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Outpatient Physical Therapy Management of a Patient Diagnosed with Myotonic Muscular Dystrophy Type 2

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OUTPATIENT PHYSICAL THERAPY MANAGEMENT OF A PATIENT DIAGNOSED WITH
MYOTONIC MUSCULAR DYSTROPHY TYPE 2

A Scholarly Project Submitted to the Graduate Faculty of the

Department of Physical Therapy

School of Medicine

University of North Dakota

by

Ashley Jobe

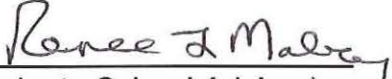
In Partial Fulfillment of the Requirements for the Degree of

Doctor of Physical Therapy


Grand Forks, North Dakota

May, 2019

This Scholarly Project, submitted by Ashley Jobe in partial fulfillment of the requirements for the Degree of Doctor of Physical Therapy from the University of North Dakota, has been read by the Advisor and Chairperson of Physical Therapy under whom the work has been done and is hereby approved.



(Graduate School Advisor)



(Chairperson, Physical Therapy)

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Department Physical Therapy

Degree Doctor of Physical Therapy

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Signature Ashley Jobe

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ABSTRACT

Background and Purpose. This article describes the 3-week outpatient Physical Therapy management of a 63-year-old female with a diagnosis of Myotonic Muscular Dystrophy (MMD) Type 2. MMD is an inherited disorder, which progressively impacts muscular, respiratory, cardiac, nervous, and endocrine systems. MMD Type 2 is characterized by muscle weakness, muscle pain, and myotonia. The patient in this case study presented to physical therapy with upper and lower extremity weakness and balance impairment. The purpose of this article is to describe the initial physical therapy evaluation process for a patient with MMD, as well as the interventions used for this patient and the patient's response to treatment. Research involving the physical therapy management of a patient with MMD Type 2 is scarce, and this article intends to add to the limited available information. **Description.** The treatment of this patient involved a generalized strengthening program for the upper and lower extremities, aerobic conditioning, balance training, and soft tissue massage for tissue restrictions. **Outcomes.** Following physical therapy intervention, the patient improved in functional outcome scores and balance assessment scores. The patient also demonstrated increased tolerance to aerobic exercise. Grip and pinch strength dynamometry were recorded revealing increased strength. Variable results were compiled from the manual muscle testing strength assessment, with a general trend of increased strength following the 3-week period of therapy. **Discussion.** The rationale for treatment was based on addressing the patient's presenting symptoms and goals. Information gathered from a thorough literature review was used to guide treatment. Treatment was altered or progressed based on the patient's response.

CHAPTER I

INTRODUCTION AND BACKGROUND INFORMATION

Genetics

The origin of Myotonic Muscular Dystrophy (MMD) is genetic. MMD results from an abnormal repetition within a gene's DNA sequence. The mutated gene is unstable, and produces aberrant messenger-RNA that clumps inside the cell and interferes with the production of many proteins.¹ These changes impede normal cell function, and lead to the signs and symptoms of MMD. Due to the etiology of MMD, the disease is further classified as a spliceopathy.² MMD is an autosomal dominant disorder. So, when a mutated gene is present it will be dominant over the paired normal gene. If an individual inherits a mutated gene, they will develop the disease. The likelihood of a parent with the disease passing the mutated gene on to their children is 50%. As the disease is passed on through generations, the signs and symptoms become more severe and begin earlier in life due to a phenomenon called anticipation.³

Two different forms of MMD have been identified. Genetic variations between the two subtypes can help with differential diagnosis. MMD Type 1 is classified by genetic mutations in the DMPK gene.¹ MMD Type 2 results from mutations in the CNBP gene. The exact functions of these genes are still unclear. However, the protein produced by the DMPK gene appears to play a role in cell communication and correct cell functioning within

the heart, brain and skeletal muscles. The protein produced by CNBP is found primarily in cardiac and skeletal muscles, where it is involved in protein synthesis and possibly the regulation of other genes. Additional genetic differences have also been found. For instance, the repeat expansion observed in MMD Type 2 is much larger than MMD Type 1.² However, the size of the repeat does not correlate with the disease severity or age of onset for patients with MMD Type 2, unlike patients with MMD Type 1. Physical examination, electromyography, and muscle biopsies are used to diagnose MMD. Genetic testing is the only definitive test, though, and is the best for determining the subtype of MMD.³

Prevalence

MMD is estimated to affect at least 1 in 8,000 people worldwide.¹ The prevalence of MMD varies by subtype and geographic location. MMD Type 1 is the most common inherited myopathy that presents in adulthood.⁴ MMD Type 1 is typically more common than MMD Type 2, with the exception subpopulations in Germany and Finland where the two subtypes are equally common. The prevalence of MMD is 3-15/100,000 people in Europe. It is extremely rare in some areas (0.9/100,000 people in Taiwan) and extremely common in others (189/100,000 in Saguenay-Lac-Saint-Jean, Canada). Since most research has been done outside of the United States of America, the exact prevalence of MMD in the United States is unknown at this time. There are currently no statistics for the prevalence of MMD Type 2 within the general population.

