



# Role of Physical Therapy in the Assessment and Management of Individuals with Myotonic Dystrophy

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# INTRODUCTION

Physical therapists (PT) are healthcare professionals who hold a post-baccalaureate graduate degree (MPT, DPT) from an accredited college or university. They also may be certified specialists in an area of expertise, such as pediatrics (PCS), geriatrics (GCS), neurologic (NCS), cardiopulmonary (CCS) or orthopedic physical therapy (OCS). Physical therapists practice in a variety of settings including hospitals and nursing homes, outpatient clinics, home health, and schools. Most individuals with myotonic dystrophy (DM) will probably first encounter a physical therapist in the multidisciplinary clinic where they receive care for their DM related problems. In this setting, the physical therapist plays a consultative role providing evaluation, education, instructions and recommendations based on individual patient needs. They may also act as a liaison and help coordinate care with school or community based therapists who may be providing direct care services.

Common areas that are addressed by physical therapists are related to exercise/physical activity, pain and/or fatigue management, orthotics/braces and assistive/adaptive equipment. The goals of physical therapy management for DM are to maximize functional ability, delay and prevent secondary complications associated with DM and aging, and improve quality of life for individuals with DM.

Myotonic dystrophy is the most common form of muscular dystrophy in adults. It is an autosomal dominant disorder, meaning that a person carrying the gene has a 50-50 chance of passing it on to a child. It is a multi-systemic progressive disorder that affects the muscular, respiratory, cardiac, nervous, gastrointestinal and endocrine systems. Currently two variants of DM are recognized; DM1 which arises from a defect on chromosome 19 and DM2 which results from a defect on chromosome 3.<sup>1</sup> DM1 was first described in the early 1900's thus is a more studied entity while DM2 was described in the late 1990's. DM1 and DM2 share many common features, but there are also significant differences. Individuals with DM1 can present with symptoms at different ages; at birth (congenital), during childhood (pediatric), during adulthood, or later in life, thus four clinical phenotypes are described in the literature. Congenital phenotypes have not been described in DM2 and most patients present in adulthood.

Weakness and muscle wasting (atrophy) are prominent features in DM1 whereas muscle pain and myotonia are prominent in DM2. Individuals with DM1 primarily exhibit facial and distal limb weakness whereas individuals with DM2 exhibit proximal weakness. Muscle related problems such as weakness, wasting and functional difficulties are very often the concerns that lead individuals to seek attention and help from physical therapists. However, DM is a multi-systemic disorder thus it is essential to understand all of the systemic complaints and help manage the muscle-related symptoms in the overall context of concerns for an individual.<sup>1-3</sup> Congenital and childhood onset DM1 have unique features so the physical therapy management of these conditions are addressed separately later in this section.



# PHYSICAL THERAPY ASSESSMENT

During an initial physical therapy evaluation, the physical therapist will obtain a detailed history of the patient's symptoms and/or problems, how they have changed over time, factors that make them better or worse and how they affect the daily activities and lives of the affected individual. Information regarding the person's occupation, lifestyle, leisure activities, and their role in the family unit is essential to the evaluation process.

As DM is a systemic condition, it is important for the physical therapist to perform a systems review according to the *Guide to Physical Therapy*,<sup>4</sup> including review of cognition, communication, musculoskeletal system, neuromuscular system, cardiovascular/pulmonary system, and integumentary/skin system.

Individuals with DM can have difficulties in both cognition and communication.<sup>1-3</sup> Symptoms include somnolence, apathy, specific personality traits, deficit in executive functions, depression and fatigue. These cognitive deficits may impact a person's ability to follow through with recommendations and are important to take into consideration when establishing a plan of care or management program. Communication difficulties can arise as a result of weakness of the facial muscles as well as the presence of myotonia in the jaw and tongue. This not only impacts proper communication between patients and care providers, but also has an effect on social communication leading to some of the psychosocial issues mentioned previously.

