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The patient signed an informed consent allowing the use of medical information and 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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Background and Purpose: Charcot Marie Tooth Disease (CMT) is the most common progressive inherited neurological disorder[1]. Characteristics include muscle weakness and reduced sensation beginning in the distal lower extremities. Individuals with CMT have increased difficulty sensing and maintaining balance. There is minimal research on the effectiveness of balance training and outcome measures in this population. The RUSK Hospital Modified Romberg Protocol (RUSK MRP) is used as a balance-training program for patients with a neurological diagnosis, however no information is available on its effectiveness in CMT. The purpose of this case report was to investigate the effectiveness of the RUSK MRP in a patient with CMT.

Case Description: The patient was a 60-year-old male diagnosed with CMT over 40 years ago and reported more than seven falls in the last six months. The RUSK MRP intervention included balance training with foot placement variation, surface type and visual cues as well as strengthening and mobility training (See Appendix A) twice a day for seven days over 12 weeks.

Outcomes: The patient demonstrated improvement in balance per Berg Balance Scale score from 31 at initial evaluation to 41 at discharge. RUSK MRP balance improved from 20 seconds at six inches apart to ¾ Romberg for 1 minute (See Appendix A).

Discussion: CMT can be a debilitating disease that causes significant balance challenges. The RUSK MRP was found to be successful in the treatment and outcome evaluation of a 60-year-old patient with CMT. Although the patient made improvements in his balance,

further research is needed to assess the validity and reliability of the RUSK MRP.

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Background and Purpose

Charcot Marie Tooth Disease (CMT) is the most common progressive inherited neurological disease, affecting 1 in 2,500 individuals in the United States.[1] CMT is a genetic disorder in which the genetic encoding of proteins in myelin sheaths in both motor and sensory nerves become damaged. The distal lower extremities (LE) are most affected, followed by the distal upper extremities (UE). Signs and symptoms include: muscle wasting, reduced sensation, skeletal deformities and gait abnormalities. It is important to note that the progression of CMT is slow and symmetrical. Individuals with CMT have difficulty performing activities of daily living (ADL) and maintaining balance, resulting in an increased risk for falls. There is currently no cure for CMT, but management of the disease is maintained with medication and physical therapy.

It is currently unclear which interventions or exercise prescriptions are most effective with this patient population.[2] Generally, studies have focused on strength training to help improve patient function, with mixed results. One study examined the effects of a 12-week home-based upper and lower extremity strength-training program in regards to improved ADL participation.[3] The program focused specifically on knee and elbow flexion and extension muscles (See Appendix A for detailed exercise prescription). Patients demonstrated improved muscle strength at the conclusion of the study. A six-month follow up demonstrated that individuals who continued and discontinued the exercise program had a reduction in muscle strength. However, ADL performance was maintained in the group that continued with the home exercise program compared to those who didn't. Another study examined the effects of a hip flexor-strengthening program.[4] The results from this study demonstrated that patients improved right hip

flexor strength but there was no change in left hip flexor strength. The authors from this study believe that there may have been no statistical significance due to lack of exercise guidelines in this patient population.[4]

Studies have demonstrated the importance of sensation in terms of static and dynamic balance in patients with CMT.[5,6] However, there is minimal research on the effectiveness of a balance-training program in this patient population. In patients with a neurological disorder, clinicians at RUSK hospitals outpatient neurological clinic employ a balance program. The RUSK Hospital Modified Romberg Protocol (RUSK MRP) varies the patient's base of support, surface, and vision (See Appendix A for exercise progression). Depending on the patient's safety, this program may be performed at home or in the clinic under the direct supervision of the clinician. Although this exercise regimen is regularly given to patients, there are no reports on its effectiveness. The purpose of this case report was to determine the effectiveness of the RUSK MRP in order to improve balance and reduce risk of falls in patients with CMT.

History and System Review

CB was a sixty year-old male with a diagnosis of CMT since 1972. His past medical history included, skin cancer (currently in remission), a family history of CMT, femoral fracture (8 years ago), facial paralysis and gold weight surgery to bilateral eyelids. Two years prior to the evaluation date, CB received physical therapy (PT) in order to address balance impairments. His prior course of PT treatment primarily focused on LE strengthening and compensatory strategies. CB reported improvements in balance at that time. However, within the last year, he had multiple falls. Due to his feeling of

imbalance, CB restricted his overall activity levels. He became fearful of walking to the grocery store and entering and leaving his home.

During the fall months, CB and his spouse lived in an apartment high rise with a ramp at the entrance and an elevator. During the summer months, CB and his spouse lived in a single-story home with fourteen steps and bilateral handrails with no ramp access. CB's goal for PT was to improve his balance, improve his ability to negotiate obstacles outdoor, and gain greater independence negotiating stairs. Upon his PT evaluation, CB signed an informed consent to authorize the release of his medical information for the purpose of this case report.