Presentation

MMD progressively impacts muscular, respiratory, cardiac, nervous, and endocrine systems. The most notable signs and symptoms of MMD include muscle atrophy, muscle weakness, myotonia, and muscle pain. These impairments are many times accompanied by balance deficits and difficulties with gait. The multisystem effects of MMD manifest in a wide variety of health impairments, such as cataracts, cardiac conduction deficits, and diabetes mellitus. It is important to note that these comorbidities are in fact related to the patient's diagnosis of MMD.

Not only are there differences in genetic characteristics, there are also differences in the patients' physical presentation when comparing the subtypes of MMD. This knowledge is very useful for clinicians. Patients with MMD Type 1 typically develop symptoms of the disease earlier than those with MMD Type 2. MMD Type 1 can present at birth, but most often appears in the patients' 20s or 30s.³ MMD Type 2 typically presents in the patients' 40s. Although many of their symptoms are similar, MMD Type 1 tends to be more severe. MMD Type 1 generally impacts more distal muscles including those of the legs, hands, neck, and face. Patients with MMD Type 2 typically experience involvement of more proximal muscles including those of the neck, shoulders, elbows, and hips. Prominent muscle pain, stiffness, and fatigue are more commonly experienced by patients with MMD Type 2 in comparison to MMD Type 1.⁵ These are general trends that can be helpful in coming to a diagnosis and for treating a patient with MMD, but it is important to note that symptoms vary largely from person to person so these criteria may not apply to all patients.

Research by Dalton, Ranum, and Day describes the clinical presentation of patients with MMD Type 2. The most prevalent findings are myotonia (90%) and proximal muscle weakness (82%).⁵ Patients with MMD Type 2 also commonly report muscle pain, stiffness, fatigue, and excessive daytime sleepiness. Other clinical features include defects in cardiac conduction (19%), cataracts (36-78%, increasing with age), and insulin insensitivity (25-75%, increasing with age). Diabetes mellitus may be more common in MMD Type 2 patients in comparison with MMD Type 1 patients. Cognitive manifestations of MMD Type 2 include difficulties with organization, concentration, and word finding. When performing a review of systems and history for a patient, these are important factors to address.

Current Literature

The majority of literature and research reviewed in preparation for this case focused on the patient's primary diagnosis of MMD Type 2. Pandya and Eichinger described the critical pieces of an initial evaluation for a patient with MMD.⁶ Systems review, manual muscle testing, dynamometry, functional ability assessment, as well as pain and fatigue assessments provide important data for monitoring a patient's disease progression. The article also addresses physical therapy management of patients with MMD. General range of motion is emphasized to maintain joint function and decrease occurrence of muscular imbalances. Strengthening exercises at a moderate intensity may help to avoid muscular weakness from disuse. High intensity exercise is not advised, as this can overstress the patient leading to an increase in symptoms and worsening of the disease progression. Cardiovascular exercise at a low to moderate intensity is safe for patients with MMD and

may benefit the patient. Since MMD can affect the heart, clearance from the patient's physician is recommended before beginning aerobic exercise.

A study by the United States Department of Health and Human Services suggests that individuals with chronic conditions or disabilities, such as MMD, would benefit from 2 hours and 30 minutes of moderate intensity exercise every week.⁷ This exercise program should include full body strengthening 2-3 times a week and aerobic conditioning lasting at least 10 minutes each session. According to Ted Abresch, the director of the Research and Training Center for Neuromuscular Disease at the University of California at Davis, adults with slowly progressing muscular disorders (such as MMD) show improvements from moderate intensity exercise.⁸ Walking studies have been particularly effective in decreasing the patients' weight and increasing their quality of life assessment scores.

When selecting modalities, it has been shown that patients with MMD typically have positive results with therapeutic heat.⁶ Cold is less therapeutic, as it increases the likelihood of myotonic muscle contractions. Adaptive equipment and assistive devices or orthotics may be helpful to improve the patient's independence and safety. Assessing the patient's need for such devices is an important part of physical therapy management for MMD.

