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A Case Study of Functional Improvements in a Patient with Myotonic Dystrophy Type 1 through Physical Therapy Interventions

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ABSTRACT: This case study involves 30-year-old Jayantibhai who was diagnosed with Myotonic Dystrophy Type 1 (DM1) 6 years ago and is currently presenting with global muscular weakness. Jayantibhai was referred to physical therapy to improve his mobility and manage the effects of his progressive disorder. Jayantibhai and the physical therapy team worked together to create patient-centered goals to guide his treatment plan. Interventions included: diaphragmatic breathing; muscle strengthening; balance, coordination and gait training; and orthotics and mobility aids. After Jayantibhai's participation in physical therapy, he demonstrated improvements in muscular strength, gait speed, and balance, allowing him to successfully ambulate and participate within the community. Jayantibhai will continue to attend physical therapy sessions and be referred to a speech language pathologist and occupational therapist for a home assessment. Despite the significant improvements in Jayantibhai's functional capacity, he still expresses concerns regarding his progressive condition, indicating that he may also benefit from appointments with a social worker and continued physical therapy treatment.

I. INTRODUCTION

Myotonic dystrophy (DM) is a rare progressive disease[1] with multisystem effects seen in the muscles and other body systems[2]. In addition it is very common for patients with DM to present with cardiac conduction defects and cataracts[1]. DM is considered to be a subgroup of myopathies [1] which refers to a variety of diseases which primarily affect the skeletal muscle and leads to muscular weakness[3]. Although physical therapy cannot assist in the reversal or curing of this disease, it can help to manage and slow the progression of the disease, along with minimize possible comorbidities and secondary effects[4].

DM affects approximately every 1 in 8,000 people, making it the most common adult-onset muscular dystrophy[5], despite the disease itself being quite rare. DM is often diagnosed through physical and subjective examinations performed by a Physician[6]. Additionally, since patients with DM often present with cataracts, an optometrist may be able to recognize this and refer the patient to a specialist for further examination and diagnosis[6]. Diagnoses can be confirmed through genetic DNA testing, to confirm whether the genes associated with DM are present[6].

DM is subdivided into two types:

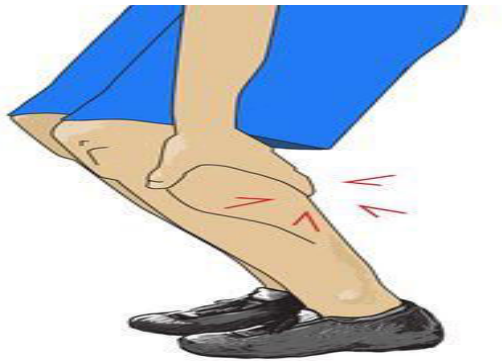

Figure 1: individuals with DM1 suffer from myotonia

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Type 1 Myotonic Dystrophy (DM1) also known as Steinert disease[1] affects both skeletal and smooth muscle, along with the eyes, heart, endocrine system and central nervous system[7]. DM1 is the more common type seen in individuals with DM[8]. DM1 is caused by "expansion of the CTG repeat in the noncoding region of DMPK"[7] with a CTG repeat of greater than 34 repeats in length being considered abnormal[7]. DM1 is further divided into one of three phenotypes including: mild DM1, classic DM1, and congenital DM1, each presenting with different symptoms.

Signs/symptoms of Mild DM1 include: cataracts, and mild myotonia, with a normal life span[7]

Signs/symptoms of Classic DM1 include: muscle weakness and wasting, myotonia, cataracts, cardiac conduction abnormalities, decreased physical capacity, and potential decreased life span[7]

Signs/symptoms of Congenital DM1 include: hypotonia, severe generalized weakness at birth, respiratory insufficiency, intellectual disability and decreased life span[7]

Type 2 Myotonic Dystrophy (DM2) is the less common of the two subtypes. Patients with DM2 present with myotonia and muscle dysfunction[9], similar to DM1; however, effects of other body systems, such as cataracts and cardiac deficits are less commonly seen[9]. DM2 is caused by the "expansion of cytosine-cytosine-thymine-guanine (CCTG) tetranucleotide repeat located in the intron of the CCHC-type zinc finger nucleic acid-binding protein (CNB or ZNF9) gene or chromosome 3q21.3" [1].

