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Functional School-Based Physical Therapy Management For A Child With Pallister-Killian Syndrome: A Case Report

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The child's parent provided written consent in allowing the use of medical information and photo/video footage for this case report. The parent received information from the university's Health Insurance Portability and Accountability Act (HIPAA) policies.

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Key Words: Pallister-Killian Syndrome, body-weight support treadmill training

Abstract

Background and Purpose: Pallister-Killian Syndrome (PKS) is a rare genetic disorder resulting from an additional short arm in chromosome 12. This condition leads to varying levels of multi-system impairments that impact a child's development in gross motor skills yielding functional limitations. Pediatric physical therapy (PT) management is crucial for these children to facilitate improvements in sitting, standing, and ambulation. The purpose of this case report was to describe functional school-based PT management for a child with PKS.

Case Description: The child was a seven-year-old male diagnosed with PKS at 20 weeks' gestation. The child was seen for PT five times a week in a specialized school setting for six weeks. The child presented with hypotonia, delayed gross motor skills, generalized weakness, and impaired posture. The child's level of function was classified as Gross Motor Function Classification System Level V. The child required minimum assistance in sitting and maximum assistance for standing and ambulating. Interventions included: standing exercise, body-weight support treadmill training (BWSTT), overground gait training, and a standing program.

Outcomes: The child's standing time progressed from 16 seconds to 3:05 minutes, reciprocal stepping during BWSTT improved from 3 to 63 steps, and overground gait training progressed from 0 to 6 steps. Improvements in observational posture and strength were also seen. The standing program was not tolerated by the child, therefore removed from the plan of care (POC).

Discussion: This six-week POC resulted in improved standing time, reciprocal steps, strength, and posture for a child with PKS. Further research is warranted on the benefits of these interventions to elicit improvements in gross motor function and declines in disability for children with PKS.

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Background and Purpose

Pallister-Killian Syndrome (PKS) is a rare and sporadic genetic disorder caused by mosaic tetrasomy of the p arm of chromosome 12.[1] PKS may also be known as Pallister-Killian Mosaic Syndrome, Tetrasomy 12p, Pallister Mosaic Aneuploidy, and Tischler-Nicola Syndrome. PKS is a non-hereditary condition that results in clinical manifestations affecting multiple systems.[2] Common manifestations include hypotonia, intellectual disability, diaphragmatic hernia, epilepsy or seizures, hearing loss, visual impairment, hypo- or hyperpigmentation, and congenital heart defects.[3] Children with PKS exhibit distinct craniofacial anomalies that may involve frontal bossing, frontal-temporal alopecia, hypertelorism, nasal bridge depression, a long philtrum, a short neck, and micrognathia.[3] Srinivasan and Wright[4] mention that a prenatal diagnosis through cordocentesis, amniocentesis, or chorionic villus sampling is possible. Postnatal and childhood diagnosis is done by a genetic specialist examining skin fibroblasts and buccal smears. Diagnostic detection is at its highest through examination of amniocytes and bone marrow cells resulting in a rate of 100%.[4]

The literature has reported a prevalence of 1/20,000 live births from prenatal diagnosis.[5-6] In the medical literature, more than 150 cases of PKS have been documented.[2] Under-diagnosis has been theorized due to a low detection rate from peripheral blood testing and mild cases being unreported.[7-8] Saito et al[9] authored a case report on two patients aged 43 and 37 years old whom required significant medical needs as they progressed in age. Izumi and Krantz[4] state that life expectancy for this condition has not been formally established, although individuals ranging between 40 to 50 years old living with PKS were mentioned.

Children with PKS experience developmental delay, ranging from mild to profound. Wilkens et al[3] reported children with PKS walking at 38.8 months within a cohort (refer Appendix 3). Topcu and Uzuner[10] found improvements in gross motor function from

physiotherapy (also known as physical therapy in the United States) and rehabilitation for a three-year-old male with PKS. Based on their findings, it is evident that individuals born with PKS may benefit from physical therapy (PT) interventions to assist in achieving gross motor skills.[10] Unfortunately, the evidence for recommended PT interventions for this population is inadequate.