When searching for further articles about MMD Type 2, it became apparent that research is severely lacking. Physical therapy intervention has been proven effective for other myopathies, but there is limited information available that is specific to MMD Type 2. The purpose of this article is to describe the initial physical therapy evaluation process for a patient with MMD Type 2, as well as the interventions used for this patient and the

patient's response to treatment. This article aims to add information to the limited supply that is available, and to increase awareness about the need for research concerning MMD Type 2.

CHAPTER II

CASE DESCRIPTION

History

The patient is a 63-year-old female. She was at a check-up for her sleep apnea when a nurse practitioner specializing in pulmonology referred her to physical therapy. The practitioner identified upper and lower extremity weakness and difficulty with standing during the check-up, which led to the referral. The patient presented to physical therapy the following week with bilateral upper and lower extremity weakness, decreased muscle tone of her hands, bilateral grip myotonia, and impaired balance. She was formally diagnosed with MMD Type 2 approximately five years ago by a neurologist who used diagnostic EMG studies.

The patient recalled symptoms of grip myotonia beginning when she was around 30-years-old. She reported a history of muscle weakness starting when she was approximately 40-years-old, which gradually worsened. The patient began using a cane at age 58. The patient reported a suspected family history of MMD, as she recalled relatives with similar presentations and health conditions. The patient was of European descent, and may be of heritage with increased prevalence, although this had not been confirmed. The patient's past medical history included (but is not limited to), diabetes mellitus, hypertension, hyperlipidemia, silent aspiration, pharyngeal dysphagia, decreased

peristalsis, diaphragm weakness, mixed restrictive and obstructive lung disease, obstructive sleep apnea, mild aortic insufficiency, bradycardia, cardiac pacemaker (insertion in 2002 and replacement in 2009), bilateral cataract removal (2008), cholecystectomy (1992), breast cancer with mastectomy (2008), hysterectomy (1992), degenerative joint disease, osteopenia, osteoporosis, ligamentous injury to the left knee, and MMD Type 2. The patient exhibited systemic involvement and had been seen by specialists for each of her diagnoses. She maintained regular check-ups and treatment by a multifaceted care team involving cardiologists, neurologists, general practitioners, pulmonologists, endocrinologists, and orthopedic specialists. Upon review of systems, the patient reported no changes or new complaints in these areas. The patient also had a history of falls, which resulted in traumatic coccydynia in 2010, a fractured left radius in 2012, and a distal left femur fracture in 2013. Following each fracture, she was seen in physical therapy for rehabilitation.

The patient's symptoms associated with MMD Type 2 were worsened by prolonged activity. Following a day of increased activity, the patient experienced high levels of fatigue. She reconciled the fatigue with sleep, which would help her feel better. By increasing the amount of sleep she received over the duration of a few days, she would return to her baseline level of function.

The patient lived in a rural area of the Midwest. She lived independently with her husband in their home. Her home had a ramp to the entrance, and approximately 5 stairs inside that she used on a regular basis. She had family in the area, and often cared for her teenage grandson when he was done with school. Due to her health conditions, the patient

had retired from waitressing. Her leisure activities were limited, and she did not make many community outings. She typically did not go into the community without her husband. She reported that driving to her physical therapy sessions was the first time she had traveled on her own in the last 5 years. The patient had limited resources in regards to her healthcare. There were no area programs for seniors to exercise, she was not a member of a support group or care organization related to her diagnosis, and she had to travel for her healthcare.

The patient's chief complaint was her decreased balance, as she reported multiple falls. Her main goals for physical therapy included improving her balance, increasing her muscle strength and endurance so that household chores were less challenging, improving her walking tolerance for community ambulation, and relief of her muscle pain.

Clinical Impression

The initial clinical impression following the subjective interview was that the patient would benefit from skilled physical therapy intervention. Observations of her sit-to-stand transfers and gait prior to formal examination, along with her reports of decreased balance and difficulties with household chores indicated that the patient might benefit from physical therapy. Skilled physical therapy would work to improve her balance and decrease her risk of falls. She would also benefit from an exercise program that had been adjusted to her personal needs, which would counteract her symptoms by increasing both her aerobic and muscular endurance. Manual therapy techniques, such as soft tissue massage, would help relieve her pain.

