcha no tapete. A aplicação do protocolo será constantemente supervisionada e ocorrerá em um local seguro, sem mobílias próximas e em uma superfície estável.

Benefícios:

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Participando deste estudo estarei auxiliando no fortalecimento dos músculos dos MMII e independência na locomoção. O participante e família estarão promovendo maior conhecimento acerca da condição de saúde. Ganharão atendimento fisioterapêutico especializado, orientado à tarefa e com grandes chances de apresentar resultados positivos e significativos.

Comentários e Considerações sobre a Pesquisa:
Trata-se de uma pesquisa que deve seguir os preceitos éticos estabelecidos pela Resolução CNS nº 466/2012 suas complementares.

Considerações sobre os Termos de apresentação obrigatória:
Apresentada anuência da Santa Casa de Misericórdia de Porto Feliz anexada junto ao documento projeto de pesquisa

Recomendações:
Vide campo "Conclusões ou Pendências e Lista de Inadequações"

Conclusões ou Pendências e Lista de Inadequações:
ANÁLISE DAS PENDÊNCIAS CONFORME CARTA RESPOSTAS E DOCUMENTOS TCLE E PROJETO ANEXADOS NA PB EM 04/10/2023

1. Apresentar o TCLE em forma de convite AO RESPONSÁVEL LEGAL DA CRIANÇA. Segundo a resolução 466 de 2012: IV – DO PROCESSO DE CONSENTIMENTO LIVRE E ESCLARECIDO
O respeito devido à dignidade humana exige que toda pesquisa se processe com consentimento livre e esclarecido dos participantes, indivíduos ou grupos que, por si e/ou por seus representantes legais, manifestem a sua anuência à participação na pesquisa. Entende-se por Processo de Consentimento Livre e Esclarecido todas as etapas a serem necessariamente observadas para que o convidado a participar de uma pesquisa possa se manifestar, de forma autônoma, consciente, livre e esclarecida.
Resposta à pendência 1: O documento TCLE foi editado para forma de convite e está grifado em amarelo para destacar a alteração realizada no documento.

ANÁLISE DO CEP: PENDÊNCIA ATENDIDA

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UNIVERSIDADE FEDERAL DE SÃO CARLOS - UFSCAR

Continuação do Parecer: 6.454.170

2. Adequar TCLE conforme prevê a resolução 466 de 2012: IV.3 - O Termo de Consentimento Livre e Esclarecido deverá conter, obrigatoriamente:
a) justificativa, os objetivos e os procedimentos que serão utilizados na pesquisa, com o detalhamento dos métodos a serem utilizados, informando a possibilidade de inclusão em grupo controle ou experimental, quando aplicável; b) explicitação dos possíveis desconfortos e riscos decorrentes da participação na pesquisa, além dos benefícios esperados dessa participação e apresentação das providências e cautelas a se em empregadas para evitar e/ou reduzir efeitos e condições adversas que possam causar dano, considerando características e contexto do participante da pesquisa;
c) esclarecimento sobre a forma de acompanhamento e assistência a que terão direito os participantes da pesquisa, inclusive considerando benefícios e acompanhamentos posteriores ao encerramento e/ ou a interrupção da pesquisa;
f) garantia de que o participante da pesquisa receberá uma via do Termo de Consentimento Livre e Esclarecido;
g) explicitação da garantia de ressarcimento e como serão cobertas as despesas tidas pelos participantes da pesquisa e dela decorrentes; e
h) explicitação da garantia de indenização diante de eventuais danos decorrentes da pesquisa

Respostas à pendência 2:
a) a justificativa foi acrescentada no termo de consentimento livre e esclarecido (TCLE) na página 1, linhas 26 a 30. Este estudo é imprescindível ao passo que auxiliará na verificação da eficácia de um protocolo de treino para aquisição da marcha independente em crianças com síndrome de Joubert. Ao comprovar a eficácia do mesmo, poderemos auxiliar no tratamento de outros indivíduos com a síndrome de Joubert, além de proporcionar maior visibilidade acerca da condição de saúde.