This case report was needed due to limited information suggesting appropriate physical therapy interventions for children with PKS. The purpose of this case report was to describe a school-based PT intervention plan for a child with PKS, which involved a standing exercise, body-weight support treadmill training (BWSTT), overground gait training, and a standing program.

Patient History and Systems Review

The child's mother provided written consent for participation in this case report. The child was a seven-year-old male student in a specialized day school for children with brain injuries and disorders. He was of Hispanic descent and lived with his mother and two typically developing adolescent sisters. At 20 weeks' gestation, a primary medical diagnosis of Pallister-Killian Syndrome was confirmed. The child was born 39 weeks with jaundice. Phototherapy was received for three days, followed by spending five days in the neonatal intensive care unit. Magnetic resonance imaging (MRI) at three months of age revealed left germinal matrix hemorrhage, followed by mild ventriculomegaly at two years of age.

The medical history also revealed the following: hypotonia; global development delay; cortical visual impairment (CVI); intermittent nystagmus; bilateral hearing impairment; atrial septal defect (ASD); oropharyngeal dysphagia. It was noted that this presentation was parallel with the manifestations of PKS. The child also presented with dysmorphic facial features, low-

set ears, hypopigmentation of the trunk represented as spots, ASD, visual and auditory impairments, and finger flapping. There were no reports of medication administration while in school. The child presented with signs of sensory processing issues, which was reported by his occupational therapist (OT). When touched lightly at the hands or feet, the child would immediately withdraw. When overstimulated, the child would shake and scream.

The main concern for this child was the limited attainment in gross motor skills for functional school-based activities. The primary problems involved his walking ability, need for maximum assistance with transfers, and inability to perform class activities while standing. The child received PT services five days a week in school. The child received other related services, including occupational therapy, speech therapy, and vision therapy. Prior PT interventions included neurodevelopmental treatment (NDT), standing exercises, and upper extremity (UE), lower extremity (LE), and trunk strengthening. A differential diagnosis was not made due to the prenatal diagnosis. The plan for examination, based on existing data, involved administration of the Pediatric Evaluation of Disability Inventory (PEDI), range of motion (ROM) measurements, postural observation, and strength testing through observation of functional skills.

A systems review was completed based on the child's examination and medical history. Impairments were found in the systems of cardiovascular/pulmonary, musculoskeletal, neuromuscular, and integumentary. The child's communication, affect, cognition, language, and learning style also presented with impairments. The child presented with bilateral solid ankle-foot orthoses (AFOs), a tilt-in-space wheelchair, bilateral hearing aids, and eyeglasses for hyperopia with astigmatism. It should be noted that the physical therapists had limited interaction with the child's mother to provide perspective on PT interventions for this case. Refer to Table 1 for further details.

The child was an ideal candidate for this case report based on the rare prevalence of his genetic diagnosis and his unique characteristics involving sensory input and regulation. In addition, the literature is currently lacking in physical therapy interventions and outcomes for children with Pallister-Killian Syndrome.

Examination – Tests and Measures

Examination procedures were completed to further assess the child's impairments and limitations. Passive ROM measurements were done, according to Norkin and White[11], with an EMI Plastic 12" Goniometer (Elite Medical Instruments EMI Inc., Fullerton, CA). Due to tactile sensitivity at the hands and feet, the child did not tolerate further ROM measurements (refer to Table 2). The PEDI was administered to measure the child's level of independence and functional skill ability due to maximum assistance required for his daily functions. The PEDI measures functional performance and caregiver assistance in three domains: self-care, mobility, and social function. Nichols and Case-Smith[12] concluded that the PEDI is a reliable and valid functional assessment for children with disabilities. The child scored significantly below the normative range in all domains (refer to Appendix 1). Strength testing was done by observation of functional skills as described by the Strength Testing in Pediatric Physical Therapy Fact Sheet by the Academy of Pediatric Physical Therapy.[13] This was done by observing the child's movement activities and making conclusions about the child's strength based on performance. This method was selected to accommodate the child's cognitive impairment and inability to perform a standard manual muscle test. Refer to Appendix 2 for further information.

