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A Case Report: Treatment of Facioscapulohumeral Muscular Dystrophy

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A CASE REPORT: TREATMENT OF FACIOSCAPULOHUMERAL MUSCULAR DISTROPHY

by

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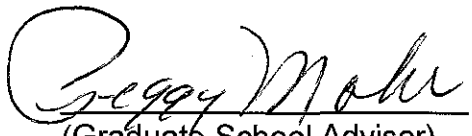

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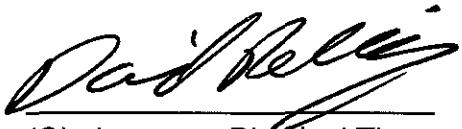

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Date 6/11/2018

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I would like to thank my clinical instructor for all of her support throughout this clinical rotation. I would also like to thank my advisor Peggy Mohr, PT, PhD for assisting me with the organization of this project.

ABSTRACT

Purpose: The purpose of the case study was to explain a treatment program involving low/moderate intensity strength training and stretching for a patient with facioscapulohumeral muscular dystrophy (FSHD). **Background:** FSHD is the third most common type of muscular dystrophy. This pathology results from a mutation in chromosome 4 and results in progressive weakening of facial, scapular, and hip musculature. Research behind effective strengthening programs is often inconclusive and varies in results. **Case Description:** The patient was a 34-year-old female, who was diagnosed with FSHD 3 years prior to our first visit. She had complaints of left shoulder pain (2-3/10), shoulder and hip weakness, and limited shoulder range of motion (ROM). The patient reported her lower extremities fatigued and had multiple falls over the last 2 years. **Plan of Care:** Interventions included upper and lower extremity strengthening exercises and shoulder ROM exercises. Prior to discharge, exercise program to continue strength training independently was provided to the patient. The patient received a significant amount of education throughout her plan of care. **Outcomes:** At discharge, the patient had no significant changes in strength or ROM. She reported no left shoulder pain and significant increases in functionality as recorded on an Upper Extremity Functional Index (UEFI). **Conclusion:** After 6 weeks of low/moderate intensity strength training and stretching, strength and ROM did not improve, but functionality based on the UEFI did improve.

CHAPTER I

BACKGROUND AND PURPOSE

Muscular dystrophy is a genetic condition marked by progressive muscular weakening. There are many forms of muscular dystrophy, with FSHD being the third most common.[1] Muscular dystrophy is caused by genetic mutations resulting in the dysfunction of proteins that protect muscle fibers during muscular contractions.[2-3] Although each type of muscular dystrophy results from a different mutation, all result in muscular weakening. Due to the similar cause of each form of muscular dystrophy, the different variations are often studied together when researching different methods of treatment.

While the most common form of muscular dystrophy, Duchenne muscular dystrophy (DMD), results from a mutation on the X-chromosome, FSHD results from a mutation on an intron on the 4th chromosoms.[4] Symptoms of FSHD include facial weakness, shoulder/scapular weakness, and often leg and pelvis weakness.[5] Physical therapy interventions have been researched for decades, but there continues to be limited evidence on the most effective treatment options. Due to the nature of this disease, a major concern of treatment is causing further muscle damage. Since the muscle cells within patients with muscular dystrophy are already susceptible to tearing, eccentric muscle training is commonly accepted as a poor intervention option.[6]