b) os possíveis desconfortos e riscos decorrentes da participação na pesquisa, foram incluídos e estão especificados na página 3, linha 29 e 30 e na página 4, linhas 1 a 8. Durante a aplicação do estudo podem ocorrer risco de queda da própria altura, risco de quedas nos obstáculos do treino de marcha no tapete. A aplicação do protocolo será constantemente supervisionada e ocorrerá em um local seguro, sem

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Continuação do Parecer: 6.454.170

mobílias próximas e em uma superfície estável.
Seu (sua) filho (a) será beneficiado (a) participando deste estudo sendo auxiliado (a) no fortalecimento dos músculos dos MMII e independência na locomoção. O (a) participante e família estarão promovendo maior conhecimento acerca da condição de saúde. Ganharão atendimento fisioterapêutico especializado, orientado à tarefa e com grandes chances de apresentar resultados positivos e significativos.

c) Acrescentado parágrafo sobre esclarecimento sobre a forma de acompanhamento e assistência dos participantes na página 3, linha 25 a 28. O participante será acompanhado por um profissional da saúde com graduação na área da fisioterapia, e será respaldado para orientações e se necessário acompanhamentos por meio de avaliação e atendimento posteriores ao encerramento e/ou a interrupção da pesquisa.

f) a garantia que o participante receberá uma via do Termo de Consentimento Livre e Esclarecido, foram incluídos na página 4, linhas 14 e 19. O (a) senhor (a) receberá uma via deste termo, rubricada em todas as páginas por você e pela pesquisadora, onde consta o telefone e o endereço da pesquisadora principal com quem você poderá tirar suas dúvidas sobre a pesquisa e da participação do (a) seu (sua) filho (a) agora ou a qualquer momento.

g) Sobre a explicitação da garantia de ressarcimento e como serão cobertas as despesas tidas pelos participantes da pesquisa e dela decorrentes, foram incluídos e especificados na página 4, linhas 9 a 12. O (a) senhor (a) não terá nenhum custo ou compensação financeira ao participar do estudo. Entretanto, todas as despesas com o transporte e a alimentação decorrentes da sua participação na pesquisa, quando for o caso, serão ressarcidas no dia da coleta.

h) a explicitação da garantia de indenização diante de eventuais danos decorrentes da pesquisa, foram incluídos e especificados na página 4, linhas 12 e 13. Você terá direito a indenização por qualquer tipo de dano resultante da sua participação na pesquisa.

ANÁLISE DO CEP: PENDÊNCIA ATENDIDA

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Continuação do Parecer: 6.454.170

3. O recrutamento foi realizado por conveniência, uma vez que a pesquisadora possui contato profissional com o paciente em questão e com a responsável pelo mesmo.
PELO TEXTO A PESQUISA JÁ FOI INICIADA E HOUVE RECRUTAMENTO. ESCLARECER ESTE PONTO
Resposta à pendência 3: Já realizamos o recrutamento, mas não qualquer intervenção. Isto se deve ao fato que sou a terapeuta deste paciente há alguns meses no centro de reabilitação, adicionalmente se trata de uma síndrome rara, portanto, seria muito difícil encontrar mais crianças com o mesmo diagnóstico clínico e quadro funcional. Ademais, o desenvolvimento deste estudo se deu pela necessidade que senti de mais relatos sobre a síndrome. Retomo ainda que a coleta de dados para o estudo não foi iniciada, estou aguardando a liberação do comitê de ética para poder realiza-lo.

ANÁLISE DO CEP: PENDÊNCIA ATENDIDA

4. Local de coleta de dados: As avaliações serão realizadas no ambiente domiciliar em que a criança reside e no Centro de Reabilitação da Santa Casa de Misericórdia
de Porto Feliz.
COMO ENVOLVE OUTRA INSTITUIÇÃO E UM RESPONSÁVEL LEGAL, É NECESSÁRIO APRESENTAR ANUÊNCIA DA INSTITUIÇÃO COPARTICIPANTE PARA PARTICIPAR DO ESTUDO. ALÉM DISSO, SE ESTA INSTITUIÇÃO (Santa Casa de Misericórdia de Porto Feliz) POSSUIR CEP, SE FAZ NECESSÁRIA A INCLUSÃO DESTA INSTITUIÇÃO COMO COPARTICIPANTE NA PLATAFORMA BRASIL.
Resposta à pendência 4: A anuência da instituição coparticipante foi assinada e carimbada, adicionalmente, a instituição Santa Casa de Misericórdia de Porto Feliz não possui um Comitê de Ética (CEP) específico para o seu funcionamento, o documento foi incluído no projeto de pesquisa na página 40.

