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Case Report

The High-level Mobility Assessment Tool (HiMAT) in Myotonic Dystrophy type 2: A Case Report

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Abstract

Myotonic dystrophy type 2 (DM2) is a progressive muscular dystrophy with multi-system manifestations and can affect functional mobility, gait, and balance. Currently, there are few reports of functional outcome measures in this population. This case describes the change in function detected by the High-level Mobility Assessment Tool (HiMAT) and 30-second Chair Stand Test (CST) in a high-functioning adult female with DM2 after physical therapy treatment. The patient's chief complaint was muscular pain and fatigue that affected activities of daily living (ADLs). Multi-modal physical therapy focused on neuromuscular re-education, balance and strength training, patient education, and moderate cardiorespiratory training. Improvements were observed in manual muscle testing and in single-limb standing balance. A nine-point total improvement was observed in HiMAT score, and the CST detected a 100% improvement in number of sit-to-stands. The patient returned to prior level of function and was able to resume ADLs and recreational activities without complaints of muscular pain or fatigue.

Keywords: Physical function in DM2; HiMAT; Physical therapy; Neuromuscular rehabilitation; Chair stand test; Functional outcome measures

Abbreviations: ADLs – activities of daily living; CCTG – tetranucleotide (cytosine-cytosine-thymine-guanine) expansion; DM1 – myotonic dystrophy type 1; DM2 – myotonic dystrophy type 2; J.D. – patient pseudonym (Jane Doe); LE – lower extremity; LLE – left lower extremity; LOB – loss of balance; MMT – manual muscle testing; PROMM – proximal myotonic myopathy; PT – physical therapy / physical therapist; RLE – right

Introduction

Myotonic Dystrophy type 2 (DM2), or Proximal Myotonic Myopathy (PROMM), is a late-onset inherited and progressive muscular dystrophy with multi-system manifestations [1-5]. The etiology of DM2 is traced to a tetranucleotide (CCTG) repeat expansion located on the CNBP gene resulting in abnormal cardiac and skeletal muscle function [6-9]. Despite having similar features to Myotonic Dystrophy type 1 (DM1), DM2 is clinically distinct, originating from a different gene locus [1,2,4,10]. DM2 is characterized by a milder clinical presentation, later onset of symptoms, and more proximal versus distal weakness [4,5,9,11]. Other clinical features include somnolence, weakness, muscle pain and stiffness, cramping and fatigue, which impact quality of life [1,6,8]. Compared to DM1, DM2 is less severe and the exact prevalence is a topic of ongoing study [7-9,12].

Similar to DM1, a hallmark feature of DM2 is myotonia [6-8]. Grip myotonia is often the first symptom experienced with noticeable delay in the relaxation of the long finger flexors [6,8]. Myotonia may also be noted in the calf musculature or in the thumb. DM2 selectively affects the neck flexors, long finger flexors, elbow extensors, and proximal girdle musculature [1,6,8]. Hip flexor and extensor weakness can disrupt functional mobility, particularly during ambulation and stair climbing [3,5-8,11]. Persons with DM2 may vary in the degree to which the disease impacts functional mobility [6]. Therefore, any number of physical performance or functional outcome measures may be selected to monitor change or response to treatment. However, there are few published studies that have utilized functional outcome measures in the assessment of persons with DM2.

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The High-level Mobility Assessment Tool (HiMAT) appears to be an appropriate measure of functional mobility for persons with DM2. The HiMAT has previously demonstrated sound psychometric properties and has been normative referenced in healthy subjects [13-19]. These properties have led to the application of the HiMAT as a valid and reliable measurement tool in the outpatient rehabilitation of persons with other neurological disorders. Furthermore, repetitive sit-to-stand testing is frequently used as an indicator of functional strength in the lower extremities and has been widely studied across populations [20-23]. However, to date, there have been no published reports examining the use of either the HiMAT or the 30-sec Chair Stand Test (CST) in persons with DM2. Therefore, the purpose of this case report is to describe the use of the HiMAT and CST in a patient with DM2 undergoing an outpatient multi-modal Physical Therapy (PT) treatment.

