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Functional Mobility Management Of A Patient With Adult-Onset Hereditary Proximal Motor Neuropathy Following A Tibial Fracture: A Case Report

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The patient signed an informed consent allowing the use of medical information and picture/video footage for this report and received information on the institution's policies regarding the Health Insurance Portability and Accountability Act.

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Abstract

Background & Purpose

Adult-onset hereditary proximal motor neuropathy (AHPMN) is a subcategory of spinal muscular atrophy, caused by survival motor neuron gene mutation. This rare disease, affecting approximately 1 in 10,000 people, presents as proximal weakness and muscle wasting, more commonly in the lower extremities, in addition to gait unsteadiness and difficulty standing. Additionally, the lifetime risk of developing CHF is one in five; Since the diagnosis poses a risk factor for falling, it may increase the likelihood of falls. Regarding seniors over the age of 65, fall-related injuries increased from 49.4% to 58.8% between 2005 and 2013, accounting for 2.5 million older adults treated in the emergency department for falls. There is limited literature currently describing the physical therapy (PT) management for AHPMN. The purpose of this case report was to document the outcomes of various functional mobility interventions for a geriatric patient with a left tibial fracture, secondary to a fall, with AHPMN and CHF.

Case Description

GL was a 77 year-old male with limited baseline activity, who sustained a tibial fracture secondary to falling while walking up a ramp. Significant medical history included AHPMN and CHF.

Outcomes

The Lower Extremity Functional Scale, manual muscle testing, and gait pattern assessment were all used to assess the functional progress, with improvements in all three categories demonstrated.

Discussion

This case report provides opportunity to describe the PT management of a patient with AHPMN who sustained a tibial fracture. Upon discharge, the patient had achieved all set goals to assist with improvements in independent functional mobility. There is opportunity for further investigation in this area of PT for comparison in the benefits of the interventions performed.

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Background & Purpose

Adult-onset hereditary proximal motor neuropathy (AHPMN) is a subcategory of spinal muscular atrophy, which is caused by survival motor neuron gene mutation.[1] This rare disease, affecting approximately 1 in 10,000 people, presents with the primary symptoms of proximal weakness and muscle wasting of the limbs, more commonly in the lower extremities, in addition to gait unsteadiness and difficulty standing.[1, 2] Concerning the diagnosis of congestive heart failure (CHF), the lifetime risk of development is one in five, and as the diagnosis poses a risk factor for falling it may, in turn, increase the likelihood of falls.[3, 4] Furthermore, regarding seniors over the age of 65, fall-related injuries increased from 49.4% to 58.8% (per 1000 population) between 2005 and 2013, which, for this population, accounts for 2.5 million older adults treated in the emergency department for falls.[5, 6] This patient, with both AHPMN and CHF diagnoses, was referred to physical therapy (PT) after acquiring an acute left (L) tibial fracture secondary to falling while walking up an inclined ramp. An open reduction internal fixation (ORIF) procedure was performed the following day. There is limited literature currently describing the physical therapy (PT) management for AHPMN; thus, this case report may help fill a gap in the literature, which is sparse with functional mobility treatment of a patient with an acute L tibial fracture, who has AHPMN. For that reason, this case could provide future clinicians the opportunity for intervention recreation with a comparable patient with relatively similar clinical presentation, symptoms, or comorbidities. The purpose of this case report was to document the outcomes of various functional mobility interventions for a geriatric patient with an acute L tibial fracture, secondary to a fall, with AHPMN and CHF.