ANÁLISE DO CEP: PENDÊNCIA ATENDIDA

5. NA DESCRIÇÃO DA METODOLOGIA NO TCLE CONSTA EM EM TODO O TCLE:
'O protocolo de treino será realizado em quatro dias da semana, sendo destes, dois dias (segundas e quintas-feiras) realizados pela pesquisadora e dois dias (terças e sextas-feiras) pelos pais/responsáveis da criança. A sessão do protocolo de treino de marcha terá duração de aproximadamente 40 minutos e os exercícios serão realizados conforme descrito na tabela 1 e 2. A ordem dos exercícios será alterada a fim de contemplar a prática

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Continuação do Parecer: 6.454.170

aleatória randomizada proposta para este protocolo de treino'.
É PRECISO, NESTE TRECHO EM TODO O TCLE APRESENTAR DETALHES DA METODOLOGIA E NÃO CITAR ANEXOS, QUE NÃO PODEM CONSTAR NO TERMO, E DESCREVE EM LINGUAGEM ACESSÍVEL AO RESPONSÁVEL LEGAL PELA CRIANÇA.
Resposta à pendência 5: Obrigada, foi retirado do texto do documento TCLE a citação de anexos.
Documento editado:
O protocolo de treino será realizado em quatro dias da semana, sendo destes, dois dias (segundas e quinta-feiras) realizados pela pesquisadora e dois dias (terças e sexta-feiras) pelos pais/responsáveis da criança. A sessão do protocolo de treino de marcha terá duração de aproximadamente 40 minutos. A ordem dos exercícios será alterada a fim de contemplar a prática aleatória randomizada proposta para este protocolo de treino.

ANÁLISE DO CEP: PENDÊNCIA ATENDIDA

Considerações Finais a critério do CEP:

Diante do exposto, o Comitê de ética em pesquisa - CEP, de acordo com as atribuições definidas na Resolução CNS nº 466 de 2012 e 510 de 2016, manifesta-se por considerar "Aprovado" o projeto. A responsabilidade do pesquisador é indelegável e indeclinável e compreende os aspectos éticos e legais, cabendo-lhe, após aprovação deste Comitê de Ética em Pesquisa: II - conduzir o processo de Consentimento e de Assentimento Livre e Esclarecido; III - apresentar dados solicitados pelo CEP ou pela CONEP a qualquer momento; IV - manter os dados da pesquisa em arquivo, físico ou digital, sob sua guarda e responsabilidade, por um período mínimo de 5 (cinco) anos após o término da pesquisa; V - apresentar no relatório final que o projeto foi desenvolvido conforme delineado, justificando, quando ocorridas, a sua mudança ou interrupção. Este relatório final deverá ser protocolado via notificação na Plataforma Brasil. OBSERVAÇÃO: Nos documentos encaminhados por Notificação NÃO DEVE constar alteração no conteúdo do projeto. Caso o projeto tenha sofrido alterações, o pesquisador deverá submeter uma "EMENDA".

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Continuação do Parecer: 6.454.170

Este parecer foi elaborado baseado nos documentos abaixo relacionados:

Tipo Documento	Arquivo	Postagem	Autor	Situação
Informações Básicas do Projeto	PB_INFORMAÇÕES_BÁSICAS_DO_PROJETO_2152272.pdf	04/10/2023 13:30:39		Aceito
Projeto Detalhado / Brochura Investigador	Projeto_de_pesquisa.docx	04/10/2023 13:28:39	CAROLINE TELES	Aceito
Projeto Detalhado / Brochura Investigador	Projeto_de_pesquisa.pdf	04/10/2023 13:28:27	CAROLINE TELES	Aceito
Outros	Carta_Resposta_versaoX.docx	04/10/2023 13:28:11	CAROLINE TELES	Aceito
Outros	Carta_Resposta_versaoX.pdf	04/10/2023 13:27:56	CAROLINE TELES	Aceito
TCLE / Termos de Assentimento / Justificativa de Ausência	TCLE.docx	04/10/2023 13:27:35	CAROLINE TELES	Aceito
TCLE / Termos de Assentimento / Justificativa de Ausência	TCLE.pdf	04/10/2023 13:27:28	CAROLINE TELES	Aceito
Folha de Rosto	folhaDeRosto_carol_assinado.pdf	21/07/2023 16:51:06	CAROLINE TELES	Aceito