The goal of physical therapy was to increase her upper and lower extremity strength, allowing her increased functional capabilities at home. The purpose of continuing the strengthening program would be to maintain her functional strength for as long as possible. The goal of balance training was to decrease the patient's likelihood of falling and to increase her mobility. All of the proposed goals coincide with the reported goals of the patient.

Examination Plan

The plan for examination was to document her current functional status. Observations of her bed mobility, sit-to-stand transfers, and gait would assist in assessing her transferring abilities and mobility level. Pinch strength, grip strength, and full body manual muscle testing would gauge her muscular strength and identify areas of weakness. Balance tests, such as the Berg Balance Scale and the Timed Up and Go Assessment, would be used to determine her risk of falls and areas of difficulty. Range of motion measurements would assess if the patient had full and pain-free motion of all body parts.

CHAPTER III

EXAMINATION AND EVALUATION

Observation

When walking into the clinic, the patient used a cane in her right hand. Proper gait sequence was used, although she leaned heavily on her cane. The patient navigated non-complicated environments well, but with increasing complexity she demonstrated decreased tolerance. For example, in crowded areas or on uneven surfaces she self-selected a slower gait speed and reported feelings of unsteadiness and weakness. For these reasons, the patient used her cane for community ambulation and for walking distances longer than approximately 50 feet. On stairs, the patient used a step-to-step pattern with bilateral upper extremity assistance on hand railings. She flexed forward at her hips and relied heavily on her upper extremities to pull herself up onto the next stair. The patient demonstrated use of proper lead foot, and she reported her right side being dominant and stronger.

The patient had difficulty with transitional movements, but with compensatory patterns and adaptations she remained independent. When transferring from sit-to-stand, she demonstrated a Gower's sign. She leaned very far forward and walked her hands up her legs to assume an erect position, and generally required multiple attempts to stand. At home, she used a front-wheeled walker to assist with sit-to-stand transfers. The patient

reported that she did not usually use any form of assistive device within the home for ambulation.

When observing her bed mobility, similar trends appeared. The patient pulled with her arms to assist with log rolling to the side, which required multiple small weight shifts to occur and was physically demanding. She had to support her head with a pillow during this transition, as her neck muscles were too weak to assist with this movement. When transferring to and from supine, she was able to swing her legs and controlled her upper body descent with her arms. She kept her core and neck stable, but was unable to dynamically contribute to the transfer with her core stabilizers.

She presented with a right lateral shift during gait, standing, and sitting. Postural observation revealed increased thoracic kyphosis and cervical lordosis, as exhibited by a forward head and rounded shoulders.

Tests and Measures

The patient presented with full range of motion of the legs and arms bilaterally. Strength was assessed using manual muscle testing and dynamometry for grip and pinch strength measurements. Special tests were selected to test the patient's balance and functional abilities. The psychometric properties of all tests and measures used in this episode of patient care can be found in Table 3.

Initial evaluation data indicated proximal muscle weakness. Manual muscle testing was performed according to Magee.⁹ For full initial evaluation data, see Table 1. Significant weakness was found in both the upper and lower extremities, as well as core stabilizers. Shoulder flexion and extension, gross wrist motions, gross finger motions, gross hip

motions, and gross neck motions scored within the range of grade 4-/5 to 4+/5. The patient's area of greatest weakness was trunk flexion and extension, which were both found to be grade 2/5 at initial evaluation.

Table 1. Initial and Discharge Manual Muscle Testing and Dynamometry Results

Initial Manual Muscle Testing for Strength				Discharge Manual Muscle Testing for Strength			
Upper Body	Action	Right	Left	Upper Body	Action	Right	Left
Shoulder	Flexion	4+/5	4/5	Shoulder	Flexion	4+/5	4/5
	Extension	4+/5	4+/5		Extension	5/5	5/5
	Abduction	5/5	5/5		Abduction	4-/5	4-/5
	Adduction	5/5	4/5		Adduction	4+/5	4+/5
	Internal Rotation	5/5	5/5		Internal Rotation	4/5	4-/5
	External Rotation	5/5	5/5		External Rotation	4+/5	4/5
Elbow	Flexion	5/5	5/5	Elbow	Flexion	5/5	4+/5
	Extension	5/5	5/5		Extension	4+/5	4/5
Wrist	Flexion	4+/5	4/5	Wrist	Flexion	4+/5	4/5
	Extension	4+/5	4/5		Extension	4/5	4-/5
	Ulnar Deviation	4+/5	5/5		Ulnar Deviation	4/5	4/5
	Radial Deviation	5/5	5/5		Radial Deviation	4+/5	4/5
	Supination	4/5	4-/5		Supination	5/5	4/5
Fingers	Pronation	4/5	4/5	Fingers	Pronation	5/5	4/5
	Extension	4-/5	4-/5		Extension	4/5	4-/5
	Flexion	(see grip and pinch strength dynamometry)			Flexion	(see grip and pinch strength dynamometry)	
Initial Dynamometry				Discharge Dynamometry			
		Right	Left			Right	Left
	Grip Strength	8 lbs	4 lbs		Grip Strength	8 lbs	5 lbs
	Pinch Strength	4 lbs	3 lbs		Pinch Strength	4 lbs	3 lbs
	3 Jaw Chuck	4 lbs	4 lbs		3 Jaw Chuck	6 lbs	3 lbs
	Lateral Pinch	7 lbs	6 lbs		Lateral Pinch	7 lbs	8 lbs