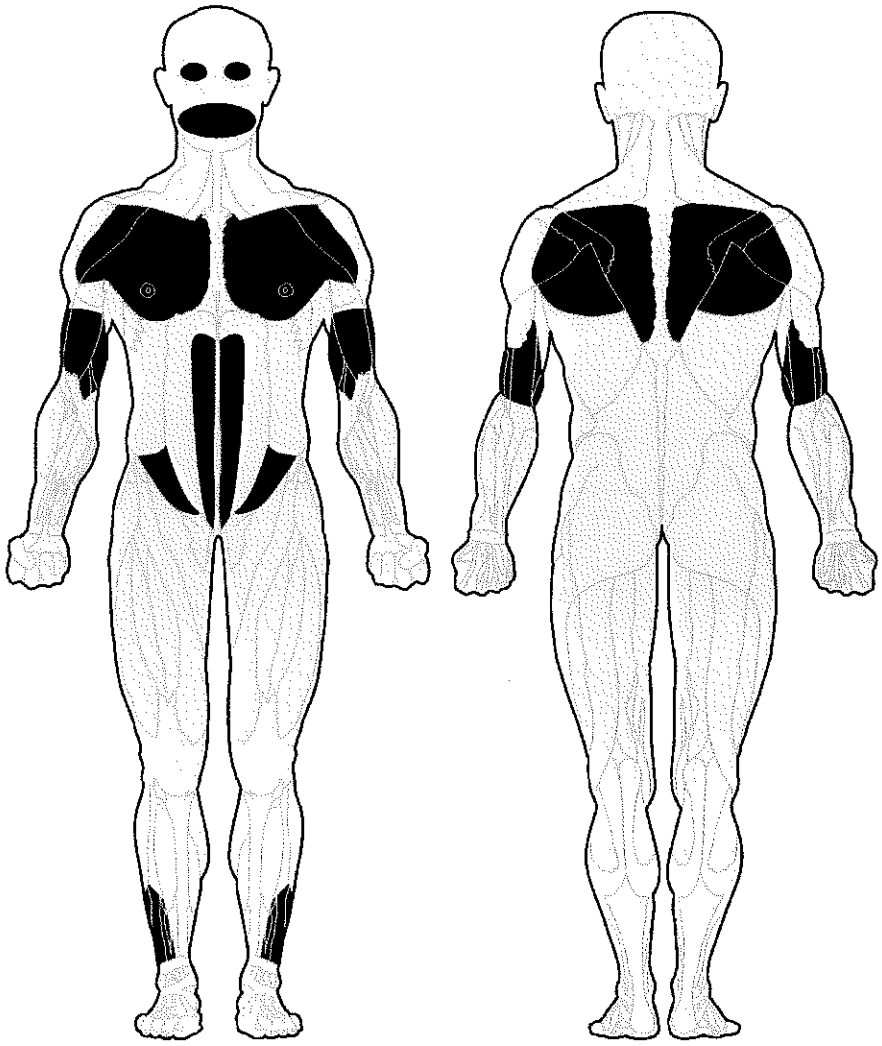

Figure 1. Distribution of Commonly Affected Muscles in FSHD Patients.

The effects of strength training on individuals with muscular dystrophy have been studied; however, the results are variable. During a 6-month trial of 4 patients with DMD, de Lateur et al[7] found that unilateral submaximal quadriceps strengthening resulted in a greater max torque compared to the non-trained quadriceps. Lindeman et al looked at patients with myotonic dystrophy (MyD) and found that 24 weeks of knee extension/flexion exercises resulted in no significant improvement in knee torque.[8] Van der Kooi et al[9] found that 52 weeks

of strengthening elbow flexors and dorsiflexors along with Albuterol was effective at strengthening elbow flexors but not dorsiflexors. Although many studies[8,10] reported no significant improvement in strength with treatment, the progressive nature of the disease must be considered.

Although the effectiveness of strength training continues to be researched, most studies[11-13] that look at the effects of endurance training report positive effects. Olsen et al[11] found that 12 weeks of low-intensity aerobic training was effective at improving VO_2 max with no signs of muscle damage for patients with FSHD. Orngreen et al[12] also found 12 weeks of aerobic training to be effective at improving the cardiovascular fitness of individuals with MyD. A similar study by Sveen et al[13] found that 12 weeks of aerobic training on a cycle ergometer improved VO_2 max and significantly improved hip abduction strength in individuals with Becker muscular dystrophy (BMD). In summary, there is support for the use of submaximal aerobic training for patients with muscular dystrophy.

CHAPTER II

CASE DESCRIPTION

The patient was a 34-year-old female with a history of facioscapulohumeral muscular dystrophy (FSHD). She lived alone in an apartment with 6 steps down to enter. She was diagnosed about 1.5 years prior to her initial evaluation, but reported seeing a decline in strength over the last decade. She reported pain with swinging her arms while walking and lifting her arms above her head while showering. She reported being constantly fatigued and reported having multiple falls from tripping over the previous couple years. Past medical history included a follicle stimulating hormone (FSH) imbalance that was controlled with a prescription medication. She also reported 5 previous surgeries on her left elbow which resulted in decreased range of motion. She reported getting a poor night sleep on a regular basis, although pain was not the cause. Her most difficult activities of daily living (ADL) were anything that required her to have her arms above her head including washing and combing her hair. She also had difficulty donning and doffing clothes on occasion. Her occupation required a lot of walking which she stated was very fatiguing to her lower extremities as well as her body as a whole.

The patient originally reported to physical therapy with an order to treat her left shoulder pain. She stated her symptoms appeared 3-4 months prior to

her initial visit. Her shoulder pain had a gradual onset with no trauma to the shoulder that she could recall. She stated her pain during therapy was 2-3/10 using the visual analog scale (VAS) with pain fluctuating from 0-5/10 throughout her day. The patient reported her symptoms occasionally went down her left arm, but the symptoms were not "shooting" in nature. She reported pain was generally worse in the morning and diminished slightly with the use of a heating pad. The patient also reported a painful pulsing sensation in the back of her neck on occasion with flexion and extension movements. The patient's chart had a recent non-significant X-ray of the left shoulder. Her chart also reported that she would be started on Diclofenac, a nonsteroidal anti-inflammatory drug, and Nortriptyline, an antidepressant. The patient was very aware of the progressive nature of her condition and stated that her goal with therapy was to prevent further decline of her strength.