Situação do Parecer:
Aprovado

Necessita Apreciação da CONEP:
Não

SAO CARLOS, 24 de Outubro de 2023

Assinado por:
Sonia Regina Zerbetto
(Coordenador(a))

Endereço: WASHINGTON LUIZ KM 235
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ANNEX B. PARENTS' CONSENT OBTAINED THROUGH THE SIGNATURE ON THE FREE AND INFORMED CONSENT FORM.

APÊNDICE 1 – TERMO DE CONSENTIMENTO LIVRE E ESCLARECIDO (TCLE)

(Consentimento Pós-informação para Pesquisa com Seres Humanos)

LABORATÓRIO DE PESQUISAS EM ANÁLISE DO MOVIMENTO (LAPAM) E LABORATÓRIO DE AVALIAÇÃO DO DESENVOLVIMENTO FUNCIONAL (LADeF) - Departamento de Fisioterapia, Universidade Federal de São Carlos

Consentimento formal da participação no estudo intitulado "TREINO DE MARCHA EM UMA CRIANÇA COM SÍNDROME DE JOUBERT: Estudo de caso".

Responsável: Caroline Teles

Orientadora: Profª. Drª. Luiza Ribeiro Machado

Eu, Suzana Paula Miranda Moreira, portador (a) do RG nº 43 661 620-1, residente a Rua Paulino de Bernardes, nº- 121, bairro: Cidade Jardim na cidade de - Porto Feliz, telefone (11) 952472134, responsável pelo (a) menor: Isaac Miranda Moreira, autorizo a participação do meu (minha) filho (a) na pesquisa intitulada "TREINO DE MARCHA EM UMA CRIANÇA COM SÍNDROME DE JOUBERT: Estudo de caso" conduzida por Caroline Teles, sob orientação da professora Drª. Luiza Ribeiro Machado.

Objetivo do estudo:

Verificar a eficácia de um protocolo de treino de marcha na aquisição da marcha independente por aproximadamente 10 passos, em uma criança com síndrome de Joubert.

Explicação do procedimento:

A avaliação ocorrerá em ambiente domiciliar e no Centro de Reabilitação da Santa Casa de Misericórdia de Porto Feliz na presença dos pais/responsáveis. Primeiramente serão retomados os objetivos e esclarecidas as dúvidas dos pais/responsáveis referentes ao estudo. A assinatura do Termo de Consentimento Livre e Esclarecido (TCLE) será coletada pela pesquisadora. A avaliação será agendada respeitando o intervalo de uma hora após o período de alimentação da criança. Essa deve se encontrar em alerta 3 ou 4 segundo a Escala Comportamental de Prechtl e Beintema (1964). O agendamento da avaliação também não deverá coincidir com os dias de vacinação da criança.

A avaliação será iniciada pela análise do comportamento motor por meio do instrumento IMP. A avaliação do IMP será filmada para a posterior análise e pontuação dos vídeos. A criança será posicionada sobre um tatame de EVA, nas posturas supina, prona, sentada no chão e no colo do cuidador e ortostática, sendo as posturas avaliadas na sequência preferida pela criança. A avaliação prévia a intervenção deverá ser realizada em três momentos, com intervalo de dois dias entre eles, com o intuito de se estabelecer a linha de base da avaliação para o início da aplicação do protocolo de treino de marcha. Em seguida será aplicada por meio de entrevista a escala PEM-CY para avaliar o contexto da participação da criança.

O estudo será aplicado no período de seis semanas. Na 1ª e 6ª semana serão realizadas a avaliação e a reavaliação por meio do IMP para avaliar o comportamento motor (estruturas e funções do corpo, e atividades da criança) e da escala Medida da Participação e do Ambiente - Crianças e Jovens (PEM-CY) para avaliar o contexto da participação da criança. Entre a 2ª e 5ª semana do protocolo, será aplicado o instrumento IMP a cada dia em que a pesquisadora aplicar a intervenção do estudo.