Clinical Impression: Evaluation, Diagnosis, Prognosis

The child's symptoms of hypotonia, global developmental delay, and CVI were present

resulting from the primary diagnosis of PKS. The child's impairments in tone and motor function, strength and ROM, and cognition led to his inability to stand, ambulate, transfer, and participate independently in his school. The child's level of function was classified as Gross Motor Function Classification System (GMFCS) Level V. No further referral or consultation was needed to initiate the plan of care (POC). The child continued to be appropriate for this case report based on his potential for functional improvements with school-based PT.

A specific ICD-10 Code for Pallister-Killian Syndrome is currently unavailable. Other specified chromosome abnormalities (ICD-10 code: Q99.8) was used as a medical diagnosis. Congenital hypotonia (ICD-10 code: P94.2) was also chosen due to the child's history of low tone and clinical observation upon PT examination. Specific developmental disorder of motor function (ICD-10 code: F82) was selected as the primary PT diagnosis based on the delay in gross motor skills.

The child's prognosis was determined to be good based on current gross motor abilities, absence of seizures, and the ability to tolerate PT services five times a week.[14-15] Signs of intellectual disability, impairments in multiple systems, and use of a wheelchair as primary means of mobility were negative factors that contributed to his prognosis. It should be noted that there is mixed research in the prognosis for patients with PKS.[9-10, 14] Topcu and Uzuner[10] found improvements, at a two-year follow-up, in gross motor function after physical therapy for a three-year-old child. Blyth et al[14] found that individuals within their cohort (n = 22) began ambulating between 16 months and 8 years.

The child's interventions from a prior POC involved NDT and various strengthening activities. An improvement in sitting balance was reported by the previous physical therapist. Detailed information on preceding treatment interventions and outcomes were not documented in this case report. The current six-week POC involved a transition to promote standing and

ambulatory function. These interventions were administered 5 times a week and involved a standing exercise, body-weight support treadmill training (BWSTT) using a Rifton Dynamic Pacer Gait Trainer (Community Products, LLC dba Rifton Equipment, Rifton, NY), overground gait training, and a standing program in a Rifton Small Mobile Stander (Community Products, LLC dba Rifton Equipment, Rifton, NY). Short and long-term goals were established in a quarterly and annual fashion (see Table 3).

Intervention and Plan of Care

Coordination and communication to the PT treatment schedule was made with other related service providers and the child's teacher to avoid any conflict. Documentation was completed through the school's computer system. The child's paraprofessional, which is another term for a teacher's aide, assisted with transfers and interventions when necessary.

The plan was to administer interventions for six weeks, but due to illness, the child missed one week of treatment. The interventions were selected based on the child's functional limitations and goals. These interventions included: a standing exercise; BWSTT; overground gait training; and a standing program. Standing time during the exercise was measured using a Fitbit Charge 3 (Fitbit Inc., San Francisco, CA). The number of reciprocal steps taken during BWSTT and overground gait training was measured using observational methods by two clinicians.

Standing Exercise

Examination observation and medical history review revealed hypotonia and weakness in the lower extremities. The standing exercise was an intervention that was added to the child's POC to assist in achieving independent standing and ambulation for school-based activities. Due

to the lack in LE strength, the child would intermittently buckle his knees and collapse to the PT mat when assisted to stand and ambulate. Evidence suggests exercises for children diagnosed with Prader-Willi Syndrome who have hypotonia, including "stand from floor lying".[15] A standing exercise was selected as a modification due to the advanced nature of standing from floor lying. The equipment used for this intervention involved a Ladder Back Chair (Smirthwaite Ltd., Heathfield, Newton Abbot, Devon, UK) and a Kaye S2A Adjustable Bench (Kaye Products Inc., Hillsborough, NC). The PT sat behind the child and provided maximum assistance to facilitate standing. Tactile cues and guarding were provided to the posterior and anterior hip region, creating a grasp surrounding both hips. The child was cued and encouraged to stand for an extended time for five trials. When the child was tired or reluctant to maintain the stand, he would show apprehension to holding the ladder and forcibly attempt to sit. If this occurred, he was slowly returned to a seated position on the bench. Six repetitions per session were completed with a frequency of three times a week. Refer to Table 4 and Figure 1.