On the patient's third visit, 1 week after starting therapy, the therapy department received orders from her physician to treat her muscular dystrophy as a whole. Due to her initial evaluation 1 week prior to her third visit, we were able to bypass much of her history during therapy.

Examination, Evaluation and Diagnosis

Examination

During the patient's initial examination, cervical ROM and upper extremity ROM were assessed. Flexion and rotation were within functional limits (WFL) bilaterally with lateral flexion limited to 25 degrees to the right and 29 degrees to the left. Shoulder flexion was 78 degrees on the right and 73 degrees on the left

while shoulder abduction was 80 degrees on the right and 65 degrees on the left (see Table 1).

Upper extremity strength was assessed using manual muscle testing (MMT) which is a 0-5 scale with 5/5 indicating normal strength (+'s and –'s indicate strength slightly above or below the preceding number). The patient had 5/5 strength with shoulder adduction bilaterally and 5-/5 strength with shoulder flexion (see Table 2). After receiving orders to treat the patient's muscular dystrophy, her lower extremity was assessed via MMT. The patient had 2/5 strength for hip flexion and abduction bilaterally and 2-/5 strength for hip adduction. Knee extension strength was normal (5/5) and knee flexion strength was slightly below normal (5-/5).

While sitting, she had a slumped posture with rounded shoulders. The patient was negative bilaterally for an Anterior Apprehension Test, Drop Arm Test, and Lift Off Sign. She had a positive O'Brien's Test on the left shoulder but not the right shoulder indicating a possible superior labrum tear on the left shoulder. She was not tender to palpation anywhere on her shoulders, and reported that pain resulted from muscle activation. The patient did have tight suboccipital muscles and upper trapezius muscles bilaterally.

She was independent with ambulation and her standing/sitting balance appeared functional. The patient stated that her balance often declined throughout the day due to her muscles fatiguing. She had a slow gait pattern with excessive hip internal rotation bilaterally. The patient also displayed bilaterally

winging and anterior tilt of her scapula. During all shoulder motions, her upper trapeziuses were dominant and unable to relax.

The patient's initial functionality was assessed using the Upper Extremity Functional Index (UEFI). The patient had a score of 43/80 on her initial UEFI (80/80 indicates maximal functionality). During the examination of her muscular dystrophy, she was given a Fatigue Severity Scale on which she scored a 54/63 (0/63 indicates no fatigue).

Evaluation

Patient's shoulders were very limited into flexion and abduction ROM bilaterally; however, upper extremity strength appeared intact. Lateral cervical flexion was limited, but did not appear to be a major concern of the patient. Lower extremity hip and knee strength were limited with hip flexion, adduction, and abduction being affected the greatest. The patient's fatigue was reported to be very inhibiting for the patient and it was anticipated that this was likely to decrease her participation in physical therapy. Although she had a positive O'Brien's Test, no other signs indicated a torn labrum. According to the UEFI, her upper extremity function was 46.3% impaired. She also had a significant amount of fatigue according to the Fatigue Severity Scale. Although the patient had normal upper extremity strength via MMT, she appeared to have very weak scapular stabilizers as she had severe winging during her upper extremity motions. Due to the gradual onset of her shoulder pain without any trauma, her lack of scapular stability was one possible issue that may have led to her shoulder pain.

Table 1. ROM during Initial evaluation and Final Assessment

	Initial Assessment (10/30/2017)	Final Assessment (12/8/2017)
Active R shoulder flexion	78 degrees	70 degrees
Active L shoulder flexion	80 degrees	80 degrees
Active R shoulder abduction	73 degrees	73 degrees
Active L shoulder abduction	65 degrees	65 degrees
Active R Elbow ROM	9-135 degrees	8-136 degrees
Active L Elbow ROM	20-127 degrees	21-128 degree
Passive R shoulder flexion	144 degrees	145 degrees
Passive L shoulder flexion	152 degrees	152 degrees

Table 2. Strength via MMT during Initial Evaluation and Final Assessment

	Initial Assessment (10/30/2017)	Final Assessment (12/8/2017)
R shoulder abduction	5/5	5-/5
L shoulder abduction	5/5	5-/5
R shoulder flexion	5-/5	4+/5
L shoulder flexion	5-/5	4+/5
R hip flexion	2/5	2/5
L hip flexion	2/5	2/5
R hip abduction	2/5	2/5
L hip abduction	2/5	2/5
R hip adduction	2-/5	2-/5
L hip adduction	2-/5	2-/5
R knee extension	5/5	5/5
L knee extension	5/5	5/5
R knee flexion	5-/5	4+/5
L knee flexion	5-/5	4+/5

Diagnosis

The patient had limited shoulder flexion and abduction, weakness of her hip flexors, adductors, and abductors and pain in her left shoulder. She had difficulty with upper extremity activities and generalized fatigued throughout her body.