The neuromuscular and musculoskeletal systems are often the focus of the examination, as weakness and resulting functional difficulties are often the most disabling features of the disorder. The most common pattern of muscle involvement in DM1 includes the facial muscles (masseter and temporalis), neck muscles (sternocleidomastoid), long finger flexors of the hand and distal ankle muscles.<sup>1-3</sup> Muscle involvement usually begins in the teens,

twenties or thirties and is slowly progressive. The weakness progresses from the distal to proximal muscles. Muscular weakness in congenital myotonic dystrophy presents during the neonatal period with generalized hypotonia. In DM2, the muscular involvement is predominantly proximal (trunk, shoulders and pelvis) and also slowly progressive, beginning in the 'mid-adult' life.<sup>1</sup> It is critical that physical therapists are knowledgeable about manual muscle testing for all muscles, as the pattern of weakness can be predictive of both the disease itself as well as mobility concerns that may arise. Strength can also be measured more objectively by hand-held dynamometers as well as fixed systems such as the Quantitative Muscle Assessment (QMA) system. QMA systems are often utilized in the research setting. Normative data for both of these methods have been established in the pediatric and adult populations.<sup>5-9</sup>

Myotonia is the other musculoskeletal manifestation of myotonic dystrophy. Myotonia is the inability to relax a muscle after a forceful contraction. Individuals with myotonia affecting the hand musculature often report difficulty releasing their grip after a vigorous handshake, which can create embarrassing social situations. Complaints of myotonia are also reported in the jaw and tongue, leading to difficulties with speech, swallowing and chewing.<sup>1,3</sup> Myotonia in the leg muscles may lead to difficulty with movements like climbing stairs, running, walking, etc. Symptoms of myotonia may also be present in other parts of the body. Often patients will report that their myotonia symptoms are worse in cooler temperatures. Myotonia can be managed with medications such as Mexilitene.<sup>10, 11</sup>

The progressive decline of strength in DM results in limitations that affect functional tasks, including the ability to get up from a chair, ambulate, and climb stairs. These functional activities can also be timed and used as outcome measures to monitor the progression of the disease or to document benefits of interventions.

Assessment of hand function, including grip and pinch strength, is also important in this population. Detailed information related to hand function testing and treatment is provided in the section on occupational therapy.

Individuals with DM also report decreased balance abilities.<sup>14, 17</sup> Balance requires the interaction of sensory systems (visual, vestibular and somatosensory), the musculoskeletal system (range of motion and strength), and the central nervous system. It is important to assess static and dynamic balance as well as balance reactions/strategies to determine appropriate recommendations for safe mobility.<sup>14, 17</sup>

The cardiovascular system can be compromised by the presence of cardiac arrhythmias and conduction defects as well as involvement of the cardiac muscle itself.<sup>1</sup> Insufficiency of the respiratory system may be a result of both myotonia and weakness in the muscles that control respiration.<sup>1</sup> Respiratory muscle involvement often leads to a reduced vital capacity later in the disease. Individuals with DM1 who have reduced respiratory function are often at higher risk for pulmonary complications such as pneumonia.<sup>3</sup> When making exercise recommendations for a home program, it is essential to educate individuals about how to monitor their cardiorespiratory responses with simple tools like heart rate monitors, Borg scale, etc. It is essential that individuals report their responses to exercise to the person overseeing and/or monitoring the home program. Exercise response may be effectively monitored by using the American College of Sports Medicine (ACSM) recommended FITT guidelines (Frequency, Intensity, Time, Type).<sup>18</sup> It is also important to explain the need to continue breathing during exercise as holding ones breath may affect the heart rhythm. Depending on the progression of their disease, individuals with DM may have limited exercise tolerance, thus should be monitored carefully.

Pain and fatigue are common complaints among individuals with DM1 and DM2.<sup>19-22</sup> In a study by Peric et al. the frequency of pain was found to be similar between individuals with DM1 (88.5%) and DM2 (86.4%).<sup>19</sup> Additionally, similar patterns of pain have been reported by individuals with DM1 and DM2 with complaints of pain most commonly in the low back and legs.<sup>19, 22</sup>

More than 60% of patients with neuromuscular disorders complain of fatigue.<sup>20</sup> Fatigue can have a major impact on the employment status of patients with DM.<sup>23</sup> In addition, disordered sleep, a common symptom of DM, may contribute to fatigue.<sup>2</sup> These symptoms can have a significant impact on a person's quality of life and socialization. Due to the multifaceted nature of cognitive and physiological fatigue and pain, these complaints should be addressed by a multidisciplinary team and included in the physical therapy assessment and treatment plan.