Body-Weight Support Treadmill Training (BWSTT)

The purpose of intervening with BWSTT was to initiate and promote ambulation. Damiano & Dejong[16] suggested improved gait performance and gross motor development for children with developmental delay. This intervention allows the therapists to manipulate the environment and provide facilitation when necessary. Facilitation was done by grasping the child's ankles and placing a foot forward while following a reciprocal gait pattern. The treadmill used was a Smooth Fitness 6.25 Treadmill (ICON Health & Fitness Inc., Logan, UT). A Rifton Pacer Gait Trainer was placed on the treadmill and used as the mechanism for body weight support. The intervention lasted 15 minutes per session. See Table 4 for detailed parameters and Figure 2 for illustration.

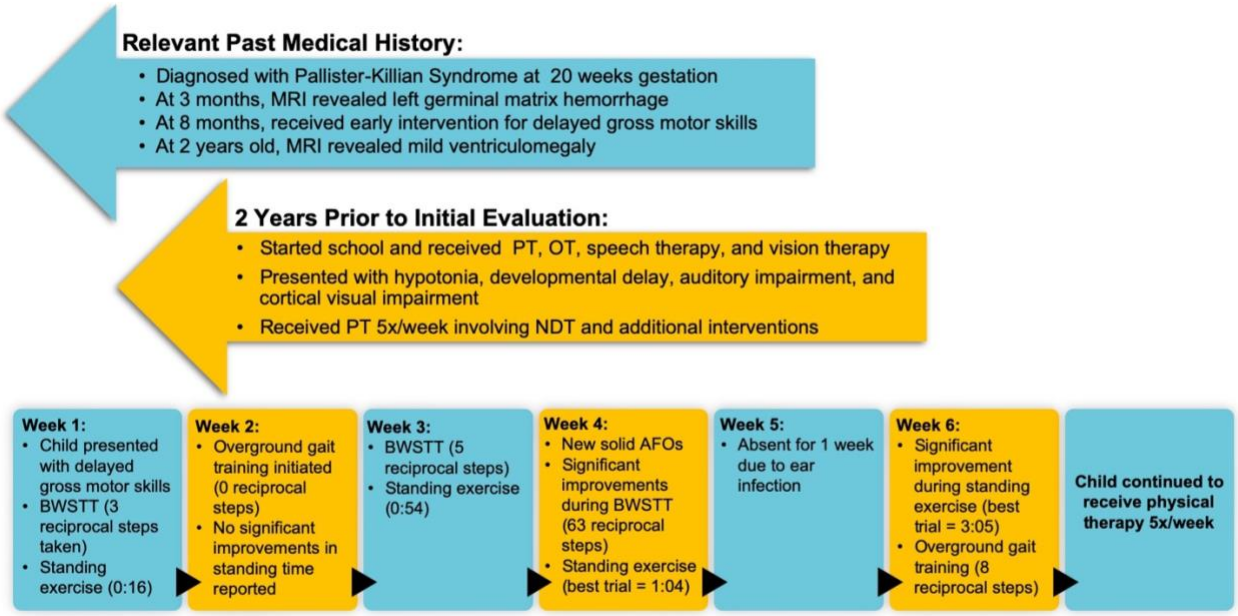
Overground Gait Training

This intervention was selected to promote step initiation and to assess if BWSTT provided carry over to a typical walking environment for the child. Paleg and Livingstone reported that gait trainers may aide in the development of ambulation for children who are not able to perform this without support.[17] Their findings suggested that gait trainers may provide positive change in outcomes for participation and function. This intervention was also used as a means to evaluate if BWSTT assisted in overground gait training. This intervention lasted 15 minutes per session. Refer to Table 4 for detailed parameters and Figure 2 for illustration.

Standing Program

Paleg et al[18] found that adherence to a standing program for five days a week has been shown to improve bone mineral density, hip stability, and range of motion. The child received a standing program based on the stated benefits and his inability to stand independently. The child was suggested to use the Rifton Small Mobile Stander five days a week for one hour in the classroom. The child's paraprofessional and physical therapist reported that the child was unable to tolerate the program for the recommended time.