Clinical Impression #1

CB had a clinical diagnosis of CMT and presented to physical therapy with signs and symptoms related to this diagnosis. Tests and measures used to effectively rule in the patients diagnosis included touch awareness, proprioception, kinesthesia, manual muscle testing (MMT), and skeletal deformities, specifically to the distal UE and LE. Due to the patient's reported increase in falls, the Berg Balance Scale (BBS) and the Dynamic Gait Index (DGI) were also administered to determine his fall risk potential. CB was an appropriate candidate for this case report due to his demonstrated improvement with past physical therapy. Although CB received positive results from a plan of care focused on strength training, this balance program may yield a long-term positive result.

Tests and Measures

Touch awareness was performed with a cotton ball in order to determine the patient's perception of tactile touch input. Touch awareness was found to be impaired from the patient's great toe bilaterally to bilateral knees. Proprioception and kinesthesia

were also performed and found to be impaired at the great toe and ankle bilaterally. A gross MMT was performed to bilateral LE. Testing revealed impaired strength to the ankle dorsiflexors and plantarflexors bilaterally (See table 2). CB wears bilateral solid ankle foot orthosis (AFO), which were worn during his initial visit. When the patient removed his solid AFO's, shoes and socks pes cavus was noted bilaterally. Skeletal deformities were also noted in the patient's carpal and metacarpal bones.

The BBS is a 14-item measurement that is used to assess balance[7]. When performing the BBS, the patient demonstrated difficulty completing the exam, specifically with those measures requiring standing for extended periods of time. For example, the patient was only able to stand unsupported for 30 seconds before requiring assistance to maintain his balance. The maximum score for this portion of the exam is to stand unsupported for 2 minutes. CB also performed the DGI, which also quantifies the patient's fall risk potential. The advantage of the DGI in comparison to the BBS is that it gives a representation of the patient's balance in dynamic situations.[7] These dynamic situations encompass multi-tasking abilities when ambulating. The patient's score on the DGI (see Table 3) indicated that he is a high fall risk. CB specifically had a challenging experience when performing horizontal head turns, vertical head turns, and stepping over obstacles when walking. When the patient would lose his balance on these measures, he would fall posteriorly, requiring the assistance of the student clinician to maintain his balance. Both the BBS and DGI have high reliability and validity in determining fall risk potential.[7,8]

Clinical Impression 2

The findings from the examination were consistent with the patient's medical diagnosis of CMT and ICD-10-CM code of G60.0 "Hereditary motor and sensory neuropathy." The patient continues to be appropriate for this case due to his diagnosis being consistent with the literature, but interventions to improve balance remain unknown. Since ambulating safely was one of the patient's primary goals, it was important to take this into account. Intervention incorporated functional exercise in addition to a home exercise program. However, the primary focus of the interventions was the implementing RUSK MRP. The plan was to see the patient twice a week for 10 weeks. Each appointment was an hour in length and one-on-one. Table 5 lists the therapeutic goals for this course of physical therapy treatment. The patient's prognosis was fair secondary to the progression of his disease, his past success with PT, motivation to improve and family support.

Intervention

CB was seen in the clinic twice a week for half hour appointments over the course of 12 weeks. The patient did not receive any other services aside from physical therapy and the patient's family received no training. Functional updates were sent to CB's primary care physician on a monthly basis. The primary intervention in the clinic focused on balance training, specifically utilizing the RUSK MRP (See Appendix A) in order to decrease the patients fall risk. Currently there is no research on the effectiveness, reliability, or validity of the RUSK MRP in regards to improvement in balance. However, the RUSK MRP is partly based on the Romberg and Sharpened Romberg Test, which are used to assess fall risk potential. With the Romberg Test, an individual is asked to stand

with feet together and eyes closed for one minute. Individuals who are unable to maintain this position without excessive swaying and eyes closed for one minute are considered a fall risk.[11] Researchers have found that there may be a ceiling effect when utilizing the Romberg Test[11]. Thus, a modified version named the Sharpened Romberg has been used. The Sharpened Romberg further narrows the patients base of support and has the individual stand on both compliant and non-compliant surfaces.[12]

The RUSK MRP exercise program focuses on various parameters such as base of support, surface the patient is standing on, and whether eyes are open or closed for one minute. For example, if a patient is unable to maintain the Romberg testing position, then a patient begins with eyes open, feet shoulder width apart, and standing on a firm surface. The patient is asked to hold this position for one minute before any changes to the various parameters are made. As the patient progresses, the base of support becomes narrower. The surface can be compliant or non- compliant, and eyes may be open or closed depending on how well the patient tolerates the position. An individual is then instructed to maintain a particular position for 60 seconds (Refer to Appendix A for progression of RUSK MRP). If a patient is unable to hold the position for 60 seconds, they do not advance to the following position and are instructed to perform four repetitions of the exercise for as long as they can.