Case Description: Patient History & Systems Review

The patient was a retired 77 year-old male who was married and lived with his spouse and was surrounded with good family support. He lived in a private home with three steps to enter and a flight of stairs inside to reach the basement. The patient required the use of a rollator for ambulation and daily mobility. Overall health was self-rated as fair and the patient denied any major life changes during the past year, with the exception of his most recent fall and subsequent lower left extremity (LLE) ORIF. The patient gave consent for participation in both PT treatment as well as a subsequent case report by the author. The patient denied both smoking and drinking and remained as active as his mobility and independence would allow. There was no known family history of heart disease, hypertension, stroke, diabetes, cancer, psychological issues, arthritis, or osteoporosis per the patient's best knowledge; however, he was diagnosed with AHPMN and CHF years prior to his fall and PT. He had difficulty with locomotion/movement including gait on level surface, stairs, getting in and out of her car, and transitioning from sitting to standing. Additionally, he had difficulty with self-care including donning and doffing his socks. Because of difficulty with these tasks, the patient required assistance at times, and therefore, was not fully independent. The patient denied taking any medications, and was on a low sodium diet for his CHF with good effect. He had an X-Ray and ORIF following his fall, but otherwise denied any further clinical tests being performed within the past year. The primary goal of the patient was returning to and optimizing his functional mobility, both at home and in the community. Information regarding the Systems Review can be found in Table 1.

Clinical Impression #1

According to the International Classification of Functioning, the patient's primary problem or health condition was a L tibial fracture. Impairments related to both body structure and functions included decreased postural control, forward-flexed genu recurvatum gait with foot slap, decreased gross LE strength, decreased passive range of motion (PROM), and increased pain with activity.[7] The patient was

limited in functional activities such as negotiation of stairs, ambulation, getting in and out of the car, and had decreased activity tolerance due to poor exercise endurance. The patient was diagnosed with an acute L tibial fracture, with no differential diagnoses necessary. Additional information needed from the patient was information on the patients co-morbidities including AHPMN and CHF. The plan for examination was to address basic functional movements (including balance, posture, and gait pattern), which express the patient's movement efficiency, movement patterns, and functional strength through a task-related approach. Flexibility/range of motion (ROM) and LE strength was assessed additionally to better understand how any ROM and strength restrictions may impact the patient's functional mobility. The patient was a good candidate for a case report due to the abundant impairments and unique co-morbidities, including AHPMN and CHF, in conjunction with an acute L tibial fracture.

Examination – Tests and Measures

The examination started by evaluating patient's functional movements, basic transfers (such as moving from sitting to standing), and normal stance with a rollator to assess balance. A general observation of the patient's standing posture and gait pattern was observed next. Subsequently, active LE knee flexion, knee extension, ankle dorsiflexion, and ankle plantarflexion were assessed for active range of motion (AROM). PROM was assessed for the patient's soleus and gastrocnemius musculature. Active ROM of the knee was assessed with goniometry, as described by *Measurement of Joint Motion: A Guide to Goniometry*.[8] Strength was tested through manual muscle testing (MMT), with techniques as described by *Muscles: testing and function with posture and pain*, which was performed on bilateral LE's, and strength of dorsiflexors and plantarflexors to assess myotomes L4 and S1, respectively.[9] Information regarding tests and measures can be found in Table 2.

The patient's past medical history was significant for both AHPMN and CHF, with the latter of which controlled through a low-sodium diet. The patient started wearing ankle foot orthoses (AFO) since his recent injury that resulted in the acute L tibial fracture. Upon the systems review, the patient

demonstrated substantial deficits in dynamic and static standing balance and in his LE's, assesed through gait and in standing with his rollator, and severe range of motion restrictions in his gastrocnemius and soleus musculature; Strength was assessed bilaterally through MMT; however, there is great variability in the assessment of strength of patients with lower limb related dystrophic muscle impairments.[10] The patient used a rollator for ambulation, with gait observed as forward-flexed with bilateral genu recurvatum. Lower Extremity Functional Scale (LEFS) revealed a self-rated 31.25% functional level upon the day of his initial evaluation, suggesting self-perceived impaired functional mobility (with a 100% self-rated functional level meaning no self-perceived functional difficulty/deficits). The patient's activity limitations included difficulty getting in and out of bed, indoor and outdoor ambulation, negotiation of stairs, decreased strength, and decreased activity tolerance. The patient denied any restrictions in participation of various activities.