Last, many individuals with DM have gastrointestinal manifestations that may be present anywhere along the digestive tract.<sup>24</sup> Symptoms reported span the spectrum of dysphagia and heartburn to abdominal pain and changes in bowel function.<sup>25, 26</sup> Involvement of the GI system may be very disabling to the individual and may impact the person's ability to participate in employment, societal roles and exercise programs.

# PHYSICAL THERAPY MANAGEMENT

## Exercise

Exercise, including range of motion (flexibility), resistive (strengthening), cardiovascular (aerobic) and balance training, is important for the management of the musculoskeletal and cardiorespiratory manifestations of myotonic dystrophy. An appropriate exercise program should include all types of exercise.

Current recommendations from the U.S. Department of Health and Human Services (HHS) suggest that for all individuals, some activity is better than none and that the health benefits of physical activity far outweigh the risks. For individuals, including those with chronic disease, these recommendations include five hours per week of moderate intensity exercise and strengthening two times per week. If individuals with chronic conditions are unable to meet those recommendations, they should perform as much activity and/or exercise as their condition allows and avoid inactivity.<sup>27</sup>

The terms physical activity and exercise are often used interchangeably, however, physical activity is activity that is performed during daily activities, whereas exercise is a structured activity for the purpose of body conditioning, maintaining fitness, or improving health.<sup>28</sup>

Aerobic activities at a moderate intensity level are activities that raise the heart rate and respiratory rate to a level that the person can still carry on a conversation, but can't sing. Examples of these activities include walking briskly, biking on level ground, water exercise, using a stationary bicycle, dancing, gardening, household activities, canoeing, using hand cycles, and using a manual wheelchair.

Exercise intensity may be monitored by using a Rate of Perceived Exertion Scale (RPE) and/or heart rate, detailed on the following tables.

**Table 1: Exercise Intensity Based on Rate of Perceived Exertion and Heart Rate Maximum<sup>27</sup> (\*\* = target zone)**

Intensity Level	RPE (Based on a 0-10 scale)	Heart Rate Maximum %
Light	RPE < 5	50-63%
<b>**Moderate</b>	<b>RPE = 5-6</b>	<b>64-76%</b>
Vigorous	RPE > 7	77-93%

**Table 2: Strengthening Intensity Based on Training Zones<sup>29</sup> (\*\* = target zone)**

Zone Training	Type of Training	Resistance
1-3 repetitions	Muscle power	Very heavy
3-7 repetitions	Muscle strength	Heavy
<b>**8-12 repetitions</b>	<b>Strength and endurance</b>	<b>Moderate</b>
13-25 repetitions	Endurance	Light

Strength training involves using some type of resistance (body weight, weights, weight machines, elastic bands). The different types of resistance training are outlined above.<sup>29</sup> Performing moderate intensity resistance training is recommended for individuals with DM.<sup>30-34</sup>

## Benefits of Exercise

Exercise has general benefits of lowering mortality and morbidity in individuals without DM.<sup>35, 36</sup> Studies have shown reduction of co-morbidities associated with exercise including reduction of falls,<sup>37</sup> decreased anxiety, depression and pain,<sup>38, 39</sup> cognitive and psychosocial benefits of improved cognition<sup>40, 41</sup> and reduced risk of dementia.<sup>42, 43</sup>

Epidemiological evidence has shown that there is a dose response associated with physical activity and feelings of energy.<sup>44</sup>

In neuromuscular diseases (NMD), the general benefits of exercise continue to be a reason to remain active. However, the response to exercise is less known and the method to approach exercise may need to be more creative and individualized to meet different presentation patterns seen in DM. Physical therapists can assist individuals with DM in designing exercise programs that are specific to their needs. Adaptations to exercise for those with NMD mimic that of the sedentary and elderly populations.<sup>36</sup> Individuals with DM should consult with a PT to design an individualized home exercise program and to guide exercise progression for the most benefit.