Timeline



Outcomes

The child was seen for a total of 15 sessions over six weeks for this case report. The child received additional PT treatment sessions from another physical therapist due to requirements by the school. This resulted in a total of approximately 25 sessions. Improvements were seen in observational posture and strength at six weeks compared to baseline (see Appendix 2). Intervention outcomes were recorded for each session (see Table 4). Improvements in standing time were seen from 16 seconds at baseline to three minutes and five seconds at week six for the standing exercise. Reciprocal step initiation increased from three steps at baseline to 63 steps at week four during BWSTT. Reciprocal steps increased from zero steps at week two to eight steps at week six during overground gait training. Improvements in quality of movement standing and ambulation were noted. The PEDI was not re-administered at the end of the POC due to limited time. A plan to re-administer the performance measure in the future was discussed. No diagnostic or other test results were awaiting follow-up. The child adhered to all interventions excluding the standing program. The child continued to receive physical therapy treatment after

the current POC, which resulted in undocumented goal achievements for this case report.

Discussion

This case report demonstrated its intended purpose by describing school-based PT management for a child with PKS. Within the six-week POC, the child exhibited improvements in observational posture and strength, longer standing times, and increased reciprocal steps.

There is minimal research on PT management for children with PKS, however, a program for a three-year-old male with PKS lead to improved gross motor function.[10] Standing exercises, a modification to standing from floor lying, is a recommended exercise for children with Prader-Willi Syndrome (PWS).[15] Children with PWS and PKS commonly present with hypotonia and developmental delay. Improvements were seen in standing time at the end of this POC. The intervention of BWSTT has been shown to improve gait performance and motor development for children with developmental delay.[16] Improvement was seen at week four compared to baseline. Paleg and Livingstone[17] mention overground gait training having some contribution to walking distance and number of steps, which was seen in this case at week four and week six. It is emphasized that the motor development of children with PKS occurs gradually and that continued physical therapy will assist in children reaching their potential.[14] Further research is warranted on the benefits of these interventions to elicit improvements in gross motor function and declines in disability for children with PKS.

One of the strengths of this case report included the unique nature of the child's diagnosis and clinical presentation. The child was diagnosed with a rare genetic syndrome and presented with signs of sensory dysregulation. The child's past medical history included prior left germinal matrix hemorrhage and mild ventriculomegaly. Another strength of this case report was the child's level of function being classified as GMFCS Level V. There is limited research on

recommended PT interventions for children with this classification. This case report also incorporated new treatment methods for a child with PKS that have not been reported in current research, including a standing exercise, BWSTT, overground gait training.

One limitation was that the child missed one full week of school and PT due to an ear infection, therefore receiving five weeks of treatment following this POC. Another limitation included a restricted amount of time for this case report. Improvements in gross motor function have been seen after one to two years of treatment.[10] The limited time also prevented measurement for achieved short-term and long-term goals. Intervention administration and outcome measurements over longer periods may demonstrate significant clinical benefit. Another limitation was a lack of providing information on the extra PT treatment sessions. It has been noted that the extra sessions, or prior sessions before the initiation of this case study, may have contributed to the child's progress. Another barrier was the lack of communication with the child's parents to incorporate a home exercise program (HEP). If a HEP was communicated to the parents, this may have resulted in higher functional gains. A final limitation was the delay in the child receiving new solid AFOs. The child was able to achieve more steps, stand for longer periods of time, and presented with improved quality of movement with the new AFOs. Further investigation is necessary to determine the level of effect these factors contributed to the child's progress.

It may be speculated that a functional school-based PT program incorporating a standing exercise, BWSTT, and overground gait training may increase observational posture and strength, standing time, and reciprocal steps for children with PKS. Further research is warranted on the various methods of interventions to elicit improvements in function and disability for this distinctive population. These interventions may comprise of a standing exercise and program, BWSTT, overground gait training, and other approaches. Conceivably, research is to be made on

the level of effect from task-oriented interventions for children with PKS. This conjecture is based on the intervention outcomes advancing gross motor function for children with various neurodevelopmental disorders.[19] Future research on applicable and effective PT management may have the capacity to improve gross motor function and decrease the level of disability for children with this sporadic genetic condition.