Serão realizados quatro dias de intervenção na semana por quatro semanas, sendo dois destes realizados pela pesquisadora, um no Centro de Reabilitação da Santa Casa de Misericórdia de Porto Feliz e outro no domicílio da criança; os outros dois dias serão realizados em domicílio pelos pais/responsáveis. Serão realizadas 8 reavaliações previamente ao início da intervenção com intervalo de 2 dias entre si dentro da mesma semana. As reavaliações ocorrerão por meio da aplicação do instrumento IMP, e na 6ª semana do protocolo deste estudo, a criança também será reavaliada por meio da escala PEM-CY.

O protocolo de treino será realizado em quatro dias da semana, sendo destes, dois dias (segundas e quinta-feiras) realizados pela pesquisadora e dois dias (terças e sexta-feiras) pelos pais/responsáveis da criança. A sessão do protocolo de treino de marcha terá duração de aproximadamente 40 minutos e os exercícios serão realizados conforme descrito na tabela 1 e 2. A ordem dos exercícios será alterada a fim de contemplar a prática aleatória randomizada proposta para este protocolo de treino.

Benefícios previstos:

Participando deste estudo estarei auxiliando no fortalecimento dos músculos dos MMII e independência na locomoção. O participante e família estarão promovendo maior conhecimento acerca da condição de saúde. Ganharão atendimento fisioterapêutico especializado, orientado à tarefa e com grandes chances de apresentar resultados positivos e significativos.

Potenciais riscos e incômodos:

Risco de queda da própria altura, risco de quedas nos obstáculos do treino de marcha no tapete. A aplicação do protocolo será constantemente supervisionada e ocorrerá em um local seguro, sem mobílias próximas e em uma superfície estável.

Seguro saúde ou de vida:

Eu entendo que não existe nenhum tipo de seguro de saúde ou de vida que possa vir a me beneficiar em função de minha participação neste estudo.

Liberdade de participação:

A minha participação neste estudo é voluntária. É meu direito interromper a participação de meu (minha) filho (a) a qualquer momento sem que isto incorra em qualquer penalidade ou prejuizo. Também entendo que a pesquisadora tem o direito de excluir do estudo o(a) meu (minha) filho(a) a qualquer momento.

Sigilo de identidade:

As informações obtidas nas filmagens deste estudo serão mantidas em sigilo e não poderão ser consultadas por pessoas leigas sem a minha autorização oficial. Estas informações só poderão ser utilizadas para fins estatísticos, cientificos ou didáticos, desde que fique resguardada a minha privacidade.

A responsável por este estudo me explicou das necessidades da pesquisa e se prontificou a responder todas as questões sobre o experimento. Eu estou de acordo com a participação de meu (minha) filho (a) no estudo de livre e espontânea vontade e entendo a relevância dele. Julgo que é meu direito manter uma cópia deste consentimento.

Para questões relacionadas a este estudo, contate:

Caroline Teles

15 981728591

carol_ts11@hotmail.com

R. Irmã Elisa Ambrósio, 16, Jandira Diez Alcalá, Porto Feliz - SP- 18545-062 - Brasil

Declaro que entendi os objetivos, riscos e beneficios da participação de meu (minha) filho(a) na pesquisa e concordo em participar.

A pesquisadora me informou que o projeto foi aprovado pelo Comitê de Ética

em Pesquisa em Seres Humanos da UFSCar que funciona na Pró-Reitoria de Pós Graduação e Pesquisa da Universidade Federal de São Carlos, localizada na Rodovia Washington Luiz, Km. 235 - Caixa Postal 676 - CEP 13.565-905 - São Carlos - SP – Brasil. Fone (16) 3351-8110. Endereço eletrônico: cephumanos@power.ufscar.br

Assinatura da mãe ou responsável legal *

Suzana Paula Miranda Moreira
Nome por extenso

Assinatura do pesquisador

Caroline Teles
Nome por extenso

Assinatura da testemunha

Lucas Correia de Oliveira
Nome por extenso

São Carlos, 19 de julho de 2023

ANNEX C. RESULTS OF THE BRAIN MAGNETIC RESONANCE IMAGING, DATED NOVEMBER 7TH, 2020.