Several studies have supported physical therapy as a treatment intervention for individuals with DM1. It has been reported that muscular strength can increase in patients with DM1 when provided with strength training programs [10][11][12]. Specifically, Roussel, Hebert & Duschesne (2020) followed 11 men with DM1 who completed a 12-week lower extremity strengthening program. When compared to baseline, performance in both strength and functional tests improved significantly[12] indicating that this is an effective intervention in the scope of Physiotherapy. Similarly, Missaoui et al. (2010) examined the effectiveness of a rehabilitation program in patients with DM, and found significant improvements in balance (measured by the Berg Balance Scale), gait speed and muscle strength[13]. Evidence supports the importance of physical therapy in management of DM, which is why it is necessary for physical therapists to have a strong understanding of the disease, clinical presentation, and possible treatment interventions.

In this case, I will be following the treatment of 30-year-old male, Jayantibhai who has been diagnosed with DM1 and referred to physical therapy by his physician. Jayantibhai initially came to physical therapy presenting with moderate musculoskeletal weakness, decreased cardiovascular and respiratory function, which affected his ability to ambulate and participate in activities of daily living. This fictional case will discuss the initial assessment, outcome measures, treatment interventions and outcomes throughout Jayantibhai's time in Physiotherapy.

The objective of this fictional case is to provide physical therapists with a resource on how to recognize the clinical presentation of DM1 and provide information on possible treatment interventions and outcome measures.

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II. CLIENT CHARACTERISTICS

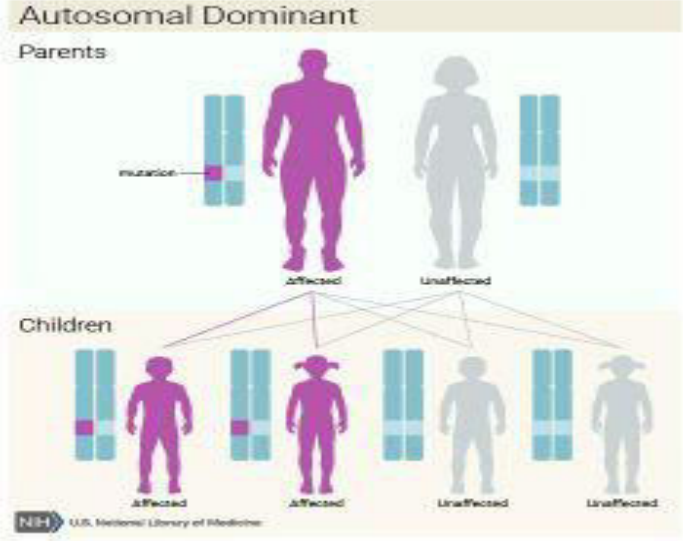

Figure 2: autosomal dominant disorders

The patient in which this case study will be revolving around is Jayantibhai is a 30-year-old male who was diagnosed with myotonic dystrophy Type 1 (DM1) six year ago. Myotonic Dystrophy Type 1 is an autosomal dominant condition that has a multi-system effect. Jayantibhai lives with his wife in their bungalow and helps her with her online business, mostly answering calls as customer support. Jayantibhai was referred to physical therapy by his family doctor to help improve his level of mobility and to try to attenuate some of the negative effects of his progressive condition. Jayantibhai has no other significant comorbidities.

III. EXAMINATION FINDINGS

Upon assessment, it was clear that Jayantibhai had mobility difficulties. Jayantibhai entered the clinic with the use of a single point cane (which he placed in his right hand) but also required the assistance of his wife on his other hand. Jayantibhai requested his wife be present for the duration of the assessment.

Prior to coming to the appointment, Jayantibhai completed the Assessment of Life Habits 3.1 short-form questionnaire which was sent to him by our clinic since this is a self-reported measure been validated for use in DM patients. Jayantibhai's total score was 7 and in particular, his mobility score was 6.

Once Jayantibhai and his wife were situated in one of our patient rooms, Jayantibhai 's subjective assessment was conducted. Jayantibhai noted a consistent and steady decline in his mobility status since being diagnosed with his condition, particularly noticing the effort it takes to walk being the most drastic change. Jayantibhai noted he is having difficulty coordinating his feet and ankles, reporting "sometimes my ankles don't move the way I want them to which is affecting my balance". He also has a hard time completing his ADLs without the help of his girlfriend and notices that within the last year, his breathing has become "a lot more labored". When asked what he hopes to get out of physical therapy, he responded with "be able to walk from the bedroom to kitchen without stopping at his desk to take a break in the morning, and to complete more daily activities independently."