CB was initially unable to achieve the Romberg position and required modifications to his position. CB began with feet shoulder width apart, standing on a stable surface, and eyes open for one minute. As CB progressed, adjustments were made to his base of support and the standing surface. Adjustments in regards to vision were unable to be performed secondary to the patient demonstrating difficulty maintaining

eyes closed for an extended period of time. The patient reported that keeping his eyes closed had been difficult since developing facial paralysis and his recent gold weight surgery.

A secondary issue that was addressed with therapy was CB's decreased strength as noted in Table 2. A Home Exercise Program (HEP) based on hip strengthening was given in order to improve walking distances and balance. The HEP was based on past research that found strengthening of the proximal LE musculature could improve gait distances and balances. [3,4] The patient was instructed to perform bilateral standing hip flexion, hip extension, and hip abduction for 3 sets of 10 repetitions. CB and his spouse were reminded of the importance of following the HEP in order to continue making advances in function. Adjustments were made in terms of exercise prescription, repetitions, and frequency as the patient perceived improved strength and form. See Table 4 for changes in frequency and intensity to CB's HEP.

CB demonstrated difficulty rising from a standard chair without assistance secondary to impaired LE strength. Therefore, sit to stands were performed in the clinic with the use of an adjustable mat table. Sit to stands are a functional exercise that focuses on the patient using lower extremity musculature to achieve a standing position. In addition, this exercise uses musculature to help control the descent when returning to sitting. This exercise focused on technique in order for the patient to gain the greatest advantage for success with this task. The patient was instructed to sit near the edge of the table, place both feet behind both knees, and use "nose over toes" to reach a standing posture. Nose over toes is a position in which the patient flexes his trunk to the point that his nose is over his toes. Close attention was given to ensure CB did not push both knees

against mat table when coming into full standing position. This reduced the likelihood of CB falling posteriorly when standing from a seat with less support.

Outcomes

CB met all long and short-term goals established for physical therapy (Table 5) and statistical significance was observed in both the BBS and DGI outcome measure (see Table 3). As CB progressed in therapy, there was a notable improvement in his ability to maintain his balance when performing the RUSK MRP in the clinic. Initially, he was unable to perform the exercise, but with improved balance he was able to reduce his base of support to the ¾ Romberg position and stand on a non-stable surface (Table 4). Improvements were demonstrated in his technique with sit-to- stand and his ability to independently rise from lower seat heights that required increased LE muscle strength and balance (Table 4). Slight improvements in gross muscle strength were noted in bilateral hip flexion, extension, and abduction. However, no improvements in strength were demonstrated with bilateral knee flexion, knee extension, ankle plantar flexion, and ankle dorsiflexion (Table 2).

Discussion

The purpose of this case report was to demonstrate an improvement in balance utilizing the RUSK MRP in a patient with CMT. There is currently no research on the effectiveness of a balance program in patients with CMT.[2] Studies have attempted to improve patient function via strength training of the LE. [2,3,4,5] The patient in this case report demonstrated an improvement in balance per the BBS and DGI, indicating that CB was at a decreased risk of falls. The clinical implication resulting from this case report is

rather than focusing on strength training, clinicians may use this balance program to reduce the risk of falls and improve functional outcomes.

Despite CB's improvements, it is unclear if the RUSK MRP was the primary contributor to the patient's improved balance. In addition to the RUSK MRP, the patient was given a HEP to improve hip flexion, extension, and abduction muscle strength. Another intervention used in CB's rehabilitation program was the functional strength-training exercise of sit-to-stands. With both of these interventions, the patient demonstrated improvements in gross muscle strength and functional strength, as noted in the outcomes section of this paper. Studies have demonstrated an improvement in gross strength and functional activities through strengthening,[2-6] but have not demonstrated any carryover to improved balance.

Further research is needed to determine if the RUSK MRP is a sufficient intervention to improve balance in patients with CMT. In order to ascertain that the RUSK MRP was the primary contributor to improvements in balance with CB, future studies should attempt to use the RUSK MRP in singularity. In addition, future research should attempt to replicate the improvements noted in this case report to patients with various onsets of CMT. The patient in this case report had CMT for over 40 years. Patients who have been diagnosed at an earlier age may respond differently to the RUSK MRP compared to CB. Interventions for patients with CMT are limited and studies are needed that investigate the best available intervention for this patient population.