Lower Body	Action	Right	Left
Hips	Flexion	4/5	4-/5
	Extension	4+/5	4/5
	Abduction	4/5	4/5
	Adduction	4/5	4/5
	Internal Rotation	4/5	4/5
	External Rotation	4+/5	4-/5
Knees	Flexion	4/5	4-/5
	Extension	4+/5	4/5
Ankles	Dorsiflexion	5/5	5/5
	Plantarflexion		
	Inversion	4+/5	4/5
	Eversion	5/5	5/5
Toes	Flexion		
	Extension	5/5	5/5
Neck	Flexion		4-/5
	Extension		4/5
	Side-bending		4/5
	Rotation		
Abdominals	Crunch		2/5
Back	Extension		2/5

Lower Body	Action	Right	Left
Hips	Flexion	4/5	4/5
	Extension	4/5	4/5
	Abduction	4-/5	4/5
	Adduction	4/5	4/5
	Internal Rotation	4-/5	4-/5
	External Rotation	4/5	4/5
Knees	Flexion	5/5	5/5
	Extension	4+/5	4/5
Ankles	Dorsiflexion	4+/5	4+/5
	Plantarflexion		
	Inversion	5/5	5/5
	Eversion	5/5	5/5
Toes	Flexion	4/5	4/5
	Extension	4/5	4/5
Neck	Flexion		4-/5
	Extension		4+/5
	Side-bending		4/5
	Rotation		4/5
Abdominals	Crunch		2/5
Back	Extension		2/5

Grip dynamometry also revealed significant weakness compared to age norms. Standardized test positioning was used with a Jamar® dynamometer to assess grip strength. According to the Rehabilitation Measures Database, normative values for women aged 65-69 is 54.9 pounds of force with a 10.1 pound standard deviation.¹⁰ The patient in this case study generated 8 pounds of force with the right hand and 4 pounds with the left hand; both grip strength values were significantly lower than the normative values.

Pinch strength was measured using a Sammons Preston Baseline® Pinch Gauge. Normative data generated by faculty at the University of Wisconsin-Milwaukee for females 60-65 years old has been compiled.¹¹ Please, see Table 2 for these normative values. The patient's initial tip pinch strength was 4 pounds with the right and 3 pounds with the left, key pinch strength was 7 pounds on the right and 6 pounds on the left, and palmar grip strength 4 pounds bilaterally. All of the dynamometry measurements were significantly below normal for her age and gender.

Table 2. Pinch Dynamometry Normative Data for Females Age 60-65

	Right Hand (Pounds)	Right Hand Standard Deviation (Pounds)	Left Hand (Pounds)	Left Hand Standard Deviation (Pounds)
Tip Pinch	10.1	2.1	9.9	2
Key Pinch	15.5	2.7	14.1	2.5
Palmar Pinch	14.8	3.1	14.3	2.7

Special tests were selected to assess the patient's balance, mobility, and fall risk. The Berg Balance Scale (BBS) was administered to assess the patient's balance with various tasks, allowing examiners to compute fall risk. The patient was instructed to perform both static and dynamic balance activities of varying difficulty. A maximum score of 56 is ideal. For patients with a history of falls and a score less than 51, or those with no history of falls and a score less than 42, the BBS is predictive of falls with 91% sensitivity and 82% specificity. A score of less than 40 on the BBS is associated with an almost 100% fall risk. Normative scores for women ages 60-69 was 55 with a standard deviation of 2. The patient in this case study scored 37, which means she had almost a 100% risk of falling at the time of her initial evaluation.