Observation:
Upon observation, with Jayantibhai seated, it appears as if he uses an apical breathing pattern and requires accessory muscle use while breathing. Jayantibhai can sit without any support but sits with a dropped head, though no other postural sitting deviations were noted. Since he is wearing shorts and a t-shirt, it appears as if Jayantibhai also has muscle atrophy of the distal lower extremity and upper extremity musculature. It was also noted that his facial expressions were small, and his speech was slurred occasionally, indicating weakness of the facial and oral muscles. When standing, Jayantibhai struggles to maintain his balance without the use of a gait

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aid and shows significant postural sway while attempting to stand statically. Also, upon a gait assessment, Jayantibhai presents with a drop foot gait pattern throughout the swing phase bilaterally, but more predominantly on his right lower extremity. As a result, Jayantibhai has a decreased heel strike during the initial contact and has a hip hike during the swing phase in order to ensure that his foot can clear the ground while walking.

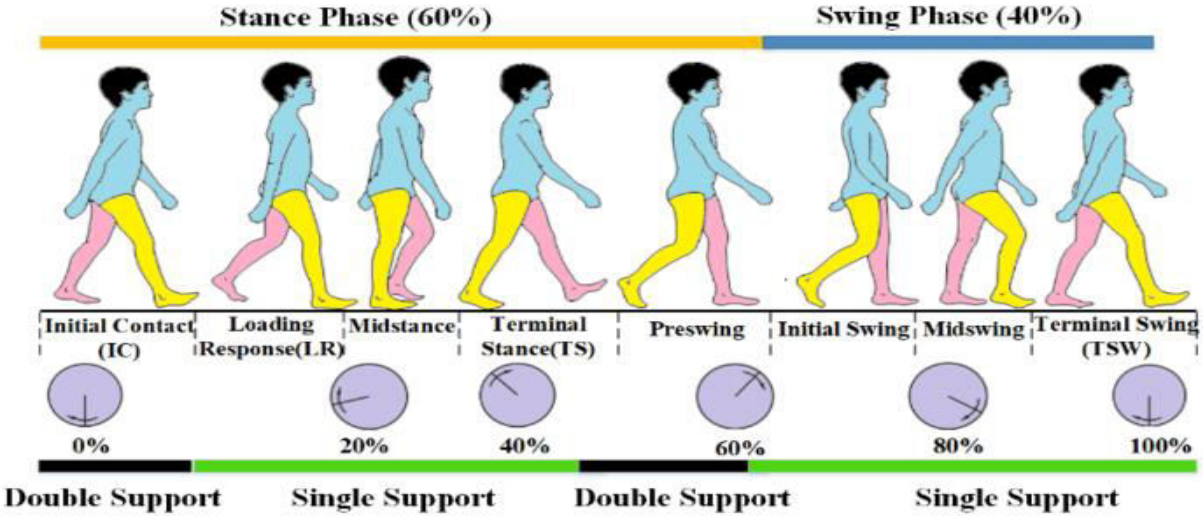

Figure 3: complete gait cycle stages

Muscle Testing:

Movement	MMT Grade	AROM (degrees)	PROM (degrees)
Neck extension	3-/5	0-50	0-80
Shoulder flexion	3+/5	0-160	0-180
Shoulder abduction	4-/5	0-173	0-175
Shoulder extension	4-/5	0-43	0-50
Elbow flexion	4-/5	0-133	0-141
Elbow extension	4-/5	0-2	0-2
Wrist flexion	3+/5	0-60	0-77
Wrist extension	4/5	0- 50	0-55
Hip flexion	4-/5	0- 121	0- 125
Hip abduction	3+/5	0- 34	0- 36
Hip adduction	4+/5	0-26	0-31
Hip extension	3+/5	0-10	0-12
Knee flexion	4-/5	0-120	0-123

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Knee Extension	4-/5	0	0
Ankle dorsiflexion	2-/5	0-3	0-15
Ankle plantar flexion	4/5	0- 35	0-39