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Table 1. Systems Review

Cardiovascular/Pulmonary	**Heart rate and blood pressure within normal limits**
Musculoskeletal	Gross strength and range of motion impaired in bilateral ankle and knee. Skeletal deformities noted in bilateral upper extremities. Pes cavus bilaterally. Bilateral UE fine motor skills absent. Trendelenburg gait.
Neuromuscular	Light touch impaired bilateral lower extremities, from great toe to knee. Proprioception impaired in bilateral great toe and ankle.
Integumentary	No impairments noted
Communication	No impairments
Affect, Cognition, Language, Learning Style	No impairments

Abbreviations: UE= Upper extremity, LE= Lower extremity

Table 2. Manual Muscle Testing

Joint	**Motion**	**Left**	**Right**
Hip	Flexion	4-/5 at admission 4/5 at discharge	4-/5 at admission 4/5 at discharge
	Extension	Able to perform Bridge	Able to perform Bridge
	Abduction	3-/5 at admission 3/5 at discharge	3-/5 at admission 3/5 at discharge
Knee	Flexion	Seated: against mod resistance	Seated: against mod resistance
	Extension	4-/5 at admission unchanged at discharge	4/5 at admission Unchanged at discharge
Ankle	Dorsiflexion	Absent at admission Unchanged at discharge	Absent at admission Unchanged at discharge
	Plantarflexion	Initiates only (at admission) Unchanged at discharge	Initiates only (at admission) Unchanged at discharge

Table 3: Outcome Measurements

Outcome Measure	At Admission	At Discharge
Berg Balance Scale	31/56	41/56
Dynamic Gait Index	9/24	19/24

Table 4. Intervention progression

	Weeks 1-3	**Weeks 4-6**	**Weeks 7-9**	**Weeks 10-12**
Sit to stand	23" mat table 3 x 10	22" mat table 3 x 10	21" mat table 3 x 10	20" mat table 3 x 10
Romberg	Feet shoulder width apart, firm surface, eyes open for one minute	Feet 2" apart, airex, eyes open for one minute	Romberg, airex, eyes open, for one minute	¾ Romberg, airex, eyes open, for one minute
HEP performed bilaterally (Hip Flexion, Hip Abduction, and Hip Extension)	2 x 10 each Seated: Hip Flexion, Hip Abduction	Standing Hip Flexion & Hip Abduction 2 x 10 each Hip Extension 1 x 10	Standing Hip Flexion, Hip Abduction, and Hip Extension 3 x 10 each.	Standing Hip Flexion, Hip Abduction, and Hip Extension 3 x 10 each.

Table 5 Physical Therapy Goals

Short Term Goals (5 weeks)	Long Term Goals (10 weeks)
The patient will perform sit to stand from 21" mat table independently 3/5 times with no upper extremity support in order to demonstrate improved safety with transfers in the community.	The patient will perform sit to stand from 18" chair with independence 3/5 times with no upper extremity support in order to demonstrate improved safety with transfers in the community.
The patient will improve his score on the Berg Balance Scale from 36/56 to 42/56 in order to demonstrate decreased risk for falls.	Patient will score 50/56 on berg balance test in order to demonstrate decreased risk of falls in the community and at home.

The patient will ambulate 100' with the least restrictive device with distant supervision with no loss of balance 5/5 times in order to reach his mailbox at home.	The patient will ambulate 2 city blocks with least restrictive device with distant supervision with no loss of balance in order to access grocery store.

Appendix A

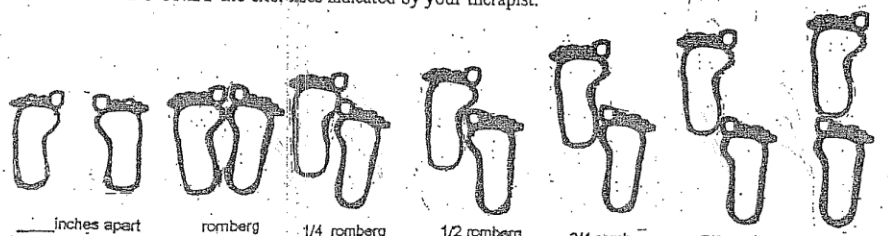
BALANCE EXERCISES

RUSK INSTITUTE OF REHABILITATION

Vestibular Rehabilitation Program 212.263.8466

Name:_____ Date:_____ Therapist:_____

Perform the following exercises ____ times per day.
Do **ONLY** the exercises indicated by your therapist.



☐ 1. Stand with your feet in the _____ position.
It does not matter which foot is in front.
Place your arms across your chest / at your sides.
Have your eyes open / closed
Hold for ____ seconds.

☐ 2. Stand with your feet in the _____ position.
It does not matter which foot is in front.
Place your arms across your chest / at your sides.
Have your eyes open / closed
Hold for ____ seconds.

☐ 3. Stand in front of a footstool or step.
Place your _____ foot up on the step
Place your foot so that it is directly in front of the other / _____inches apart
Place your arms across your chest / at your sides
Have your eyes open / closed
Hold for ____ seconds