Prognosis, Goals and Plan of Care

Prognosis

The patient had a fair prognosis. She was young and motivated; however, FSHD has no known cure with most treatments only designed to slow the progression of the condition. According to the facioscapulohumeral muscular dystrophy society,[14] approximated 25% of patients with this disorder require the use of a wheelchair by the age of 50. Fortunately, life expectancy is near normal for patients with FSHD, so maintaining quality of life before and after a patient is ambulatory is the main focus of treatment. During treatment, it was important to balance optimism and the progressive nature of this disorder.

Goals

Goals for physical therapy were focused on maintaining or improving the patient's strength and ROM and decreasing shoulder pain to allow her greater functionality. Her long term goals are below.

Following physical therapy intervention, patient will

1. Report 0/10 pain in left shoulder to allow her to complete her ADL's with less discomfort (to be met in 12 weeks).

2. Have greater than 100 degrees of shoulder flexion bilaterally to allow her to complete overhead activities with greater ease (to be met in 12 weeks).
3. Have 90 degrees of left shoulder abduction to allow her to reach above her head with greater ease (to be met in 12 weeks).
4. Have 3/5 hip abduction strength bilaterally to allow her the lower extremity strength to function efficiently at work (to be met in 12 weeks).

Her short term goals are below.

Following physical therapy intervention, patient will

1. Report a 25% decrease in their left shoulder pain to allow them to complete their ADL's with less discomfort (to be met in 3 weeks).
2. Demonstrate understanding of their home exercise plan (HEP) to allow them to improve their strength and ROM independently (to be met in 3 weeks).

Plan of Care

Patient was scheduled for therapy 2-3 times per week for 45 minute therapy sessions. The patient's evaluation indicated that lower extremity strength would be a major focus of therapy. Specifically, we planned to work on the strength of her hip abductors, adductors, and flexors to allow her to ambulate with less fatigue. We also planned to work on improving shoulder range of motion to allow her to complete overhead activities. Strengthening scapular stabilizers was also prioritized to gain proper shoulder mechanics which would prevent future shoulder pathologies. Endurance would also be incorporated into therapy as tolerated. Due to the fatiguing effects of her job, endurance training would not

be a major component of therapy; however, she would be encouraged to walk or use exercise equipment independently and, as tolerated, to maintain or improve her aerobic endurance. The patient was also sent home with a HEP following each therapy intervention to allow her to progress independently. Educating the patient on the treatment options as well as the progression of the disease was also a component of therapy since the condition was relatively new to her.

CHAPTER III

INTERVENTION

The patient was seen 2-3 times per week for 6 weeks. Her intervention consisted of strengthening exercises, ROM exercises, endurance training, and education. The patient received instructions on a HEP on her first visit and her HEP was modified as necessary throughout treatment. The patient was also instructed on proper use of exercise equipment at a fitness center located next to the therapy department.

On the first visit, an examination and evaluation were performed on the patient. Following her evaluation, patient was instructed on how to do wall slides to encourage bilateral shoulder flexion and abduction. She performed lateral cervical flexion bilaterally with and without cervical rotation, holding for 20 seconds to stretch lateral neck musculature. She performed scapular protraction from supine against resistance of theraband and scapular adduction while supine to strengthen scapular stabilizers. The patient was given handouts of these exercises before leaving her appointment.

On the second visit, she reported some soreness in her lower neck. Manual therapy was performed to her neck to stretch her suboccipital muscles, upper trapezius, and levator muscles bilaterally. Passive range of motion

(PROM) was performed on left shoulder in all planes. Supine isometric holds were performed for shoulder external/internal rotation, abduction/adduction, and flexion/extension. The patient also performed scapular retraction/protraction exercises against resistance.

On the third visit, a second examination was performed on the patient with a focus on the lower extremities. Following her evaluation, the patient reviewed her HEP with the therapist and demonstrated understanding. Due to a scheduling error, there was no time to work on exercises after reviewing her HEP.

On the fourth visit, the patient reported feeling more fatigued that usual. During therapy, she performed short arc quads (SAQs), hamstring sets, straight leg raises (SLRs), hip abduction/adduction from supine using a sliding board, and gluteal sets. She also performed hip abduction against resistance of a theraband while sitting, hip adduction while squeezing a pillow between her legs, and scapular adduction while supine. All exercises were performed bilaterally. The patient was also instructed on contracting her abdominal muscles to help provide core stability.

On the fifth visit, patient reported 0/10 pain in her left shoulder. During therapy, she performed SAQ, SLR, hamstring sets, and hip abduction/adduction from supine using a sliding board. She also performed shoulder protraction and retraction against the resistance of a theraband. All exercises were performed bilaterally. The patient also worked on balancing on the tilt board and practiced step ups on the stairs.