Table 1: recorded bilateral average results for upper and lower extremity MMT, AROM, PROM

Patient reported no pain or discomforted on active range of motion, though Jayantibhai informed the physical therapist that he is on medication to manage his underlying myotonia and that "the medication is working well and controls the issue". Passive range of motion was conducted in which a slight reduction in tone was noted and normal joint end feels were noted. Manual Muscle Tests (MMTs) were also conducted on both upper extremities and lower extremities. Of note, Jayantibhai recorded values of 3-/5 for neck extension and 2-/5 for ankle dorsiflexion bilaterally. Additionally, Jayantibhai 's grip strength was tested comparing right and left sided differences, he displayed 23.7kg with his right hand and 22.0kg with his left hand. However, even with the medication, Jayantibhai demonstrated a slight delay in relaxing his musculature after performing contractions. To confirm this, Jayantibhai was asked to repeatedly open and close a fist, which he was able to complete, but demonstrated a slight delay in muscle relaxation during the movement.

Standard measures:
Jayantibhai performed a cluster of tests to further understand his functional limitations and to outline areas of improvement for physical therapy treatment. Jayantibhai was able to complete 12 full repetitions on the 30 second sit to stand test and displayed a speed of 0.62m/s on the 10m walk test. To determine Jayantibhai functional balance a Berg balance test was administered with Jayantibhai receiving a 43/56 as a total score. Also, Jayantibhai recorded a value of 280L/min peak cough flow. The nine-hole peg test was also performed to assess Jayantibhai's finger dexterity and Jayantibhai recorded a value of 20.1 seconds on the right hand and 21.3 seconds on the left hand. Jayantibhai is right hand dominant.

Sensation:
Superficial, deep, and cortical sensations were tested on Jayantibhai as well and all tests were normal.

Neurological:
Reduced tone in upper and lower extremities as noted on passive range of motion and muscle palpation. Reflexes were also diminished when tested but not absent.

IV. CLINICAL HYPOTHESIS

Diagnosis:
Myotonic dystrophy is medically diagnosed through genetic testing. However, the physical therapy diagnosis for Jayantibhai would be, "patient presents with impaired motor function of the distal upper and lower extremities bilaterally, dropped head, impaired balance, apical breathing pattern, and requires moderate assistance for completion of ADLs"

Problem list:
1. Muscle weakness in distal upper and lower extremities due to disuse and muscle atrophy
2. Decreased upper extremity coordination due to underlying neurological impairment
3. Drop foot gait pattern throughout swing phase due to ankle dorsiflexor weakness bilaterally
4. Apical breathing pattern due to weakened diaphragm
5. Dropped head presentation due to weak neck extensors
6. Impaired balance due to coordination impairment and muscular weakness
7. Increased fatiguability due to sedentary lifestyle
8. Impaired muscle relaxation post-contraction due to underlying neurological impairment

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V. INTERVENTION

Patient-centered goals

As Jayantibhai previously indicated, his goals of physical therapy were to "be able to walk from bedroom to kitchen without stopping at his desk to take a break in the morning and to complete more daily activities independently." When taking these goals as outlined by Jayantibhai into account, the following long-term goals were created:

1. Prior to discharge from physical therapy, the patient will be able to achieve a 2-point increase in their score regarding mobility on the Assessment of Life Habits as compared to their initial assessment
2. Prior to discharge from physical therapy, the patient will be able to achieve a 0.8m/s gait speed score on their 10m walk test
3. Prior to discharge from physical therapy, the patient will be able to dress themselves with minimal assistance (no more than 3 points of contact) from the physical therapist
4. Prior to discharge, patient will be able to walk from his bedroom to his kitchen with the use of a gait aid without having to take a break

Once these long-term goals were identified and agreed upon by Mr. J, the following short-term goals were created to meet the long-term goals above:

1. After two weeks, the patient will be able to demonstrate an increase in neck extension MMT from a 3-/5 to a 4-/5 when assessed for a single activation.
2. After two weeks, the patient will be able to demonstrate a peak cough flow equal to or greater than 300L/min
3. After three weeks, the patient will demonstrate an increase in finger flexion strength by increasing hand dynamometer strength by 5 kilograms for each hand.
4. After three weeks, the patient will improve ankle dorsiflexion MMT from a 2-/5 to a 3/5 when assessed for a single activation.
5. After three weeks, the patient will be able to demonstrate an increase in their 30 second sit-to-stand result by 3 repetitions with the use of a chair's armrest
6. After four weeks, the patient will show a decrease in his time to complete the nine hole peg test of 3 seconds for his non-dominant hand and 4 seconds for his dominant hand.
7. After four weeks, the patient will demonstrate an increase in their Berg Balance Scale from 43/56 points to 48/56 points

It is critical to remember that during goal setting, the patient is heavily involved in this process and must have their voice always heard.