Case Report

Patient Background

An ambulatory 46-year-old Caucasian female, referred to as Jane Doe (J.D.) and diagnosed with DM2 by a neurologist at age 33 years, presented to an outpatient PT clinic with no apparent functional limitations. J.D. reported independence with all functional Activities of Daily Living (ADLs). J.D. is a stay-at-home mother of three school-aged children. A typical daily schedule consists of moderate physical activity demands during meal preparation, basic and heavy-duty housekeeping (cleaning/laundry), driving, shopping and running errands. The chief complaint was muscle pain and weakness during prolonged activity, especially squatting and walking, with an overall goal to restore strength and return to running and gym-based exercises. J.D. reported experiencing pain (5/10 at worst) in her legs and difficulty with sit-to-stand activities. J.D. had been unable to participate in normal exercise for the prior six months due to disease-related symptoms. Written informed consent was obtained prior to treatment, as well as for inclusion in this case report.

Examination

Upon PT examination, a standing postural assessment revealed lumbar hyperlordosis and knee hyperextension. A slight resting tremor was noted bilaterally in the upper and lower extremities. No deficits in active or passive range of motion were found in the upper or lower extremities. Isometric break testing revealed grossly normal strength for bilateral upper extremities. Manual Muscle Testing (MMT) was performed more extensively in the lower extremities and results can be found in Table 1. MMT was performed according to procedures and positions outlined by Kendall FP et al. [24] Proximal muscle weakness was noted in the hip and knee joints with the left side being slightly more affected. Ankle dorsiflexion strength was found to be within normal limits. Ankle plantarflexion strength was tested using the multiple heel-rise in standing with no deficits. However, a prolonged lowering time after heel-rise was observed indicating a delayed muscle relaxation consistent with myotonia. Observational gait analysis revealed independent ambulation and a normal reciprocal pattern without noticeable deviations. J.D. could successfully ascend and descend a flight of stairs using a reciprocal pattern without the use of upper extremity support but was slower and appeared cautious.

Outcome Measures

High-level Mobility Assessment Tool (HiMAT): The HiMAT was selected to assess the response to more challenging mobility demands in this case. The HiMAT was originally developed to assess balance in high-functioning survivors of traumatic brain injury to quantify the need for assistance with functional mobility [13-19]. The tool requires that participants be able to ambulate at least 20-meters without assistive devices or orthoses [13]. The HiMAT contains multiple test items, including: walking, running, skipping, hopping, bounding, jumping, and stair negotiation [13-15]. Participants complete each item at the safest maximum speed possible [13-15]. The test is hierarchical in nature and consists of thirteen total items based upon a five-point scale, where higher scores equate to increasing performance, as seen in Table 2. Normative values of the HiMAT have been established for healthy young adults (ages 18-25 years) with high test-retest reliability (ICC=0.88, 95% CI 0.82-0.92) [13,18]. The HiMAT has demonstrated great psychometric properties including external concurrent validity, excellent reliability [0.88-0.99], high internal consistency [0.91-0.95], and low standard error of measurement (SEM)[0.79-1.36] [16-19].

Chair Stand Test (CST)

Functional strength of the lower extremities was assessed using the CST [20]. This test was selected as it requires repetitive movement using proximal musculature over 30 seconds. This test allows for the observation of the effects of proximal weakness or the prolonged muscle relaxation characteristic of DM2 on functional mobility. Strong test-

Table 1: Results of Manual Muscle Testing.

Muscle / Muscle Group (test position)	Initial Evaluation		Week 4 Re-evaluation		Week 8 Re-evaluation prior to d/c	
	Right	Left	Right	Left	Right	Left
Hip Flexors (seated)	4+/5	4+/5	4+/5	4+/5	4+/5	4/5
Hip Abductors (side lying)	4+/5	4/5	4+/5	4/5	5/5	4+/5
Hip Adductors (side lying)	4+/5	4/5	4+/5	4/5	5/5	5/5
Gluteus Maximus (prone-knee flexed to 90deg.)	3+/4	3/5	4/5	4/5	4+/5	4/5
Biceps Femoris (prone-knee extended)	3+/5	4/5	4+/5	4+/5	5/5	4+/5
Knee Flexors (prone)	3+/5	3/5	5/5	4+/5	5/5	4+/5
Knee Extensors (seated)	3+/5	3/5	4+/5	4+/5	5/5	5/5

*Manual muscle testing (MMT) grading scale according to Kendall & McCreary, Muscles: Testing and Function (1983)

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retest reliability and validity of the CST have been found in healthy community-dwelling adults, among other populations [20-25]. This test was performed in accordance with prior studies [20-25].