On the sixth visit, the patient reported feeling fatigued from her job. She also reported some pain (1/10) in her left shoulder. During therapy, she performed mini squats, hip abduction while standing in parallel bars, and step ups on stairs. She also performed SAQ, SLR, gluteal sets, thigh fallouts while hook lying with minimal assistance during the adduction phase, and hip abduction/adduction from supine using a sliding board. All exercises were performed bilaterally. She finished therapy with 8 minutes on a NuStep.

On the seventh visit, the patient reported 0/10 pain in her left shoulder. She also appeared to have more energy than usual. During therapy, she performed shoulder flexion with a 2 lb. dumbbell, shoulder abduction with a 2 lb. dumbbell, and shoulder protraction/retraction against resistance of a theraband. She also performed SAQ, SLR, bridges while squeezing a pillow between her thighs, and hip fallouts with minimal assistance. She finished therapy with 8 minutes on a NuStep.

On the eighth visit, the patient reported she had had a bad night sleep. She stated she felt her condition was gradually regressing; however, she did not feel too sore following her last therapy session. During therapy, the patient was given instruction about exercises in the fitness center. She performed hip abduction against 10 lbs. of resistance, knee extension against 25-30 lbs. of resistance, knee flexion against 10 lbs. of resistance, seated rows against 20 lbs. of resistance, and lat (latissimus dorsi) pull downs against 10 lbs. of resistance. All exercises were performed bilaterally. The patient attempted to perform hip

adduction against the lowest resistance level (10 lbs.), but was unable to perform this exercise independently. She also exercised on a NuStep for 8 minutes.

On the ninth visit, the patient reported that she had not been sleeping well recently. She stated that her pain was being managed well, but she continued to feel fatigued. During therapy, she performed seated rows with a theraband, bridges, protraction while supine with a 1 lb. weight, SLR, and chest presses using 3 lb. weights. She also performed hip fallouts while hook lying with minimal assist for adducting thighs, shoulder flexion with a 1 lb. weights and cuing to keep scapula retracted and depressed, and tilt board for plantarflexion/dorsiflexion and inversion/eversion.

On the 10th visit, the patient reported feeling fatigued as usual. During therapy, she performed chest presses using 4 lb. weights, shoulder flexion with 2 lb. weights, shoulder abduction with 2 lbs. of resistance, and scapular protraction/retraction against resistance of theraband. She also performed SLR, SAQ, hamstring sets, hip fallouts while hook lying with minimal assist for adducting thighs, and bridges. The patient also performed hip abduction/adduction while supine and using a sliding board as well as a tilt board for plantarflexion/dorsiflexion and inversion/eversion.

On the 11th visit, the patient reported her legs felt tired prior to beginning therapy and appeared more fatigued throughout therapy. She accomplished her therapy in the fitness center during this visit. She performed hip abduction against 40 lbs. of resistance, leg press against 60lbs of resistance, knee flexion against 30 lbs. of resistance, knee extension against 35 lbs. of resistance, seated

rows with 20 lbs. of resistance, lat pull downs with 10 lbs. of resistance, and arm extensions while standing with 10 lbs. of resistance. The patient received verbal and tactile cues to keep scapula retracted and depressed during upper extremity exercises. She was instructed on proper form for each exercise and demonstrated understanding.

On the 12th visit, the patient reported that they had been sleeping slightly better the last few nights. She also stated she continues to feel fatigued throughout the day. During therapy, she performed seated rows with 25 lbs. of resistance, lat pull downs with 10 lbs. of resistance, arm extensions with 10 lbs. of resistance, leg press with 65 lbs. of resistance, and hip abduction against 40 lbs. of resistance. She finished therapy with 8 minutes on a NuStep.

On the 13th visit, the patient reported she was tired and did not get a good night sleep the previous night. During therapy, the patient's upper extremity strength and ROM was reassessed. Her lower extremity strength was also assessed. After being evaluated, she performed shoulder rows using a theraband for resistance, balanced on foam pad with feet together and on one leg for 30 seconds each, threw a ball with alternating hands while balancing on a foam pad, threw a ball against a rebounder (mini-trampoline angled toward the patient) while balancing on one leg, shoulder protraction with a 3 lb. weight while supine, shoulder flexion with a a1 lb. weight, SLR, hip fallouts while hook lying with minimal assist for adduction, and bridges. The patient was given a 1 month pass to the fitness center for strength and endurance training and then discharged following this visit.

During the 13 visits, the patient was exposed to numerous exercises that she could continue to perform following therapy. Many of the exercises that were introduced during therapy were added to her HEP so long as she had the equipment and they were safe to perform independently. She was encouraged to perform exercises at home on days when she did not have an appointment, but she stated that she only performed her HEP occasionally (2-3/week) due to being fatigued from work.