VI. TREATMENT PLAN

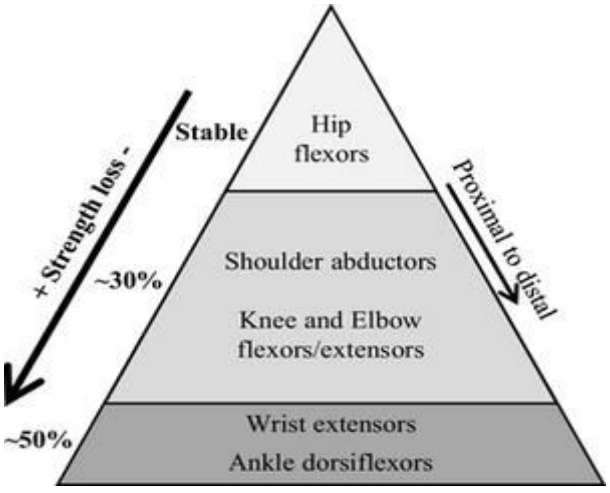

Figure 4: general strength loss in DM1[14]

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When planning intervention strategies to meet the goals above, it is important to remember that DM1 is a progressive neuromuscular disorder. This means that although it is possible for patients who have no experience with physical therapy to show improvements over short periods of time, over a long term (several years), a patient's conditions will worsen naturally. For example, in one study, it was found that over nine years, 30.3-43.5% of individuals with DM1 presented with loss of muscle strength as compared to individuals without DM1 [14]. Therefore, it is the responsibility of the physical therapist to help attenuate the consequences of such a progressive disorder as best as possible.

Interventions provided to Jayantibhai were considered in tandem with Jayantibhai to ensure that he was an active participant in this process.

The following interventions were provided:
Diaphragmatic breathing- to improve diaphragmatic breathing, patient facilitated tactile cued deep breathing was conducted for ten repetitions every waking hour. Such an intervention was used to target Mr. J's apical breathing pattern and to ensure that Jayantibhai is taking deep enough breaths to help facilitate productive coughs if required. Also, preliminary research in healthy individuals shows the potential of diaphragmatic breathing to be associated with improvements in balance[15]. Therefore, Jayantibhai may also potentially receive this add benefit from such an intervention as well.

Strengthening- exercises were provided targeting the initial limitations noted in the assessment which include: finger flexion weakness, ankle dorsiflexion weakness, and neck extensor weakness. The exercises were prescribed under parameters that were identified as being safe for patients with DM1 to perform[16] and were completed to muscular fatigue, not muscular failure. Exercises prescribed can be found in the table below (Table 1) and target the muscle groups that were observed as being weak in Jayantibhai 's assessment. In addition, isotonic exercises were avoided to prevent any exacerbation of Jayantibhai 's underlying myotonia. However, quarter squats and light weight over-head shoulder press exercises were also provided to Jayantibhai under the same parameters to help improve his general fatiguability and general muscular strength. In the literature there seems to be the possibility of those with DM1 to present with skeletal muscle growth[10] and that strength in DM1 patients can be increased when following an exercise program[11]. As Jayantibhai progressed through his therapy interventions, the following exercises were also progressed to more difficult variations or resistance of the exercises below were increased.

Type	Intensity	Reps	Sets	Rest between sets	Times per week
Seated ankle dorsiflexion	Moderate (60% 1RM)	8-12	2-4	2-3 minutes	2-3
Prone Knee extensions					
Mass finger flexion against ball					
Body weight quarter squats					
Light weight over-head shoulder press					

Table 2: strengthening exercises prescription and parameters provided to Jayantibhai

Muscular relaxation- Jayantibhai was educated on the what his underlying myotonia meant and referred back to his physician if his current medication no longer helped him.