Clinical Impression #2

Based on the examination data, the initial impression regarding the patient's acute L tibial fracture and resulting ORIF surgery could be confirmed. The medical diagnosis provided in the referral was for "pain in joint, lower leg," with an ICD-9 code of 719.46 (ICD-10 code M25.569, "pain in unspecified knee"). This was for the region of the left (L) knee.

Both of AHPMN and CHF diagnoses caused a modified plan of action and subsequent treatment due to their effects on aerobic capacity, activity tolerance, and gross muscular strengthening. The plan of action included proceeding with planned interventions, however with modifications made, which shifted from basic strengthening related interventions to also include dynamic balance, gait training, and using compensatory strategies to improve independence with mobility in the community and at home.

This patient continues to be a good candidate for a case report because of their unique presentation with rare comorbidities.

The patient had a fair prognosis for improvement with PT. The diagnoses of AHPMN and CHF negatively affected the patient's prognosis, extending his projected duration of PT. Based on the patient's low-level functional mobility and significant medical history of AHPMN and CHF, the prognosis was more impacted negatively. Some research states that patients presenting with dystrophic muscles (as this patient presented with) may be able to tolerate higher intensity exercise.[11] However, the research for patients with AHPMN remains very much limited, especially in the areas of strength improvements. The patient was highly motivated and was able to physically manage his AHPMN and CHF diagnoses for approximately 10 years prior to having his recent tibial fracture injury, thus improving his prognosis.

The plan for PT was to retain the patient to initiate a rehabilitation program. There is little to no research on rehabilitation concerning AHPMN in conjunction with CHF, especially those suffering a tibial fracture repaired by ORIF. Procedural interventions included therapeutic exercise, functional training in self-care and home management, and manual therapy techniques such as manual stretching and PROM. This involved strengthening of the LE's and trunk, especially the hip abductors, knee extensors, and trunk extensors, increasing activity tolerance, gait training, and increasing range of motion, all of which related to meeting his goals of increased independence during household and daily mobility. A follow-up evaluation was planned four weeks after his initial evaluation, which involved a reassessment of flexibility, strength, gait, posture, and a LEFS questionnaire.

Interventions

Coordination and communication included contacting the referring physician to discuss and clarify the patient's significant medical history, as this affected patient care. Other aspects included data collection and analysis through outcome measures, and documentation to state the outcomes of interventions. The documentation aspect of the POC included electronic medical record documentation through TherapySource®†, with re-evaluations completed every 30 days or 10 visits. Patient/client related instruction included education on the patient's current condition, including anatomy and physiology of bone fractures, typical fracture healing time, and the plan to address the patient's functional mobility deficits while at PT.

Upon the initial evaluation, interventions began at an extremely low level, due to the patient's limited exercise tolerance, limited strength, and limited aerobic capacity. These interventions involved both isometric and isotonic strengthening exercises, and supine ROM exercises. Isometric exercises included quadriceps sets and hip adductor isometrics while isotonic exercises included Red Thera-Band™* (medium resistance) resisted ankle plantarflexion, standing knee extension and marching exercises, and supine hip adduction/abduction. Supine range of motion exercises included supine knee flexion. The chronology of interventions per visit remained the same in that supine ROM exercises were performed first, followed by supine isometric strengthening exercises. The patient then transitioned from isotonic exercises in the supine position to that of a standing position. This chronology was chosen in order to prepare the patient for more taxing interventions. As the patient was able to progress from isometric and supine ROM exercises to increased standing, closed chain activities, the chronology remained the same; beginning with more basic interventions such as closed chain strengthening and progressing to dynamic balance and gait training activities.

† Source Medical. 100 Grandview Place, Suite 400, Birmingham, AL 35243.
* The Hygenic Corporation, 1245 Home Avenue, Akron, OH 44310, with the medium resistance band model number being 7168-02.