### **Evidence Supporting Exercise in Individuals with Myotonic Dystrophy**

There is evidence supporting the participation of individuals with DM in moderate intensity exercise without ill effects.<sup>30, 34, 45</sup> In a Cochrane review updated in 2013,<sup>34</sup> the authors examined the safety and efficacy of strength and aerobic training in neuromuscular diseases. They reviewed five randomized controlled trials involving strength and/or aerobic training programs. Based on these studies, the authors concluded that strengthening exercises at a **moderate** intensity did not worsen the disease progression in persons with DM. Exercise studies in disorders like DM are difficult to perform as they are rare diseases and it is difficult to enroll enough patients to carry out a well powered randomized control trial. In another study (not included in the Cochrane review), Orngreen and colleagues studied the benefits of aerobic exercise using bicycle ergometers in individuals with DM1 and concluded that aerobic exercise is safe and improves fitness in individuals with DM1.<sup>45</sup> Cup et al<sup>30</sup> also reviewed the evidence related to exercise in individuals with neuromuscular diseases with expanded criteria than those in the Cochrane reviews. Based on their analysis of the studies, they concluded that

strengthening exercises in combination with aerobic exercises are “likely to be effective.” Last, in a recent scoping review of 21 published studies, Roussel et al. reports positive training effects on patient-reported outcomes, muscle strength, muscles endurance, aerobic capacity and function.<sup>46</sup> Given the type of review that this was, the quality of the methods used in these studies was not analyzed and therefore the interpretation of the findings are limited. Overall, exercise studies in DM support both moderate intensity aerobic and strengthening exercises.

### **Physiology of Exercise**

Individuals with DM1 and DM2 show signs of progressive strength loss. In a nine year study, loss of muscle strength occurred in 30.3% to 43.5% of individuals with DM1 compared to individuals without DM1.<sup>47</sup> Individuals who showed very minor signs of muscle weakness with Muscle Impairment Rating Scale (MIRS) scores of 1 or 2 (no limb weakness) showed an 11.3% to 24.1% decline in strength.<sup>47</sup> Muscle weakness in DM is associated with the CTG repetitions accumulation of toxic CUG RNA leading to atrophy of type I muscle fibers, increased centrally nucleated fibers, fibrosis and fat infiltration of tissue.<sup>48</sup> However, there is a lack of understanding of the mechanism behind muscle weakness in DM1, muscle fiber response and adaptation to exercise. Studies have focused on countering the effects of progressive strength loss and atrophy in DM.

Strength training tends to increase lean body mass, contractile force and muscle power by signaling pathways to activate satellite cells that increase proliferation of contractile proteins such as actin and myosin.<sup>49</sup> Training increased the muscle fiber cross sectional area with no negative change to histopathology.<sup>45, 50</sup> Endurance exercise in mouse models have provided understanding at the cellular level on the possible physiology behind strength gains and motor improvements with exercise including decreased RNA toxicity, alternative mRNA splicing, and MBNL1 loss of function.<sup>51, 52</sup> Aerobic exercise induces physiologic adaptations to the heart, peripheral

circulation, skeletal muscle to improve oxygen delivery, capillary density and vascular conduction to muscles through mitochondrial biogenesis and oxidative enzyme activity.<sup>49</sup> This may be measured by  $VO_2$  max, reduction in fatigue with activity, and ability to perform functional activities.

For individuals with progressive weakness, overwork weakness is a possible side effect of too much exercise. Overwork weakness occurs when already weak muscles exceed the maximal strength capabilities of the muscle fiber.<sup>53</sup> In a randomized controlled trial looking at lower extremity strengthening in Charcot-Marie-Tooth (CMT) and DM, they found no evidence of overwork weakness<sup>54</sup> however, clinicians should monitor this closely as strengthening exercise should not exacerbate muscle weakness. DM, unlike other dystrophinopathies like Duchenne Muscular Dystrophy (DMD), does not have membrane fragility issues that increase the risk of mechanical load injuries thus strength training is safe and recommended.

### **Goals of Exercise for Individuals with Myotonic Dystrophy**

- Maximize range of motion and minimize muscular imbalances.
- Avoid and minimize disuse atrophy.
- Maximize functional abilities. It is important to train to maximize muscle ability to perform functional activities and daily mobility tasks such as walking, climbing stairs and getting out of chairs. Training should be adapted to encompass varying strength throughout the full range of motion required for patient identified functional goals.
- Energy Conservation. Watch for fatigue and overuse; consider that exercise may have a positive impact on fatigue over time.
- Prevent secondary injuries or repetitive injuries by utilizing good biomechanics.