The Timed Up and Go (TUG) Assessment was also used. The TUG assessed the patient's mobility and balance. It was useful in determining her risk of falling. A score of 20 seconds or less for a community-dwelling elderly individual indicates their functional ability is sufficient to safely transfer independently. If they score over 30 seconds, they are likely to be dependent for safe transfers. Normative data for community-dwelling elderly females aged 60-69 was 8 seconds with a standard deviation of 2 seconds. The patient in this case study was able to complete the TUG in 13 seconds with the use of her cane, which is significantly longer than the norms for her age and indicates an increased risk of falls. This is consistent with other examination findings. However, her balance and strength are not impaired enough to make her dependent for transfers.

Table 3. Psychometric Properties of Tests and Measures

	Sensitivity	Specificity	Test-Retest Reliability	Intrarater Reliability	Interrater Reliability	Minimal Detectable Change	Accuracy
Manual Muscle Testing	35% ¹⁰	90% ¹⁰	98% ¹⁰				
Grip Dynamometry			98% ¹⁰	98% ¹⁰			3% ¹⁰
Pinch Dynamometry			80% ¹²		97% ¹²		1% ¹²
Berg Balance Scale	91% ¹⁰	82% ¹⁰	91% ¹⁰	97% ¹⁰		3.3-6.3 ¹⁰	
Timed Up and Go	56% ¹⁰	60% ¹⁰	97% ¹⁰	99% ¹⁰			

CHAPTER IV

DIAGNOSIS AND PROGNOSIS

The examination process confirmed initial clinical impressions and revealed physical therapy diagnoses of bilateral upper and lower extremity weakness, decreased muscle tone of her hands, bilateral grip myotonia, and impaired balance. These findings are consistent with her medical diagnosis of MMD Type 2. It was determined that the patient was in need of skilled physical therapy to improve her function. The plan of care included balance and proprioceptive activities, a home exercise program, and manual therapy as needed for treatment of pain. This plan of care was to be carried out twice weekly over the duration of six weeks. This amount of time would allow her to learn her home exercise program with instruction from a physical therapist to avoid compensation patterns, develop her balance strategies and provide her with proprioceptive challenges under clinical supervision to ensure safety, and explore pain management techniques to improve her quality of life. Due to the progressive nature MMD Type 2, her prognosis and rehabilitation potential are guarded. It was hypothesized that she would see some minor improvements in her functional abilities and scores on tests and measures. Individuals with muscular dystrophies are able to benefit from physical therapy services, and one goal was to offer this patient a plan of care that would help her maintain or improve her current level of function.

The patient's goals were to improve her ability to complete household activities, increase her tolerance for walking, and to complete her self-cares with more ease. She also wished to decrease her discomfort from muscular pain. The patient highly valued her independence and interactions with her family, and wanted to maintain her function in these areas. By improving her strength and balance, completing home activities, walking, and self-care should become easier. The physical therapy goals and patient goals aligned. Both were to maintain her function and to increase her strength and endurance to assist with completion of her daily activities.

A plan for periodic reexamination was set in place. Continual reassessment of her performance at each physical therapy session was used to track her progress. More formal examinations were to be performed after three weeks and after six weeks of physical therapy intervention. At these times, a TUG assessment and strength screening would be performed to track her progress. A long term physical therapy program would include tracking her progress in monthly intervals, offering her patient education, monitoring the effectiveness of current treatment, and adjusting her plan of care as needed.

CHAPTER V

INTERVENTIONS

This patient was seen two days a week for one hour sessions over the course of three weeks. Due to the patient's systemic involvement, exercise was maintained at moderate intensity so that aerobic and strengthening effects would occur without exacerbating the patient's fatigue or cardiovascular conditions. The patient was able to continue a conversation during all of her exercises, which is consistent with moderate intensity criteria. If the patient became breathless in conversation during activity, expressed fatigue, or demonstrated integumentary color changes (e.g. cyanosis of the lips) then exercise was paused and the patient would be instructed to rest for a few minutes until the signs and symptoms dissipated. Our program followed the general guidelines described by Pandya and Eichinger⁶, as well as recommendations by the United States Department of Health and Human Services⁷ and Ted Abresch⁸.

Week One

The first week's interventions included performing upper and lower extremity strengthening exercises while teaching these to the patient for her to use as a home exercise program. The patient demonstrated independence in these exercises after the first week. With cuing and patient education, she was able to self-monitor and avoid compensation patterns when completing the exercises. The goal of these exercises was to

increase her upper and lower extremity strength, resulting in increased functional capabilities at home. The purpose of continuing the strengthening program was to counteract the progressive nature of her disease by maintaining her functional strength for as long as possible. The patient reported compliance with her home exercise program during the course of her physical therapy treatment.