Hospital Alvorada

Ilmo Dr(a):	MIRLENE CECILIA SOARES P CERN		
Registro do Paciente:	HAA-3805273	**Admissão:**	Agendado
Nome do Paciente:	ISAAC MIRANDA MOREIRA		
Data de Nascimento:	2018.05.06	**Data do Exame:**	2020.11.07 10:00
Convênio:	SUL AMERICA SERVIÇOS MEDICOS LTDA		

RESSONÂNCIA MAGNÉTICA DO CRÂNIO

TÉCNICA: Exame realizado pelas técnicas EPI/SE, FSE e GE, com ponderações em Difusão, T1, T2, T2*/SWI, FLAIR e M3D/FSPGR, com aquisições multiplanares, sem a infusão E.V. de contraste paramagnético, demonstra:

Indicação clínica: Atraso do desenvolvimento.

ANÁLISE:

Pedúnculos cerebelare superiores alongados, vermis cerebelar superior de aspecto ligeiramente displásico, certa redução de espessura do mesencéfalo, quarto ventrículo com aspecto de "asa de morcego". Aspectos inespecíficos. Síndrome de Joubert ou outra síndrome relaciona ao sinal do dente molar ("molar tooth sign") ?.

Áreas de mielinização terminal peritrigonais bilaterais, habitual na faixa etária.

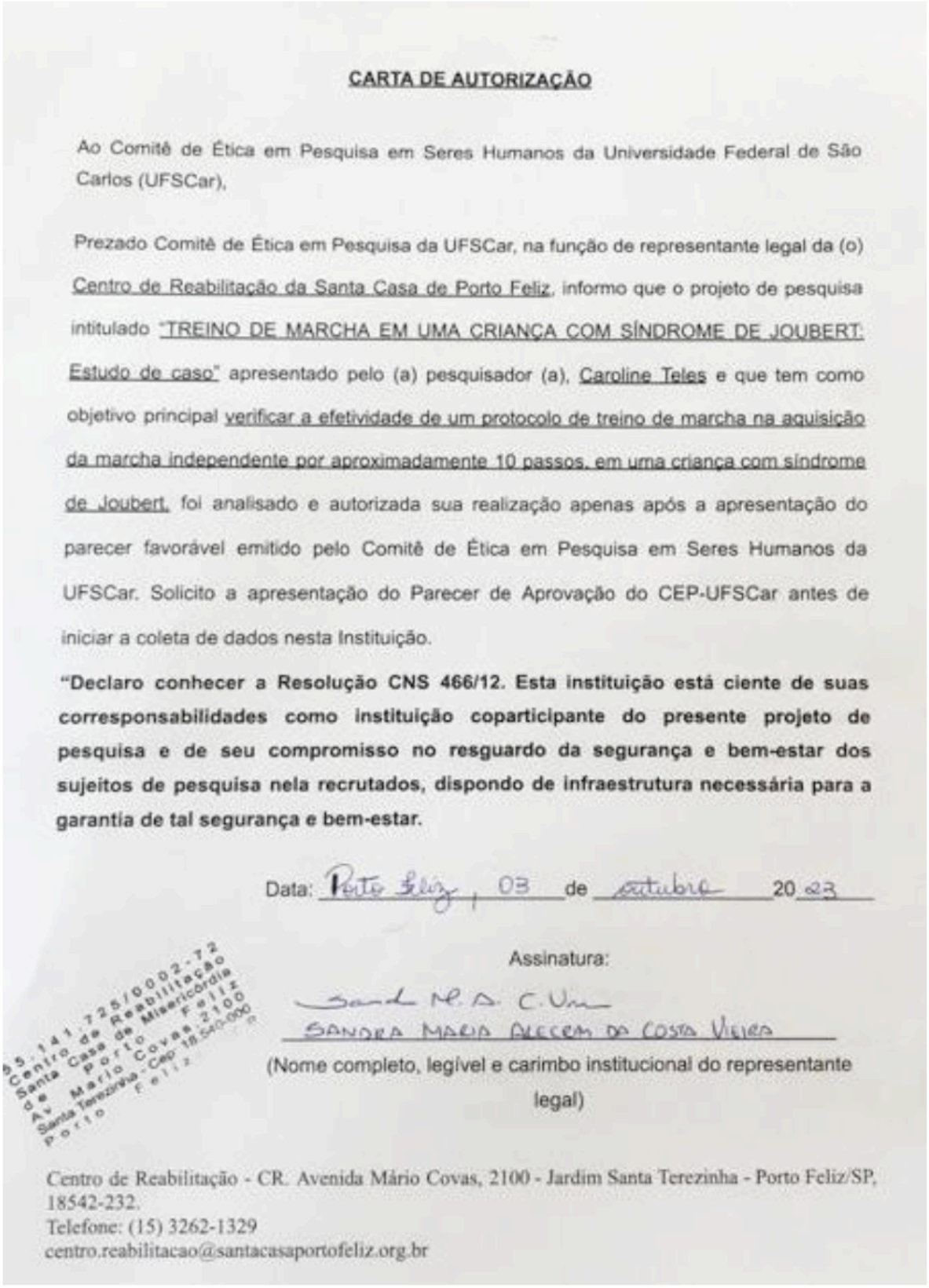
Leve ectasia não hipertensiva do sistema ventricular supratentorial.