## **Types of Exercise**

### **Range of Motion/Stretching Exercise**

Range of Motion (ROM) exercises are important in maintaining joint function and muscular balance and may play a role in reducing pain caused by muscular imbalance or tightness. One benefit of ROM/stretching is decreased muscle stiffness, because chronically tense muscles have reduced circulation and less nutrition to muscles. Other benefits include reduced post exercise soreness and pain, and improved mechanical function of muscles and joints by improving lubrication to articular cartilage of joints requiring less energy to move through a ROM.

As muscles atrophy resulting in weakness, gravitational pull may limit a person's ability to move a body part through its entire ROM, therefore it may be important to change the position of the body part to minimize the pull of gravity. For example, people may have difficulty raising their arms up in a sitting or standing position, i.e. performing shoulder abduction in an antigravity position, but may have the ability to perform this movement when lying down in a supine position where the effects of gravity are eliminated. Varying the position or assistance needed throughout the ROM is a form of strengthening throughout the ROM.

Individuals may also participate in ROM exercises that are more dynamic in nature. This includes yoga or Pilates based activity that can either be done individually or in a class setting. Education regarding ROM exercise is essential to the management of the symptoms related to the musculoskeletal system.

### **Aerobic Exercise**

Aerobic (cardiovascular) exercise has been found to be safe and may offer benefits for people with myotonic dystrophy.<sup>45, 46</sup> Aerobic exercise increases your heart rate and respiratory rate and is aimed at increasing endurance and fitness. Some examples of aerobic exercise are walking briskly, running, bicycling, swimming or exercising in water, using machines like an elliptical, dancing, or raking leaves. There are



many benefits of aerobic exercise, however, because DM can affect the heart rhythm, it is essential that individuals have a physical, appropriate cardiac evaluations and clearance from their physicians prior to initiating an aerobic exercise program.

### **Resistive Exercise**

Individuals with DM may benefit from a resistive exercise program with studies showing improvements with strength training.<sup>32, 34, 46</sup> Resistance exercise can be accomplished in several ways with resistance provided by gravity, water (in a pool), equipment such as elastic bands, free weights and machines. Yoga and Pilates type of exercises may be recommended as part of a strengthening program, but there are no specific studies that have examined the effects of these specific interventions in patients with DM. It is essential that individuals with DM work with providers who are knowledgeable about their condition, have proper baseline evaluation and appropriate follow-up to monitor and modify the program as necessary.

### **Balance Training**

Balance training improves mobility and coordination and can reduce falls and the fear of falling.<sup>55</sup> Individuals with DM tend to have fall risks similar to that of the geriatric population<sup>56</sup> due to distal limb weakness and decreased reaction time.<sup>13</sup> A comprehensive balance program involves maximizing sensory input, strength and balance strategies. Individualized exercise programs require assessment and treatment from a physical therapist. However, balance training can be incorporated into daily physical activity and exercise programs. Practicing task specific static balance and dynamic balance activities can improve balance for performing daily tasks. One example of a static balance exercise is standing on different surfaces (hard/soft) with eyes open and eyes closed or while turning your head back and forth. Another example is balancing with feet close together, in tandem, or on one foot. These exercises can be performed near a wall or kitchen counter corners for safety. Dynamic balance

exercises are activities that are performed while moving and include walking and talking or performing other mental tasks, walking and carrying objects, or more formal exercise such as yoga and tai chi. In addition to practicing balance activities, strengthening exercises that engage the trunk and hip muscles are important for stability and balance reactions and should be incorporated into an exercise program.

### **Finding Motivation for Participating in Physical Activity and Exercise**

Participation in physical activity and exercise requires motivation. There may be individual factors such as personality, knowledge, and beliefs that are associated with adhering to an exercise program.<sup>57</sup> Burnet et al. suggests that pursuing activities that are of personal interest and fun are important factors to consider when starting a new exercise routine to improve adherence.<sup>58</sup> Recently, the investigators of the OPTIMISTIC study reported that the use of Cognitive Behavioral Therapy (CBT) resulted in increased activity and reduced fatigue in individuals with DM1.<sup>59</sup> Strategies such as making exercise part of a daily routine, engaging friends and family, and monitoring progress can help individuals stay motivated to adhere to exercise. Scheduling activities using calendars and alarms, making “appointments” with friends/family members, physical therapists, personal trainers, exercise classes or other instructors can help add exercise to daily routines. Keeping a log of activities, setting goals, tracking progress, and having a reward-based system can help maintain motivation.