During her therapy treatment, all exercises were performed bilaterally with alternating limbs if possible. She also alternated between upper extremity and lower extremity muscle strengthening to decrease the chances of fatigue to any single muscle group. Most exercises were performed for 5-10 reps and 2-3 sets. The patient was able to tolerate slightly more resistance toward the end of therapy compared to the beginning of therapy.

Another factor that often contributed the patient's treatment was her state of emotion. Prior to therapy, the patient often expressed prior the therapy that she felt overwhelmed with her diagnosis. This often led to her not being in an emotional state where she was prepared to exercise in the fitness room or other settings. This occasionally resulted in therapy taking place in a private room where exercise options may have been limited.

As a way to provide further education to the patient, she was also invited to an in-service presentation on muscular dystrophy performed for the therapy staff at this facility. This allowed the patient to gain a better understanding of her

condition as well as treatment options for the future. This was presented around her 11th visit and provided as a pro bono service.

CHAPTER IV

OUTCOMES

Following treatment for 6 weeks, MMT indicated that the strength of the patient had stayed about the same. According to MMT, knee flexion decreased from 5-/5 to 4+/5 bilaterally. Shoulder flexion and abduction also decreased slightly from 5/5 to 5-/5 bilaterally. All other muscle groups that were assessed remained the same (see Table 2).

Shoulder and elbow ROM were also similar following her therapy treatment. The only major change was her right shoulder flexion which decreased from 78 degrees to 70 degrees. All other ROM measurements were similar following treatment (see Table 1).

Although strength stayed about the same, the patient scored 51/80 on the UEFI following 6 weeks of therapy, which was an improvement over her score of 43/80 on her initial visit. This score indicated 36.3% impairment, or a 10% decrease in upper extremity functional impairment compared to her first visit (46.3%).

The patient also reported no shoulder pain during the seventh visit and stated that pain was gone throughout the remainder of her therapy. She also reported no pain in her neck or limbs at discharge, but did state she continued to have fatigue in both her upper extremities and lower extremities.

Of the patient's initial 6 goals, only 2 were met. First, she had no shoulder pain following therapy. Secondly, she demonstrated understanding of her HEP and gained the knowledge she needed to continue working on strength and endurance independently.

Although quantitative data does not show improvement in strength or ROM, the patient did have improved upper extremity functionality via the UEFI. Overall, most goals were not met for this patient. However, the patient responded well to their treatment without increases in fatigue or muscle soreness.

CHAPTER V

DISCUSSION

According to the patient's ROM and MMT results at discharge, she either made no improvement or had a slight regression following therapy intervention. However, there are many reasons for these potential regressions. First, both ROM and MMT have a potential for human error. In a study that looked at the reliability of goniometric measurements in patients with DMD, Pandya et al[15] found that intratester reliability varied between .81 and .94. Florence et al[16] concluded the intrarater reliability of manual muscle testing varied between .65 and .93. Due to intrarater error, and because the differences in strength and ROM were not significant during initial evaluation and discharge, it was difficult to assess whether the patient actually declined in strength. It is also important to recognize the fluctuation in the patient's condition and her desire to put forth maximum effort.

Furthermore, a lack of improvement in strength and ROM many be a more acceptable result considering her progressive condition. Had she not received therapy, she may have further regressed due to her diagnosis. Strength and ROM were also only two elements of her treatment plan. She also gained a lot of knowledge about FSHD through education which will likely benefit her throughout

her life. She was educated on numerous exercises that she can continue to perform independently to slow the progress of her condition.

Although endurance was not the main focus of the patients treatment plan, it was one variable. She performed the NuStep on multiple occasions and stated that she really enjoyed it. Unfortunately, the fatigue that she endured from her job played a major role in the amount of energy she had on arrival to therapy. She was consistently arriving to physical therapy fatigued from work as well as her relatively long walk to the therapy department. Many of the conversations with the patient revolved around changing occupations to prevent excessive fatigue of her muscles. She stated that she hoped to change jobs soon, but she was also worried about her financial situation. By finding a desk job, she would likely decrease her overall fatigue and allow herself more energy to perform her HEP.

Throughout the 6 weeks of therapy, it is interesting to consider the improvements she made with her therapeutic exercises. By the end of therapy, she was able to perform hip fallouts with less assistance, leg presses with more weight, and many other exercises to a greater capacity. To prevent excessive muscle damage, she never performed any exercises to her maximal potential; however, she was often able to perform exercises at a higher level with the same amount of effort towards the end of therapy. There are a couple different possibilities for this occurrence. First, she may have actually gained some muscular strength via muscle hypertrophy. However, a more likely reason is neuroadaptation. Due to the repetition of many of her exercises, her neurons may have strengthened their impulses to different muscle groups. Further

research should be done to see if this could result in a muscle increasing its ability to contract without any muscle hypertrophy. While this has been evident in individuals during the first few weeks of resistance training, research may indicate where this change is possible in a patient with FSHD since the nervous system is not affected.