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Tables and Figures

Table 1: Systems Review

Cardiovascular/Pulmonary	
Impaired	• Atrial septal defect • Decreased endurance due to primarily transporting via wheelchair and decreased physical activity
Musculoskeletal	
Impaired	• Generalized weakness in the bilateral upper extremities and bilateral lower extremities
Neuromuscular	

Impaired	• Hypotonia • Gross motor delay • Difficulty processing sensory input causing overstimulation episode (shaking and brief screaming) with auditory, tactile, and visual information
Integumentary	
Impaired	• Hypopigmentation on trunk showing spots
Communication	
Impaired	• Communicates non-verbally • Use of communication board and other adaptive communication devices • Laughs, cries, and portrays facial expressions • Uses body language and withdrawal in situations of discomfort
Affect, Cognition, Language, Learning Style	
Impaired	• Affect is displayed at times of happiness, discomfort, and sadness. Child also shows signs of pseudobulbar affect. • Presents with characteristics of cognitive impairment and intellectual disability • English is the primary language • Learning style includes visual, verbal, auditory, and physical cues

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381
382

Table 2: Passive Range of Motion

Motion	Right	Left
Elbow Flexion	0 to 151°	4 to 155°
Knee Flexion	151°	145°
Knee Extension	0°	0°
Knee Extension with Hip Flexion	21°	15°
Hip Flexion with Knee Flexion	91°	95°
Hip Flexion with Knee Extension	29°	36°
Hip Extension with Knee Flexion	0 to 13°	0 to 10°
Ankle Plantar Flexion	Unable to measure with goniometry due to child's sensitivity to touching feet; Within Functional Limits based on observation	Unable to measure with goniometry due to child's sensitivity to touching feet; Within Functional Limits based on observation
Ankle Dorsiflexion	Unable to measure with goniometry due to child's sensitivity to touching feet; Within Functional Limits based on observation	Unable to measure with goniometry due to child's sensitivity to touching feet; Within Functional Limits based on observation

Table 3: Child's Goals

Short-Term Goals	Long-Term Goals
By 3 months, the child will transition from ring sitting to tall kneeling with minimal assistance (Min A) to transfer in class By 3 months, the child will transition from sitting on the bench to standing wearing B AFOs with Min A in class	By 12 months, the child will be able to bench sit with B AFOs with supervision for 15 minutes to allow safe participation in class meetings, story time, and academic sessions during the school day
By 3 months, the child will sustain a ring sitting position with Min A for eight minutes while engaging in classroom activities By 3 months, the child will ambulate 100 consecutive steps in a Rifton Gait Trainer with B AFOs and moderate assistance (Mod A) for steering to transport from the classroom to other school activities By 3 months, the child will maintain standing with Min A and B AFOs for five minutes to wait in line in class	By 12 months, the child will ambulate 250 feet with Min A, no assistive device, and B AFOs to transport from class to the school bus
By 3 months, the child will demonstrate sitting with the head and trunk in midline, hand on lap for 60 to 90 seconds with Mod A	By 12 months, the child will improve gross motor skills to sit independently for 10 minutes
By 3 months, the child will be able to take 20 steps with a reciprocal pattern with maximum assistance (Max A)	By 12 months, the child will demonstrate increased gross motor skills to walk independently

Table 4: Interventions

Day of Treatment	Intervention	Parameters	Notes
Week 1: Day 1 (Baseline)	BWSTT at 0.5 mph (15 minutes) Standing exercise (15 minutes)	BWSTT: Alternating 1-minute independent with verbal/tactile cues, followed by 1 minute with B LE facilitation	Tolerated well, minimal complaints. Took 3 reciprocal steps. Standing exercise: Trial 1: 16 sec Trial 2: 21 sec Trial 3: 27 sec Trial 4: 36 sec Trial 5: 24 sec
Week 1: Day 2	BWSTT at 0.5 mph (15 minutes)	Alternating 1-minute independent with verbal/tactile cues,	Tolerated well, moderate complaints; Took 3 reciprocal