A large aspect of the interventions performed by the patient involved dynamic balance activities, specifically dynamic balance. For patients with the diagnosis of hereditary sensory motor neuropathy, research shows that balance exercises can be statistically beneficial. Research by Matjacić and Zupan shows statistical improvement in Berg Balance scores following dynamic balance training during both standing and stepping.[12] The interventions from this research were completed within a 12-day timeframe, indicating that significant improvements in balance can be made in a 12-week period of PT, similar to the duration for this patient's plan of care (POC). In the same population of patients with hereditary motor and sensory neuropathy, strength training three times per week for 24 weeks can result in moderate strength increases including increased knee torques, and overall leg-related functional performance.[13] This study spanned a 24-week period with a strength training frequency of three times per week. The strength was measured/evaluated by isokinetic knee torque, and functional performance through both timed motor performance and questionnaires on functional performance. No timed motor performance improvements were noted with the hereditary motor and sensory neuropathy group. [13] However, based on the results of an increased knee torque and questionnaire improvements, strength training remains a viable intervention option to improve overall function.

The reason for the progression from basic isometric/isotonic strengthening and ROM to closed chain activities including strengthening, balance, and also gait training was to improve the patient's functional mobility through task training and simulation. The use of closed chain activities added a dynamic challenge to the patient's overall strengthening program. Based on the patient's significant medical history, a compensatory strategy was implemented to help the patient better progress toward his goals of more independence in functional mobility. This included standing marching, hip abduction, hip extension, and knee flexion, squats using parallel bars for assistance, gait training, and stair simulation using parallel bars via forward and lateral step-ups using a six-inch and four-inch box, respectively. More information regarding interventions can be found in Table 4 and Appendix 1.

Modifications to the POC involved utilizing increased rest breaks due to decreased aerobic capacity and activity tolerance, and including the use of parallel bars for exercises that required balance, since the patient required a rollator walker for ambulation and general mobility. The initial exercise intensity level was reduced to a lower level to meet the patient's ability at the time, since the patient demonstrated significant strength deficits. Based on the patient's reported difficulty with functional mobility, the anticipated goals shifted from their initial focus on a return to overall independent status to new goals including maximizing increased independence with household and daily mobility and facilitating the return to a pre-morbid mobility level. Information regarding goals can be found in Table 3.

The patient's PT plan of care was approximately 12 weeks long, with two sessions per week, for a total number of 24 treatment sessions. Each PT session lasted approximately 45-60 minutes long. The patient was compliant with PT throughout his stay. The 24 treatment sessions represents the amount of time the author was present for the patient's PT, however, the patient continued PT upon the departure of the author.

Outcomes

The patient was assessed using the LEFS, gait pattern assessment, strength assessment through MMT of knee extension, hip flexion, and hip abduction. Upon re-evaluation performed at visit number 27, the patient demonstrated improvements in lower extremity strength and gait pattern categories. The LEFS remained at the same level of self-perceived functioning. Specific information regarding outcomes can be found in Table 5. Specific information regarding the patient's gait pattern can be found in Figure 1.

Discussion

AHPMN presents with the primary symptoms of proximal weakness and muscle wasting of the limbs, more commonly in the lower extremities, in addition to gait unsteadiness and difficulty standing.[1, 2] The patient, with an AHPMN diagnosis, presented with decreased lower extremity strength, decreased lower extremity self-perceived function, difficulty with transfers, and a compensatory gait pattern. Considering fall risk factors include, but are not limited to, older adults (age 65 or greater) and those with a diagnosis of CHF, this case was particularly noteworthy because the patient had both of these risk factors with the added degenerative neuromuscular diagnosis and had, in fact, sustained a fall.[4, 5]

Although the patient's self-rating of perceived function through the LEFS remained the same, the patient demonstrated improvements in functional mobility, thus making this case report demonstrate its intended purpose. This was relevant due to the prevention of falls, which this patient was not only at risk for due to his co-morbidities, but personally experienced prior, resulting in physical therapy.