Demais cisternas basais, fissuras encefálicas e sulcos corticais de aspecto normal para a faixa etária.
Restante do parênquima encefálico com morfologia e sinal normais.
Flow void habitual dos sistemas carotídeo e vértebro-basilar, pela análise das sequências convencionais.
Não há sinais de eventos isquêmicos agudos, coleções extra-axiais e/ou de hemorragias intracranianas.

Gustavo C. Ferreira

LIBERADO POR:
DR(A). GUSTAVO COUTINHO FERREIRA
CRM: 123775 - SP

ANNEX D. Rehabilitation Center of Santa Casa de Misericórdia de Porto Feliz (Porto Feliz, São Paulo, Brazil) institutional authorization.



CARTA DE AUTORIZAÇÃO

Ao Comitê de Ética em Pesquisa em Seres Humanos da Universidade Federal de São Carlos (UFSCar),

Prezado Comitê de Ética em Pesquisa da UFSCar, na função de representante legal da (o) Centro de Reabilitação da Santa Casa de Porto Feliz, informo que o projeto de pesquisa intitulado "TREINO DE MARCHA EM UMA CRIANÇA COM SÍNDROME DE JOUBERT: Estudo de caso" apresentado pelo (a) pesquisador (a), Caroline Teles e que tem como objetivo principal verificar a efetividade de um protocolo de treino de marcha na aquisição da marcha independente por aproximadamente 10 passos, em uma criança com síndrome de Joubert, foi analisado e autorizada sua realização apenas após a apresentação do parecer favorável emitido pelo Comitê de Ética em Pesquisa em Seres Humanos da UFSCar. Solicito a apresentação do Parecer de Aprovação do CEP-UFSCar antes de iniciar a coleta de dados nesta Instituição.

"Declaro conhecer a Resolução CNS 466/12. Esta instituição está ciente de suas corresponsabilidades como instituição coparticipante do presente projeto de pesquisa e de seu compromisso no resguardo da segurança e bem-estar dos sujeitos de pesquisa nela recrutados, dispondo de infraestrutura necessária para a garantia de tal segurança e bem-estar.

Data: Porto Feliz, 03 de outubro 2023

Assinatura:

Sandra M. A. C. Vieira

SANDRA MARIA ALECRIM DA COSTA VIEIRA

(Nome completo, legível e carimbo institucional do representante legal)

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centro.reabilitacao@santacasaportofeliz.org.br

APPENDIX A. GUIDELINE BOOKLET FOR PARENTS/CAREGIVERS ON THE IMPLEMENTATION OF THE TRAINING PROTOCOL.