Standing Balance Assessment

Clinical balance assessment was performed using the single- and double-limb standing balance test with eyes-opened or eyes-closed on a firm surface. This test was performed in accordance with prior studies and occurred over 30 seconds in each position [26].

Treatment

J.D. received PT treatment once a week for eight consecutive weeks.

The goals of PT intervention were to:

- 1.) Return to prior levels of daily function and physical activity without onset of fatigue, pain, or weakness;
- 2.) Return to a gym-based fitness program and recreational running; and
- 3.) Restore quality of life.

PT interventions included light to moderate resistance training of the pelvic and shoulder girdle muscles and neuromuscular re-education techniques concentrating on improving motor and postural control to prevent movement compensations. Postural stabilization techniques in developmental positions such as high kneeling and quadruped were utilized. Tactile and verbal cueing was utilized to correct movement dysfunction during treatment sessions. Single limb stance balance training, reciprocal movement coordination training, and cardiorespiratory conditioning were also employed. For this case, submaximal cardiorespiratory training was performed at a light-moderate intensity (40-60% of HRR) and slowly progressed as tolerated [27] The Borg rate of perceived exertion (RPE) (6-20) was used to monitor patient effort during all activities. Patient education stressed complete muscle relaxation between repetitions and limited repetitions based upon movement quality. A key component of the treatment plan included tracking the response to treatment to allow for the optimal modification and progression of exercise dose and intensity. Therefore, J.D. kept a daily log of activities and recorded muscle pain, weakness, fatigue and functional decline. Each treatment session began with an activity log review and served as the basis to progress or modify the PT treatment plan. A summary of the patient activity log (response to prior treatment), clinical reasoning, and intervention for each PT visit can be found in Supplemental Table 1.

Results

At the completion of eight weeks in PT, strength, single-limb standing balance, and functional performance improved. Results of MMT can be found in Table 1. Hip and knee strength improved on

Table 2: HiMAT results at initial examination, re-evaluation, and discharge.

HiMAT item	Initial Evaluation		Week 4 Re-evaluation		Week 8 Re-evaluation	
	Performance	Score	Performance	Score	Performance	Score
Category						
10-m Walk	4.62 s	3	4.13 s	4	4.67 s	3
10-m Walk Backward	10.27 s	2	5.51 s	4	6.02 s	3
10-m Walk on Toes	6.64 s	3	5.13 s	4	6.00 s	3
10-m Walk over Obstacles	6.15 s	2	4.61 s	3	3.92 s	4
10-meter Run	2.32 s	2	1.85 s	3	1.58 s	4
10-m Skip	3.32 s	3	3.01 s	3	3.29 s	3
10-m Hop Forward	5.40 s	2	5.13 s	3	6.31 s	2
Bound- Affected LE	93.1cm	2	100.43cm	2	121.6cm	3
Bound – Less-Affected LE	105.3cm	2	110cm	3	124.7cm	3
Up Stairs- Dependent	Reciprocal no rail	5	Reciprocal no rail	5	Reciprocal no rail	5
Up Stairs-Independent	7.66 s	2	6.82 s	3	5.22 s	4
Down Stairs-Dependent	Reciprocal no rail	5	Reciprocal no rail	5	Reciprocal no rail	5
Down Stairs- Independent	5.54 s	4	4.43 s	4	3.08 s	4
Total Score ___/54		37		46		46

High-Level Mobility Assessment Tool1-3, cm – centimeter, m – meter, LE – Lower extremity, s – seconds

MMT. J.D. returned to exercising 4-5 times/week alternating between the gym, running, and a home exercise program. Single-limb balance eyes-open on a firm surface improved from 25 sec (LLE) and 30 sec (RLE) to 60 sec bilaterally; single limb balance eyes-closed on a firm surface improved from inability to perform (0.0 sec) (bilaterally) to 17.29 sec on RLE and to 5.65 sec on LLE.