Balance- patients with DM1 are seen as similar falls risks to elderly individuals[17] and have impaired ankle strategy usage when balance is lost due to weakness of the ankle musculature [16]**.** Also, the drop foot that a person with DM1 presents with puts them at an increased risk of tripping when walking[16]. Therefore, the dorsiflexion strengthening exercises as listed above may improve balance as well, but balance exercises were also provided to Jayantibhai Based on Jayantibhai's presentation, a prescription of narrow stance static standing holds for ten seconds for ten repetitions daily were provided. As Jayantibhai 's balance improved, exercises progressed to more challenging interventions (tandem stance) and then to more mobile and task-specific interventions (reaching outside base of support). However, given Jayantibhai 's falls risk, such exercises were

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completed in parallel bars and under contact supervision of the therapist. Recent research has also suggested that post-balance intervention with DM1 patients is likely to result in a self-reported increase in balance confidence[18].

Coordination- In order to improve Jayantibhai's coordination, task-specific interventions were used. These interventions included tasks that Jayantibhai deemed meaningful such as: typing, washing the dishes, dressing himself, and being able to open the drawers in his house. Coordination exercises were initially blocked prior to progressing to more random practice as Jayantibhai improved. In addition, time was provided to Jayantibhai after each task for self-reflection and to allow Jayantibhai to problem solve as well as to identify any errors in his task completion prior to him receiving feedback of the results from the physical therapist. However, thumb and finger tapping were also provided to Jayantibhai for at home completion.

Gait training- gait training was a substantial portion of Jayantibhai's therapeutic intervention. However, for such a condition, therapists used a bandwidth feedback approach and allowed the patient time to self-reflect prior to providing feedback on the knowledge of results. Jayantibhai also completed body weight supported treadmill training in a closed environment in order to help facilitate more coordinated gait movements prior to being progressed to gait training in more open environments with great variability (without a balance system) to provide more generalizability to his life environment and to increase his endurance.

Orthotics/Gait aids - given the progressive nature of the condition, Jayantibhai was placed in an Ankle Foot Orthosis (AFO) to help address his foot drop as well as being properly fit, educated, and taught how to use a 2 wheeled walker. Both changes were implemented to help improve Jayantibhai's stability while ambulating. An Occupational Therapist (OT) was consulted in order to ensure all aids provided were appropriate for Jayantibhai's condition.

In addition, a referral was also made to a Speech Language Pathologist (SLP) to help target Jayantibhai's facial and oral musculature weakness. Throughout the intervention process, constant education was provided to Jayantibhai regarding several concepts. Some of these concepts included: his condition, the reason why intervention techniques were chosen, any potential risks or benefits from a given intervention, any alternative intervention strategies, and how DM1 may progress and continue to affect his mobility. The goal of this was to ensure Jayantibhai properly understood the therapy being provided to him and to properly prepare him for what may be his future reality.

VII. OUTCOME

Muscle Testing:

Movement	MMT Grade	AROM (degrees)	PROM (degrees)
Neck extension	3+/5	0-65	0-80
Shoulder flexion	4-/5	0-172	0-183
Shoulder abduction	4+/5	0-174	0-176
Shoulder extension	4-/5	0-46	0-50
Elbow flexion	4+/5	0-135	0-143
Elbow extension	4-/5	0-2	0-2
Wrist flexion	4/5	0-64	0-78
Wrist extension	4/5	0- 50	0-55
Hip flexion	4/5	0- 124	0- 126

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Hip abduction	4-/5	0- 36	0- 38
Hip adduction	4+/5	0-26	0-31
Hip extension	4/5	0-11	0-12
Knee flexion	4/5	0-123	0-125
Knee Extension	4/5	0	0
Ankle dorsiflexion	3/5	0-5	0-15
Ankle plantar flexion	4+/5	0- 37	0-39

Table 3: recorded bilateral average results for upper and lower extremity MMT, AROM, PROM prior to discharge

Grip strength improved slightly: R 29.1kg L 26.9kg.
30 second sit-to-stand test improved to 15 full repetitions.
10m walk test improved to 0.78m/s which is just below the 0.8m/s cut-off required for safe community ambulation, though Jayantibhai was able to complete his goal of walking from his bedroom to his kitchen without having to take a break.
Peak cough flow increased to 310L/min.
Nine hole peg test- right hand: 16.73s, left hand: 18.12s.
BERG Balance Scale score improved to 48/56 which indicates that his balance meets the score to be able to ambulate without a mobility aid. However, this improvement in score is also highly attributed to the improvements in strength of the lower extremity which allowed for better control of motor movements when completing dynamic items in this test such as, sit-to-stand and transfers. Therefore, Jayantibhai will continue to use a mobility aid (2 wheeled-walker) when ambulating long distances. This improvement was also attributed to the balance interventions that improved his ankle strategy.