The potential contributing factors to improved outcomes were patient motivation, improved gait pattern, improved dynamic balance, and improved strength. The patient's high level of motivation resulted in quality therapy sessions thus maximizing the potential outcomes of the therapy performed. Strength and dynamic balance improvements may have helped carry over to functional mobility by supplementing activities where they are required, such as gait, which also improved throughout therapy.

Suggestions for future work related to this project could involve the utilization of functional testing in instances of acute injury in the presence of chronic conditions and proactive referral for training in fall reduction strategies for individuals identified to be at risk.

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Tables and Figures

Table 1. Systems Review

Cardiovascular/Pulmonary	Impaired: Decreased activity tolerance and endurance were present with a diagnosis of CHF. Vital signs were within normal limits.
Musculoskeletal	Impaired: Gross symmetry presented as normal along the frontal plane, and with increased thoracic kyphosis along the sagittal plane. Generalized gross strength presented as significantly decreased with right (R) LE more impaired than the L LE.
Neuromuscular	Impaired: Gait/locomotion presents as genu recurvatum with foot slap, impaired on the R side greater than the L side. Forward trunk lean was present throughout ambulation with use of a rollator and bilateral AFOs.
Integumentary	Not impaired.
Communication	Not impaired.
Affect, Cognition, Language, Learning Style	Not impaired. The patient had good affect, with no observable barriers to learning. The patient preferred demonstration for optimal learning style.

Legend 1. Acronyms defined as: CHF (congestive heart failure), R (right), L (left), LE (lower extremity), AFO (ankle foot orthotic).

Table 2. Tests & Measures

Tests & Measures	Initial Evaluation Results	Left	Right
Lower Extremity Manual Muscle Testing	Knee Extension	2/5	1+/5
	Knee Flexion	4+/5	4+/5
	Ankle Dorsiflexion	4/5	4/5
	Ankle Plantarflexion	5-/5	5-/5
Goniometry	Knee extension	2°	126°
Lower Extremity Functional Scale	Total percentage score: 31.25% of self-perceived function.		

Legend 2. There is no current available reliability or validity for lower extremity manual muscle testing.[9] Lower Extremity Functional Scale test-retest reliability and construct validity was found to be 0.86 and 0.80, respectively, with a minimal detectable change value of 9 points.[14] Goniometric inter-rater and intra-rater reliability was found to be 0.57-0.79 and 0.91-0.96 for PROM of knee extension, respectively.[15] The intraclass correlation coefficient (ICC) validity for use of the universal goniometry for knee flexion and extension was found to be 0.99.[16]

Table 3. Goals

Short term goals:
The patient will be able to navigate 3 steps into house with step-through pattern, using bilateral railings as needed.
The patient will demonstrate moderate restriction in his gastrocnemius and soleus musculature so that he is able to position his ankles neutrally allowing for more upright posture throughout gait.
The patient will demonstrate gross lower extremity strength through MMT graded at 3/5 so that he is able to lift his legs to get in and out of bed independently and without difficulty.
Long-term goals:
The patient will be able to navigate a full 14-step flight of stairs into the basement without difficulty reported.
The patient will demonstrate mild restriction in his gastrocnemius and soleus musculature so that he prevents genu recurvatum during ambulation making gait more efficient.
The patient will demonstrate gross lower extremity strength through MMT graded at 4/5 so that he is able to weight shift appropriately making ambulation less difficult.

Legend 3. The definitions mild and moderate were defined in the electronic medical records system at patient's clinic, with definitions including none, slight, mild, moderate, and severe. The MMT acronym is defined as manual muscle testing.