		followed by 1 minute with B LE facilitation	steps.
Week 1: Day 3	BWSTT at 0.5 mph (15 minutes)	Alternating 1- minute independent with verbal/tactile cues, followed by 1 minute with B LE facilitation	Attempted with ankle weights for 2 minutes but did not show active movement in BLE; Mod to max complaints towards 10 minutes but was able to complete full 15 minutes; Took ~4 reciprocal steps.
Week 2: Day 4	Overground gait training (30 minutes)	Independently and no facilitation	Did not tolerate for more than 5 minutes; Child began to cry. Took 0 steps
Week 2: Day 5	BWSTT 0.5 mph (15 minutes) Overground gait training (30 minutes)	B LE facilitation by swinging leg at the foot	Took 4 steps reciprocally Took 6 steps reciprocally
Week 2: Day 6	Standing exercise (15 minutes)	5 trials with seated rest breaks in between	Tolerated well during classroom activity Trial 1: 15 sec Trial 2: 26 sec Trial 3: 32 sec Trial 4: 36 sec Trial 5: 30 sec
Week 3: Day 7	BWSTT 0.5 mph (15 minutes)	Alternating 1- minute independent with verbal cues and motivation 1 minute with B LE facilitation	Closed eyes in mid-session Took 5 steps
Week 3: Day 8	Standing exercise (15 minutes) Overground gait training (30 minutes)	Mostly needed tactile cueing for weight shifting	Trial 1: 18 sec Trial 2: 31 sec Trial 3: 43 sec Trial 4: 54 sec Trial 5: 35 sec Took 4 steps reciprocally
Week 3: Day 9	BWSTT 0.5 mph	Alternating 1- minute	Did not actively

	(15 minutes)	independent with verbal cues and motivation 1 minute with B LE facilitation	initiate (2 reciprocal steps) Did not tolerate well; Closed his eyes and pretended to sleep most of the session
Week 4: Day 10	Standing exercise (15 minutes)	5 trials with seated rest breaks in between	Trial 1: 45 sec Trial 2: 54 sec Trial 3: 1 minute Trial 4: 1 minute 4 seconds Trial 5: 55 seconds
Week 4: Day 11	Overground gait training (30 minutes)	B LE facilitation by swinging leg at the foot	Took 6 reciprocal steps
Week 4: Day 12	BWSTT 0.5 mph (15 minutes)	Alternating 1- minute independent with verbal cues and motivation 1 minute with B LE facilitation	New B solid AFOs were administered Took 63 reciprocal steps Tolerated well
Week 5	Absent and missed PT for 1 week due to ear infection		
Week 6: Day 13	Standing exercise (15 minutes) BWSTT 0.5 mph (15 minutes)	5 trials with seated rest breaks	Trial 1: 3 minutes 5 seconds Trial 2: 1 minute 7 seconds Trial 3: 1 minute 51 seconds Trial 4: 1 minute Trial 5: 43 seconds
Week 6: Day 14	Standing exercise (15 minutes)	5 trials with seated rest breaks	Trial 1: 1 minute 50 seconds Trial 2: 2 minutes 52 seconds Trial 3: 1 minute 30 seconds Trial 4: 1 minute 13 seconds

			Trial 5: 40 seconds
Week 6: Day 15	Overground gait training (30 minutes)	B LE facilitation by swinging leg at the foot	Took 8 reciprocal steps

B= bilateral, LE = lower extremities, BWSTT = body weight support treadmill training

Figure 1: Standing exercise

Week 6 of the treatment intervention showing equipment and level of assistance required

Figure 2: a) BWSTT b) Overground gait training

a) Week 1 of intervention administration with required equipment

b) Week 6 of intervention administration with required equipment and clinician positioning

Appendices

Appendix 1: Pediatric Evaluation of Disability Inventory Scores

PEDI	Domain	Raw Score	Normative Standard Score Ranges for a Child 7 years-old or older*
	Self-Care – Function Skills	4	30.7-55.3
	Mobility – Function Skills	5	29.9-54.7
	Social Function – Function Skills	1	31.8-59.4
	Self-Care – Caregiver Assistance	1	37.4-61.9
	Mobility – Caregiver Assistance	3	31.5-57.9

	Social Function – Caregiver Assistance	1	37.1-74.1

*Taken from: https://www.sralab.org/rehabilitation-measures/pediatric-evaluation-disability-inventory