Upper Body Strengthening

The patient was instructed in bilateral upper extremity exercises against the resistance of a Theraband®. The Theraband® selected was lime green in color and offered medium resistance. The patient performed 12 repetitions of each exercise, 3 times per week. The exercises included seated shoulder flexion, abduction, scaption, and bicep curls. Due to her grip weakness, one end of the band was tied to the chair and the opposite end was fashioned into a loop for her hand. Standing shoulder extension, internal rotation, and external rotation were also performed. One end of the band was secured to a door and the other was made into a loop for her hand.

Lower Body Strengthening

The bilateral lower extremity strengthening exercises were completed against gravity with body weight resistance. The patient performed 15 repetitions of each exercise, 3 times per week. The exercises included seated hip flexion, seated long arc quads, and sit-to-stand transfers. While standing the patient performed heel and toe raises, marching, mini squats to 45 degrees, hip abduction, hip extension, hamstring curls, and crossovers. Crossovers were performed by bringing the leg into adduction and tapping the toe in front of the contralateral foot, then repeating to the opposite side. The lower extremity exercises

were completed in parallel bars at the outpatient clinic, and with bilateral upper extremity support on a countertop at home.

Weeks Two and Three

During the second and third weeks of therapy, more challenging strengthening and balance exercises were performed under supervision. The goal of balance training was to decrease the patient's likelihood of falling and to increase her mobility. The exercises incorporated functional movements and elicited righting reactions. The purpose was to train her body to respond to environments that challenged her balance by using proprioceptive feedback and righting reactions to prevent falls. The patient was initially apprehensive to perform these activities. The patient was educated on the benefits of balance training and the safety measures used to protect her while she completed them, which made her feel more comfortable and willing to perform the exercises. The patient was compliant once she understood the purpose. The patient was adequately challenged, as she was able to maintain balance for longer than 10 seconds but not over 30 seconds at a time. The patient used the parallel bars often by tapping her hands on them to maintain balance, however she did not have to grip them to maintain balance. She was unable to multitask during the rocker board activities, and had to stop talking and focus specifically on her balance. For the other exercises, she was able to multitask and maintain a conversation while completing the task.

Balance Exercises

Her lower extremity home exercise program was completed with foam balance pads under each foot. She also performed rocker board activities by shifting her weight in anterior-posterior and side-to-side directions for 15 repetitions each. The patient also performed 15 repetitions of weight shifting in diagonals with alternating lead leg. All of these balance activities were performed in parallel bars with stand by assist. A gait belt was donned for these activities.

Manual Therapy

Manual therapy techniques, including soft tissue massage and myofascial release, were used throughout the 3-week duration of therapy to treat muscular pain as symptoms arose. The patient did not tolerate deep pressure. Each time soft tissue massage was used, light pressure was selected and as the patient accommodated pressure was gradually increased to a moderate amount. The patient expressed relief following the use of manual therapy and was happy with the results.

Aerobic Exercise

Aerobic exercise was incorporated into every physical therapy session. The patient used the NuStep® and was progressed based on her presentation. At the initial evaluation, the patient was able to complete 12.5 minutes on the NuStep® at level 2 resistance. By discharge, the patient increased her duration to 40 minutes and resistance to level 5. The length of time was increased first, then resistance was increased as the patient was able to further progress. Conversation was maintained during the aerobic exercise to ensure moderate intensity.

CHAPTER VI

OUTCOMES

The patient responded positively to treatment. She did not have symptom exacerbation after her physical therapy sessions, and maintained compliance with her home exercise program. The patient reported satisfaction with her therapy. She felt that it was helping increase her functional abilities and understood the importance of continued activity in relation to her diagnosis. She felt challenged by her exercises, but did not experience set-backs in her condition after completing them.

At discharge, the patient demonstrated improvements in her strength, balance, and aerobic capacity. The patient improved in her grip and pinch strength dynamometry readings by 1-2 pounds of force bilaterally. Variable results were compiled from the manual muscle testing strength assessment. The patient improved in both the TUG and BBS. The patient completed the TUG in 12 seconds with her cane, and 11 seconds without use of an assistive device. She scored 47/56 on the BBS. This 10-point improvement exceeds the minimal detectable change criteria for the BBS (Table 3).¹⁰ This score indicates that she is still at risk for falls, but she is no longer at a 100% risk. The patient demonstrated increased aerobic capacity as well. She was able to increase her resistance on the NuStep® from level 2 resistance to level 5, and increase the duration from 12.5 minutes to 40 minutes while maintaining a conversation.