355 Table 4. Interventions.

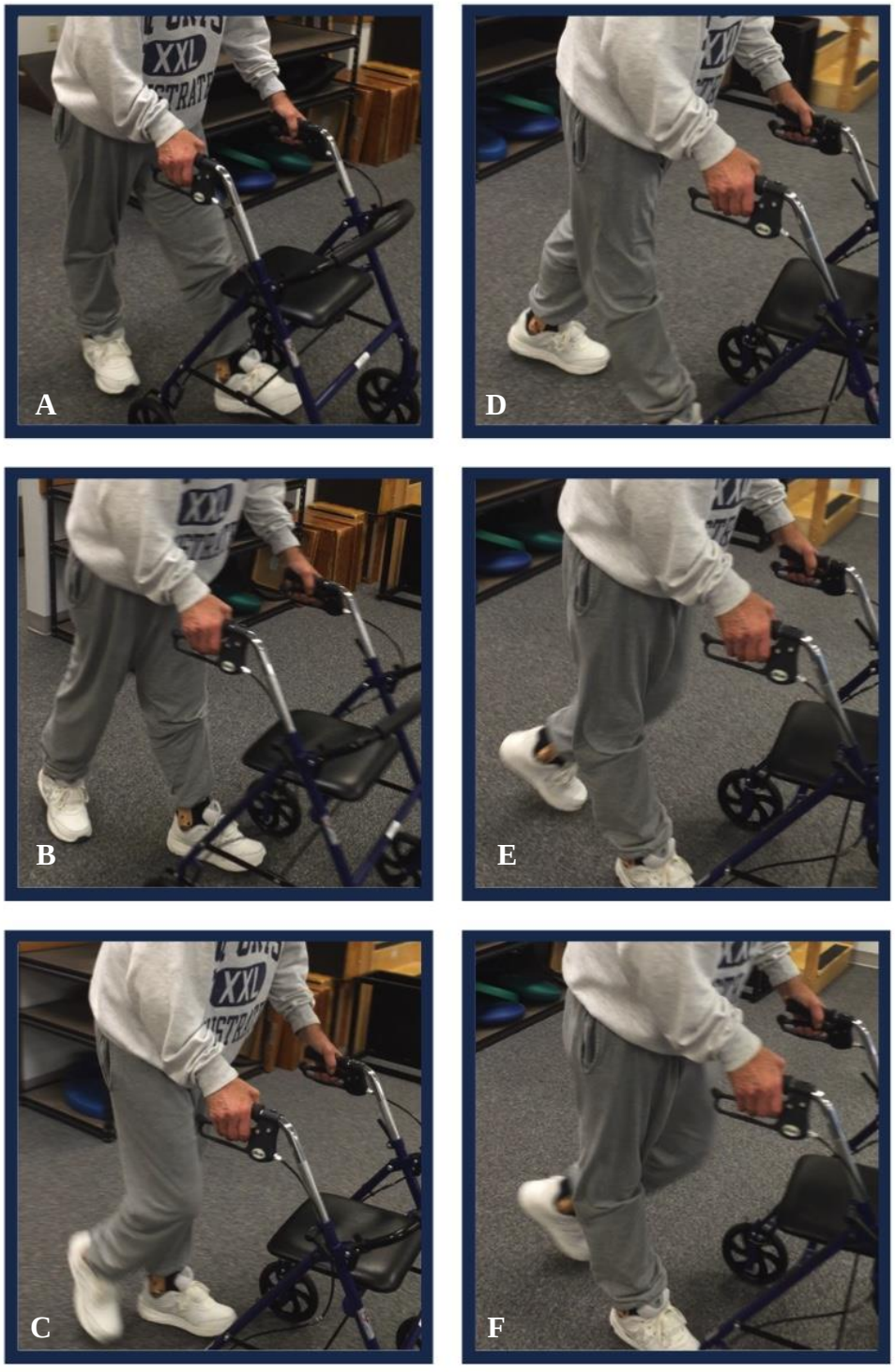
Interventions	**Treatment Sessions**							
	1-4	**5-8**	**9-12**	**13-16**	**17-20**	**21-24**	**25-28**	**29-32**
Heel slides in supine position	X	X	X					
Quadriceps squeezes in supine position	X							
Hip abduction/adduction in supine position	X	X	X	X				
Bridging in supine position		X	X	X				
Seated adductor squeezes with pillow between knees		X	X	X	X	X	X	
Standing hip abduction, extension, flexion, and knee extension, flexion	X	X	X	X	X	X	X	X
Standing from a seated position (sit-to-stands)			X	X	X	X	X	X
Standing balance in parallel bars				X	X	X	X	X
Stair simulation							X	X
Flat and inclined floor gait training						X	X	X
Forward/lateral stepping strategies							X	X