Appendix 2: Observational Examination Procedures

	Baseline	Week 6
Posture	**Sitting:** The child presented with a kyphotic posture and a posterior pelvic tilt when sitting in the wheelchair. When transferred to the PT mat, the child ring sat and was able to maintain this position without his hands propped. While sitting on a therapy bench, he presents with kyphotic posture and requires min A. **Standing:** After providing mod A to stand from the wheelchair, the child required a two hand-held assist to maintain standing. The child was able to maintain this position, although manual weight shifting was required to prevent a loss in balance.	**Sitting:** The child sits with mild kyphotic posture while sitting in the wheelchair. On the PT mat, he was able to cross-leg sit with min A with mild kyphotic posture. On a therapy bench, he is able to sit with supervision with mild kyphotic posture. **Standing:** The child continued to require mod A to stand from the wheelchair with a two hand-held assist. The child was able to maintain this position and did not require manual cues for weight shifting.
Strength Testing by Observation of Functional Skills	The child is able to sit without support for greater than one minute and is able to right body from perturbations. The child is unable to squat, stand, or walk without max A. In order to maintain a standing position, the child must use upper extremity to prevent the knees from buckling. The child is able to complete a sit-to-stand with moderate to maximum assistance. The child is able to use upper extremities and trunk strength to return to a seated position on a Large Tumble Forms 2 Roll (Patterson Companies Inc., Mendota Heights, MN). The child is able to use upper extremity strength to bring his body back into the wheelchair.	The child is able to sit upright on the therapy mat independently and able to respond to perturbations. The child is able to squat with mod A, stand with mod A, and walk with moderate assistance for weight shifting and reciprocal stepping in the gait trainer. The child is able to maintain standing while a two hand-held moderate assist. The stand from a therapy bench requires min A to mod A, with cues for weight shifting and hip muscle activity (See Figure 1). The child continues to have functional core strength to return to a seated position on a Large Tumble Forms 2 Roll. The child is able to use upper extremity strength to push off of a surface or person to stand.

min A = minimal assistance, mod A = moderate assistance, max A = maximum assistance

Appendix 3: Achieved Gross Motor Milestones for Typically Developing Children[20] vs. PKS Cohort[3]

Gross Motor Milestone	Average Age (in months): Typically Developing	Average Age (in months): PKS Cohort
Rolling	6-8	10.8
Sitting	8	21.2
Walking	12	38.8

CARE Checklist

CARE Content Area	Page
1. **Title** – The area of focus and "case report" should appear in the title	1
2. **Key Words** – Two to five key words that identify topics in this case report	1
3. **Abstract** – (structure or unstructured) a. Introduction – What is unique and why is it important? b. The patient's main concerns and important clinical findings. c. The main diagnoses, interventions, and outcomes. d. Conclusion—What are one or more "take-away" lessons?	2
4. **Introduction** – Briefly summarize why this case is unique with medical literature references.	3-4
5. **Patient Information** a. De-identified demographic and other patient information. b. Main concerns and symptoms of the patient. c. Medical, family, and psychosocial history including genetic information. d. Relevant past interventions and their outcomes.	4-6
6. **Clinical Findings** – Relevant physical examination (PE) and other clinical findings	6-7
7. **Timeline** – Relevant data from this episode of care organized as a timeline (figure or table).	11
8. **Diagnostic Assessment** a. Diagnostic methods (PE, laboratory testing, imaging, surveys). b. Diagnostic challenges. c. Diagnostic reasoning including differential diagnosis. d. Prognostic characteristics when applicable.	7-8

9. **Therapeutic Intervention** a. Types of intervention (pharmacologic, surgical, preventive). b. Administration of intervention (dosage, strength, duration). c. Changes in the interventions with explanations.	8-10
10. **Follow-up and Outcomes** a. Clinician and patient-assessed outcomes when appropriate. b. Important follow-up diagnostic and other test results. c. Intervention adherence and tolerability (how was this assessed)? d. Adverse and unanticipated events.	11-12
11. **Discussion** a. Strengths and limitations in your approach to this case. b. Discussion of the relevant medical literature. c. The rationale for your conclusions. d. The primary "take-away" lessons from this case report.	12-14
12. **Patient Perspective** – The patient can share their perspective on their case.	N/A
13. **Informed Consent** – The patient should give informed consent.	4