With the diagnosis of FSHD, a lack of upper extremity flexion and abduction was expected. However, there are surgical options for patients to regain upper extremity ROM. One surgery option that was discussed with the patient is called a scapulothoracic fusion. This surgery involves fusion of the scapula, in a slightly abducted position, to the posterior ribs (usually ribs 4-6) to assist the patient with overhead activities.[17] Diab et al[18] examined the outcomes of 11 different scapulothoracic fusion procedures and found that abduction was initially improved from 75 degrees to 145 degrees. After 6.3 years, 7 shoulders still averaged 139 degrees of abduction while 4 shoulders averaged 48 degrees of abduction due to the progressive loss of deltoid muscle strength.

Another treatment option that has shown positive results in the management of muscular dystrophy is the use of neuromuscular electrical stimulation (NMES).[19-22] Colson et al[19] found that NMES was effective at improving shoulder flexion/extension strength and knee extension strength after 5 months of treatment 5 times a week. However, Scott et al[20] found that 8 weeks of NMES treatment to the anterior tibialis muscles was unable to improve strength. Lack of dorsiflexion strength is a major concern of patients with FSHD and often results in frequent tripping. Unfortunately, many studies on muscular

dystrophy patients have found that dorsiflexors are very difficult to strengthen.[20-21]

Overall, this patient had very few objective changes in her ROM and strength with potentially mild regressions in bilateral knee flexion, shoulder flexion and abduction, and right shoulder flexion. However, her treatment was a success in many ways. She learned many new exercises to perform independently, received a significant amount of education to help her be more informed moving forward, and showed improvement in upper extremity functionally via the UEFI.

Reflective Practice

Although this case study had a few positive outcomes, there are many ways in which it could have been improved. The patient received instructions on many different exercises throughout her treatment; however, she was only given written instructions on two different occasions with only a few of the exercises she had been performing. It would likely have been very beneficial for her to receive a list of exercises before discharge to give her an idea of what to work on independently. It may have also been beneficial to give her exercises that she could work on as her condition progresses.

I also never objectively reassessed ROM or strength until discharge. It may have been informative to measure ROM and strength more frequently to observe whether the treatment being provided was making a difference. Some

exercises may not have been beneficial, but it is difficult to know without assessing for improvement.

My goals for this patient may have been unrealistic considering her prognosis. I anticipated improvements in strength and ROM even though this patient's condition was progressive. I should have made goals related to quality of life rather than objective improvements in strength and ROM.

Another area of treatment I could have focused on more was her dorsiflexor strength. She reported frequent tripping, but I spent very little time addressing the strength of her dorsiflexors. Since very little evidence on effective strength training for this muscle group was found, I was apprehensive about treating them in fear of further progressing their decline. However, spending some time on this muscle group may have been beneficial for my patient.

Finally, the patient may have benefitted from NMES. Although much of the research on NMES for FSHD is relatively old, most of it shows very positive results compared to conventional strength training. I also could have instructed the patient on how to use this device independently and given her the ability to continue this treatment option following discharge. Due to her constant fatigue, this may have been a good alternative to maintain or improve her strength.

Even though there were many ways in which this case study could have been improved, the patient did express significant gratitude for the treatment she received. She expressed confidence in her treatment options as well as the many exercises she learned. Her shoulder pain was eliminated following treatment and

her functionality was improved via the UEFI. Overall, the treatment provided to this patient resulted in a positive outcome considering her pathology.

APPENDIX

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A Case Report: Treatment of Facioscapulohumeral Muscular Dystrophy

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Abstract

Purpose: The purpose of the case study was to explain a treatment program involving low/moderate intensity strength training and stretching for a patient with facioscapulohumeral muscular dystrophy (FSHD).

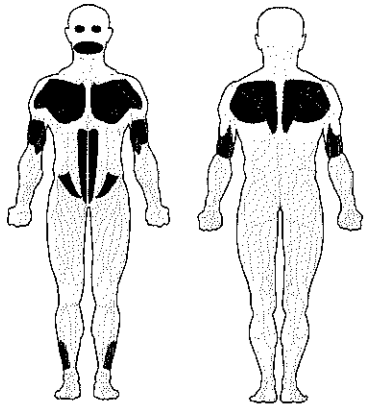
Background: FSHD is the 3rd most common type of muscular dystrophy. This pathology results from a mutation in chromosome 4 and results in progressive weakening of facial, scapular, and hip musculature. Research behind effective strengthening programs is often inconclusive and varies in results.

Case Description: The patient was a 34-year-old female who was diagnosed with FSHD 3 years prior to our first visit. She had complaints of left shoulder pain (2-3/10), shoulder and hip weakness, and limited shoulder range of motion (ROM). The patient reported her lower extremities fatigued and had multiple falls over the last two years.