10mWT improved to 0.78m/s, which is a speed that places Jayantibhai as a potential community ambulator. This improvement was due to a combination of improved aerobic capacity, strengthening of the respiratory muscles and learned deep breathing techniques. Although his lower extremity strength improved, he still had difficulty ambulating for long periods due to ankle dorsiflexor fatigue, though Jayantibhai was able to ambulate from his bedroom to his kitchen with the use of an AFO.

The discharge planning includes treatment techniques and referrals to specialists to reduce the long-term changes that will occur with DM1. Since it is a progressive disorder affecting multiple body systems, it requires a multidisciplinary treatment and management approach[19]. After the 6-week treatment intervention, Jayantibhai will continue to receive physiotherapy treatment once per week to monitor the disease progression and maintain the benefits gained from the treatment program. As noted in the initial observation, Jayantibhai exhibited signs of facial and oral muscle weakness. Therefore, referral to a speech language therapist would be beneficial to assist in the activation and maintenance of the strength, endurance, and coordination of those muscles[20]. As well, as the condition progresses, Jayantibhai may experience difficulty swallowing and eating so this would help him learn effective strategies to do so safely and effectively. Along with physical therapy once weekly, to slow the progression of muscle atrophy, Jayantibhai will be referred to a community-based exercise program close to his home, where he can continue to do light to moderate strength and endurance exercise 3-5 days per week which will also benefit his cardiorespiratory system function. The weakening of respiratory muscles results in symptoms of dyspnea, so Jayantibhai will be educated by the physical therapist about self-management positions he can do to relieve these symptoms. This includes sleeping in side-lying and upper body elevation as well as the tripod position when sitting. Before returning home, the occupational therapist will assess the safety of Jayantibhai's home and make suggestions for any adaptations that would improve his mobility and ability to be independent in his activities of daily living (i.e. grab bars in the bathroom, removal of carpets)[20].

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Approaching the end of his 6-week intervention, Jayantibhai was feeling more confident in his community ambulation and slightly stronger. He was grateful for the knowledge he learned about his condition and the things that could be done to slow its progression; however, he still expressed worry about the effect it would have on his quality of life in the future, Therefore, Jayantibhai will also benefit from a referral to a social worker to help him cope and remain socially integrated [20].

VIII. DISCUSSION

Type 1 Myotonic Dystrophy (DM1) affects both skeletal and smooth muscle, along with the eyes, heart, endocrine system and central nervous system[7]. Signs and symptoms can include muscle weakness and wasting, myotonia, cardiac conduction abnormalities, decreased physical capacity, and potential decreased life span[7]. Fortunately, several studies have supported physical therapy treatment interventions and interdisciplinary care for individuals with DM1. Dysphagia is a common symptom of DM1, Parkinson's Disease and Multiple Sclerosis, therefore, a referral to a speech language therapist is an important step to take for proper management of these conditions[21]. Jayantibhai is one of the many people who, through rehab and education, focused on task-specific and functional activities meaningful to his daily life and was able to slow the progression of his disease and gain confidence. Physical therapists play a crucial role in preventing progression, but also in teaching compensatory approaches to maintain patient functional independence and participation. Physical therapists also play a large role in communication with other health care providers, the patient and the patient's family/ caregivers. With the use of diaphragmatic breathing and lower extremity strengthening, and evidence of assistive devices (AFO, 2WW), Jayantibhai was able to achieve most of his goals. The development of rapport with patients combined with collaborative goal setting and treatment planning using the International Classification of Functioning, Disability and Health (ICF model), physiotherapists can help people with DM1 reach their goals. Ultimately, this influences their quality of life and outlook on living with a progressive disease.

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