Table 5. Patient Outcome Measures

Outcome Measure	**Initial Evaluation**		**Re-evaluation**	
A. Lower Extremity Functional Scale	31.25%		31.25%	
B. Gait Pattern	Severely forward flexed trunk, with moderate bilateral genu recurvatum during weight bearing.		Moderately forward flexed trunk, with moderate-mild bilateral genu recurvatum during weight bearing.	
Manual Muscle Testing	Left	Right	Left	Right
C. Knee extension	2/5	1+/5	3-/5	3-/5
D. Hip flexion	2+/5	3-/5	3/5	3+/5
E. Hip abduction	4-/5	4-/5	4/5	4/5

Legend 5. (A) The Lower Extremity Functional Scale (LEFS) demonstrates self-assessment of function of impaired LE. The LEFS is based on a percentage of 0-100%, with 0% being completely dependent and 100% being fully independent and functional with no difficulty with any form of functional mobility. (B) The gait pattern described was with the use of a rollator walker. Improvements were noted with increased upright torso and less dependence on upper extremity weight bearing through the assistive device, and decreased knee hyperextension (genu recurvatum). (C, D, and E) Knee extension, hip flexion, and hip abduction manual muscle testing (MMT) was assessed using the basic MMT scale of 0-5. The patient was unable to extend either knee through a gravity-dependent position (i.e. seated) upon the initial evaluation. The re-evaluation revealed the patient's increased ability to perform knee extension through full AROM, in addition to having strength to tolerate slight overpressure. Note: The re-evaluation was performed at visit number 27 when the author was no longer present.

Figure 1. Phases of Gait



Legend 6. The following photographs demonstrate the patient's typical gait pattern. These pictures show several specific phases of gait, specifically to the patient's left lower extremity. The phases of gait described and identified in the photographs are A) initial contact B) loading response C) midstance D) terminal stance E) pre-swing F) initial swing. This gait pattern shows the patient with a forward flexed trunk, and excessive plantar flexion and knee extension (genu recurvatum) during the loading response and midstance phases of gait.

Appendix 1. Regarding treatment sessions: Heel slides for active assistive range of motion included the use of a towel to improve knee flexion, and quadriceps squeezes with the use of a rolled towel for proper tactile cueing. Supine hip abduction/adduction was performed with a board and towel. Standing hip and knee exercises (hip abduction, extension, flexion, and knee extension, flexion) were all performed in parallel bars, with standing from a seated position performed with the use of the patient's rollator walker to follow a compensatory mobility strategy. Foot positioning in the parallel bars included both normal and semi-tandem stances. Stair simulation included the use of six-inch steps with bilateral railings and involved forward stair negotiation. Gait training included weight shifting for dynamic balance and an increase in floor inclination to both increase workload and functionally challenge the patient. Contact guard assistance was given during gait with verbal cues for an upright torso. The patient began ambulation with the use of a single point cane by visit 27. Stepping strategies were performed in parallel bars, including stepping over a six-inch box forwardly, and a four-inch box laterally. This also included sidestepping up an inclined surface with the use of a railing and contact guard assistance. All exercises performed with a 10-20 repetition range, for two to three sets with rest breaks given between sets and exercises as needed. This excludes standing balance in parallel bars, stair simulation, flat and inclined floor gait training, and forward/lateral stepping strategies. Standing balance in parallel bars consisted of two to three 30-60 second sets with each of the foot positions stated previously. Stair simulation involved three sets of upward and downward negotiation of three 6" steps with bilateral railings. Both forward and lateral stepping strategies were performed for two to three sets of five repetitions.