The patient was discharged after three weeks for personal reasons. The student physical therapist she had been working with was about to finish their clinical rotation, and her care would have been transitioned over to one of the physical therapists at the facility. Although she was assured the transition would be smooth and she would receive high quality care, the patient decided she did not want to continue. The patient felt she was ready to be discharged to complete her home exercise program on her own. She was independent in her home exercise program and displayed the ability to self-manage her care at this point. She would have benefitted from continued physical therapy intervention for balance. To reduce her fall risk, it was recommended that the patient attend a balance class that had just started in her community. The patient complied and signed up to start this class immediately following her discharge from physical therapy. The patient is highly motivated to prevent falls, and will likely comply with the plan that was developed.

The progressive nature of MMD Type 2 was discussed with the patient, and she was aware of the implications of this. Since the patient has systemic involvement, she has a multifaceted care team involved in her treatment. She maintains regular medical checkups and had plans to continue with this after discharge.

CHAPTER VII

DISCUSSION

The patient made improvements in strength and balance as indicated by dynamometry, TUG, and BBS measurements. Balance training and completion of a strengthening home exercise program were integral to this success. Variable results were compiled from the manual muscle testing strength assessment. The day before discharge, the patient reported completing strenuous activity. This included spending time at a firing range practicing with her pistol; during the discharge evaluation she reported lasting fatigue from the day before.

Due to her daily changes in presentation, manual muscle testing results would be more conclusive if completed at monthly intervals. Continued long-term assessment would allow practitioners to track the progression of the disorder. Taking into account the progressive nature of her disease, the patient was not expected to have large increases in strength. The results obtained are promising, as she displayed positive changes in a short 3-week period of physical therapy intervention. This is likely due to greater efficacy of muscle recruitment following regular exercise. If the patient continues her program, she may continue to see small improvements. By maintaining her health through activity and regular medical checkups, she may have a more positive future. Due to the progressive nature of MMD Type 2, she is expected to experience decreases in her strength and functional capabilities over time. Hopefully, the positive effects of exercise will help to slow

this process. The patient also displayed increases in her aerobic capacity, as shown by the increased resistance and duration completed on the NuStep®. This will positively impact her cardiovascular function and her ability to complete functional activities. This patient responded similarly to patients described in literature.⁸

Future studies should focus on the short and long term impact of regular exercise on patients with MMD Type 2. It would also be beneficial to study the effects of balance training on fall risk for patients with MMD Type 2. The patient in this case study experienced positive results from physical therapy intervention, which implicates the need for further research. If positive results are consistently found for this patient population, physical therapy intervention may become a more regular part of routine care for individuals with MMD Type 2.

CHAPTER VIII

REFLECTION

In hindsight, this patient would have benefitted from a referral to a clinic with specialists who have experience with MMD. Her care up to this point had been somewhat disjointed. She saw a wide variety of specialists, but did not have a single provider with experience treating MMD to tie the systemic manifestation together. When reviewing her medical records, she had not received genetic testing or family counseling. The patient would greatly benefit from this, as it would confirm her diagnosis and track the disease through her family. If she was willing, she could also volunteer to be involved in research.

In future appointments, a cognitive assessment should be implemented to screen for involvement. The Mini-Cog Test® would be an option to use for her. She did not present to physical therapy with any signs or symptoms of cognitive delays, and she did not express concern for this. However, changes in cognition commonly accompany the progression of MMD. So, collecting baseline data and monitoring her cognition over time would be beneficial.

The patient would have also benefitted from more formal patient education about energy conservation and home safety. These were topics that were discussed in physical therapy, however a home evaluation and ergonomic assessment were not performed.

Her family was supportive, and it would have been beneficial to include them in on the patient care and education. Her husband did not attend any of the sessions, as he was at work. Other family members had similar conflicts with work and school schedules. For these reasons, family involvement in developing a plan of care was lacking and would be an area of improvement for future appointments.

High quality evidence was applied to patient care through literature review and use of well-known tests and measures. As stated in the decision making process for accepting the patient for care, the interventions were indicated and helped the patient. Reflective thinking and ongoing assessment led to small adaptations of her HEP, such as tying loops in the Theraband® due to her grip weakness, so that her care was best suited to her individual needs.

CHAPTER IV

CONCLUSION

The clinical implications of this case study support the completion of a routine fitness program for individuals with MMD. Regular strengthening and aerobic exercises, along with balance training, may benefit other individuals with MMD as well.

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