	Tuesday	Friday	Duration
2nd week	a) Activity 1	a) Activities 2 and 3	40 min
	b) Activities 2 and 3	b) Activity 1	
3rd week	a) Activities 2 and 3	a) Activity 1	40 min
	b) Activity 1	b) Activities 2 and 3	
4th week	a) Activity 1	a) Activities 2 and 3	40 min
	b) Activities 2 and 3	b) Activity 1	
5th week	a) Activities 2 and 3	a) Activity 1	40 min
	b) Activity 1	b) Activities 2 and 3	

Activity 1

Let's practice your child's walking using the EVA mat with obstacles, as shown in Image 1, for approximately 20 minutes. To assist your child during the exercise, stand in front of him and hold his hands to provide support throughout the activity. This exercise can be performed indoors, such as in the living room.

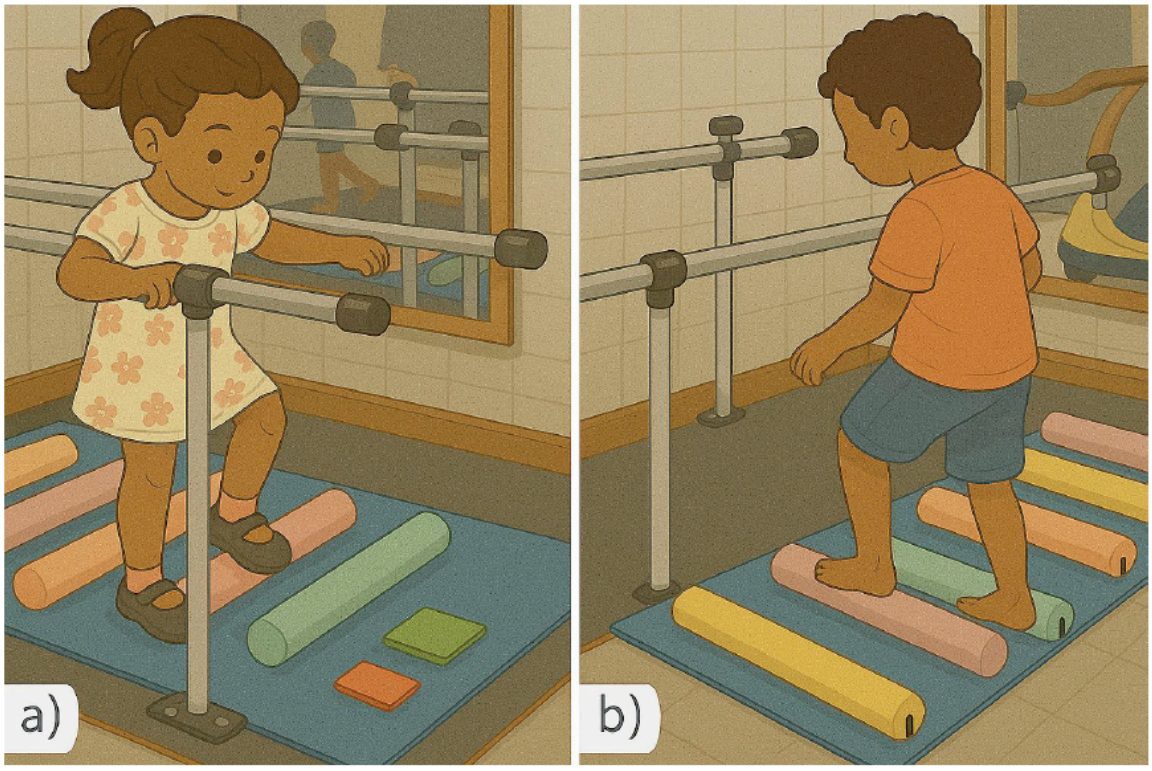

Image 1

Source: Generated by ChatGPT.

Activity 2

Your child's gait should be practiced around the house, using a walker for support if necessary, for approximately 10 minutes, as illustrated in Image 2. To assist your child during the exercise, you may remain beside him, providing encouragement and motivation to promote self-generated movement. This activity can be performed indoors, allowing the child to move from one room to another. It is important to ensure sufficient space for your child to maneuver the walker and move safely.

Image 2

Source: Generated by ChatGPT.

Activity 3

The gait training should also be practiced in the neighborhood (community setting), using a walker for support if necessary, as illustrated in Image 3. You should use encouraging words and positive reinforcement to motivate your child during the activity. To assist with the exercise, remain beside him, providing verbal encouragement and promoting independent movement.

Image 3

Source: Generated by ChatGPT.

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