Plan of Care: Interventions included upper and lower extremity strengthening exercises and shoulder ROM exercises. Prior to discharge, the patient was given an exercise program to continue strength training independently. Patient received a significant amount of education throughout her plan of care.

Outcomes: The patient had no significant changes in strength or ROM at discharge. She reported no left shoulder pain and significant increases in functionality according to an Upper Extremity Functional Index (UEFI).

Conclusion: After 6 weeks of low/moderate intensity strength training and stretching, strength and ROM did not improve, but functionality based on the UEFI did improve.



https://www.mda.org/disease/facioscapulohumeral-muscular-dystrophy

Case Description

A female patient, 34 years old and 3 years post FSHD diagnosis, presented with a history of:

- Left shoulder pain for 3-4 months with gradual onset
- Increased shoulder pain when swinging arm while walking
- Difficulty with overhead activities (washing/combing hair)
- Difficulty with stairs resulting in fatigue and tripping

History included:

- Five previous surgeries on left elbow resulting in limited ROM
- Job required a significant amount of walking and she reported her lower extremities fatigued quickly
- Patient lived alone in an apartment
- Independent with all ADL's/IADL's

Examination and Evaluation

- Scapular winging bilaterally
- Positive O'Brien test on left shoulder which indicated a possible superior labral tear

Strength

Shoulder abduction	5/5	5-/5
Shoulder flexion	5-/5	4+/5
Hip flexion	2/5	2/5
Hip abduction	2/5	2/5
Hip adduction	2-/5	2-/5
Knee extension	5/5	5/5
Knee flexion	5-/5	4+/5

*Strength was equal bilaterally for all strength assessments above. Strength was measured using manual muscle testing with 5/5 indicating normal strength.

Range of Motion

Active R shoulder flexion	78°	70°
Active L shoulder flexion	80°	80°
Active R shoulder abduction	73°	73°
Active L shoulder abduction	65°	65°
Active R elbow ROM	9°-135°	8°-136°
Active L elbow ROM	20°-127°	21°-128°
Passive R shoulder flexion	144°	145°
Passive L shoulder flexion	152°	152°

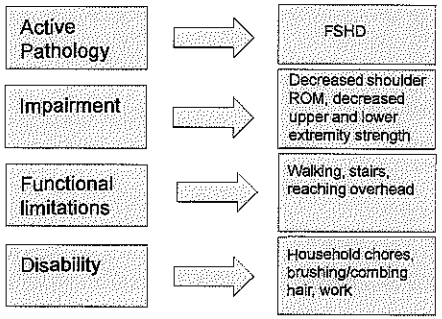
Plan of Care

The patient was seen for 13 visits over 6 weeks. Therapy focused on improving/maintaining strength. The patient received a home exercise program with strength and ROM exercises.

Intervention

- Upper extremity stretching (wall slides for shoulder abduction/flexion)
- Upper extremity strengthening (scapular protraction/retraction with/without resistance, shoulder flexion/extension against resistance)
- Lower extremity strengthening (mini squats, supine hook lying thigh fallouts, short arc quads, straight leg raises, thigh abduction/adduction against resistance)
- Fitness center exercise program (NuStep, knee extension/flexion, thigh abduction/adduction, rows, lat pull downs, chest press)

Nagi Model



Outcomes

- Knee flexion and shoulder abduction/flexion both decreased slightly and all other muscle groups stayed the same (see Strength table).
- ROM did not show significant changes with the exception of active right shoulder flexion decreasing (see Range of Motion table).
- Patient reported 0/10 pain in left shoulder following therapy intervention. Pain was reported at 2-3/10 during initial evaluation using visual analog scale.
- Patient had a significant improvement on the Upper Extremity Functional Index (UEFI) at discharge:
  - Initial Evaluation: 43/80
  - Final Evaluation: 51/80

Conclusion/Discussion

Conclusion

- While this study does not offer conclusive evidence that low/moderate strength training and stretching are beneficial for patients with FSHD, it does show that this treatment plan may be beneficial for some patients.

Discussion

- A study by Van der Kooi et al researched the effects of strength training for FSHD individuals and found dynamic strength could be improved, but not static strength. Although some research supports strength training as an intervention in many cases, not all research does. A systematic review by Voet et al did not show enough evidence to support strength training as an intervention for progressive muscular disorders.
- Neuromuscular electrical stimulation training has been found to be effective at improving strength via manual muscle testing according to a study by Colson et al and should be considered when working with future FSHD patients.
- Although this patient did not make improvements in strength or ROM, she did make improvements in function according to the Upper Extremity Functional Index. She also received a significant amount of education related to her condition, as well as exercises she can continue to do independently to maintain muscular strength.
- When performing physical therapy on patients with muscle disorders like FSHD, it is important to consider the progressive nature of the disease. No improvement in strength also means no decline in strength, which is likely a positive outcome.

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