Results for the HiMAT can be found in Table 2. On initial examination J.D. scored 38/54 and fell below the normative value range of 44-54 for a healthy adult female aged 18-25 [13,15-17,19]. At week 5, her score on the HiMAT improved by 15% to a 46/54. The HiMAT score was stable between week five and the final visit. Initially, J.D. completed 10 stands in the allotted 30-seconds of the CST and scored 15%^{ile}, equivalent for a woman aged 60-64 years [20]. Upon completion of the test she reported a score of 18 “very hard” Borg RPE (6-20) [29]. The CST improved by 50% to 15 stands on reassessment at week five and to 20 stands prior to discharge at week ten, yielding a 100% total improvement. This resulted in a total change from 15%^{ile} on initial evaluation to 75%^{ile} prior to discharge on comparative normative data.

Discussion

Overall, there is a dearth of published outcome measures for persons with DM2. This case highlights the utility of the HiMAT and the CST as novel and appropriate measures for further investigation in the DM population. This case provides an example of how PT intervention assisted a person with DM2 and demonstrates improvements in balance and function. The results of this case report support the value of PT in helping persons with DM2 learn about managing their condition to optimize functional mobility and activity participation.

In this case, the HiMAT detected an overall improvement in function that persisted across weeks of repeated measurement. J.D.’s ability to perform repetitive sit-to-stand was reflected in the 100% change from the baseline score. The increase in the HiMAT score on reassessment was further supported by her return to prior level of function and gym-based exercise and running. The increase in some HiMAT score categories and decrease in others could possibly be explained by a cumulative effect of muscles fatigue on performance through the test item progression, or by a potential training effect. Further inquiry on the HiMAT and CST is warranted across neuromuscular populations to establish reliability and validity of the tool. Future research and clinical

trials should consider utilizing the HiMAT and CST as a measure of physical function to monitor change in response to intervention.

Critical to this case was continually evaluating the patient’s response to treatment using an activity log. Managing the immediate effects of DM2 during PT treatment included allowing extra time for muscle relaxation between exercise repetitions and limiting exercise to a muscle group indicated by cramping and/or onset of tremor. Patient education was essential to the patient’s progress to address complete muscle relaxation between repetitions to reduce myotonia. The integration of developmental postures in the PT treatment plan was instrumental in promoting motor learning of the proximal and core stabilization muscles in the absence of movement substitutions patterns or compensations.

DM2 is a rare neuromuscular disorder. Persons with DM2 may benefit from physical therapy treatment to address functional impairments such as muscle weakness, pain, stiffness, decreased endurance, and activity limitations that are commonly associated with the disorder. Furthermore, other clinical features of DM2 include conductive cardiac disease and progressive cardiomyopathy [2,5,7,11,30,31]. The frequency and severity of cardiac involvement in DM2 is less than in DM1 [32]. However, the risk for cardiac arrhythmia and other complications do exist [32]. Physical therapists should be aware of the potential for asymptomatic cardiomyopathy, heart failure, and risk for serious cardiac complications when treating persons with DM2. Therefore, physician clearance prior to initiating an exercise program is imperative and should always be obtained. However, the parameters of an optimal cardiorespiratory exercise program are not yet fully understood for persons with DM2 and should be a topic of future study.

There are limitations to this case report, including a single-subject report. The lack of an objective quantitative measure of muscle strength (i.e., handheld dynamometry) was also a limitation in this case report. Therefore, the results and interpretation of the responsiveness of the HiMAT in this case is cautioned as a possible training effect could have impacted the change in performance. Additionally, various mechanisms have been found to generate musculoskeletal pain in DM2 including exercise, temperature, and palpation [32]. Routine physical activity may help to control musculoskeletal pain, and this appears to be supported in this case [33]. Rehabilitation providers should understand

the potential for symptom exacerbation with resistive exercise in persons with neuromuscular disorders. However, currently, it is not yet fully known how prescribing resistive exercise can affect the degree to which skeletal or cardiac muscle function may be enhanced or further impaired in DM2. Similarly, it is not yet fully understood how various levels of exercise training may enhance or impair myotonia [30]. Therefore, therapists should use caution when implementing a strength or cardiorespiratory training program in persons with DM2.

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Statement of ethics

The authors disclose no conflict of interest. The authors received no funding support in the completion of this work. The authors confirm that they have read the Journal's position on issues involving the ethical reporting and affirm that this report is consistent with those guidelines.

Implications for Rehabilitation

- The High-level Mobility Assessment Tool can detect change in function in DM2.
- The 30-second Chair Stand Test can detect improvements in muscle strength in DM2.
- Improvements in manual muscle testing were noted after physical therapy treatment.
- Exercise and physical therapy can promote return to function in a person with DM2.

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