### **Monitoring Exercise**

Monitoring the body’s response to exercise helps determine the level of physiological work during exercise. There are multiple methods to monitoring exercise. In an exercise lab, exercise intensity is directly measured via VO<sub>2</sub> measurement of oxygen utilization. Other means of measuring intensity outside of the clinic include heart rate (HR), rate of perceived exertion (RPE) and talk test. These methods are detailed in Table 3.



**Table 3. Exercise Intensity Measurement Methods**

Method	Method	Equipment
<b>Heart Rate (HR)</b> <i>*cardiac conditions will have modified target HR</i>	<ul style="list-style-type: none"> <li>Maximum HR = 220 – Age</li> <li>Target HR = Max HR x .65-.80 (typically for moderate intensity; but also may be training dependent)</li> <li>Beta blockers may affect HR, making it difficult to monitor HR intensity                             <ul style="list-style-type: none"> <li>Irregular cardiac rhythms may impact HR monitor accuracy</li> </ul> </li> </ul>	<ul style="list-style-type: none"> <li>Heart rate monitor</li> <li>Take your pulse (not ideal because you have to stop activity to measure)</li> </ul>
<b>Rate of Perceived Exertion (RPE)</b>	<ul style="list-style-type: none"> <li>RPE Scale: 0-10 based on perception of effort</li> <li>Effort should be based on:                             <ul style="list-style-type: none"> <li>HR, breathing, muscle fatigue, sweating, discomfort</li> <li>Be honest about how you feel</li> </ul> </li> </ul>	<ul style="list-style-type: none"> <li>Borg Scale<sup>32</sup> (6-20 scale)</li> <li>Omni Scale<sup>33</sup> (0-10 scale)</li> </ul>
<b>Talk Test (TT)</b>	<ul style="list-style-type: none"> <li>Measures “ventilatory threshold” of moderate activity.</li> <li>You should be able to speak 3-5 words comfortably</li> </ul>	<ul style="list-style-type: none"> <li>None</li> </ul>

**Table 4. FITT Based Exercise Calendar**

FITT Principles: Frequency, Intensity, Time, Type													
Day	Exercise	How long?	Reps: sets	How hard am I working: Intensity									
Monday ____/____/____	<input type="checkbox"/> Cardio_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												
Tuesday ____/____/____	<input type="checkbox"/> Strength_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												
Wednesday ____/____/____	<input type="checkbox"/> Cardio_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												
Thursday ____/____/____	<input type="checkbox"/> Cardio_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												
Friday ____/____/____	<input type="checkbox"/> Cardio_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												
Saturday ____/____/____	<input type="checkbox"/> Cardio_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												
Sunday ____/____/____	<input type="checkbox"/> Cardio_____			1	2	3	4	5	6	7	8	9	10
	<input type="checkbox"/> Strength_____												

The American College of Sports Medicine (ACSM) guidelines to monitoring a fitness program is based on the FITT principles: Frequency, Intensity, Time and Type.<sup>35</sup> Other studies also suggest to monitor FITT principles alongside exercise behavior for better adherence to exercise.<sup>58</sup> An example of a FITT based exercise calendar using the OMNI scale for measuring intensity is shown in Table 4.

### **Pain**

A wide variety of methods have been used in the treatment of pain in individuals with DM. The use of non-steroidal anti-inflammatory medications or acetaminophen, exercise (strengthening and ROM), and heat are the most common therapies used to manage pain.<sup>19, 22</sup> Individuals should consult their physician for recommendations regarding the use of medications for pain relief.

### **Fatigue**

Recently, researchers from the OPTIMISTIC study<sup>59</sup> reported that Cognitive Behavioral Therapy increased activity and participation and reduced fatigue. Further studies are needed to better understand the factors contributing to symptoms of fatigue.

### **Orthotics**

Lower extremity weakness can affect a person's ability to walk safely, especially on uneven surfaces. Ankle dorsiflexion weakness often leads to foot drop and decreased foot clearance during the swing phase of gait. Gait abnormalities typically seen are steppage gait (lifting their knees higher to help the foot clear the ground), foot slap, and lack of heel toe action. These compensatory gait patterns may result in increased energy use for walking. The use of ankle-foot-orthotics (AFO) can help to correct the foot drop, however, care must be taken in prescribing an AFO. Other reasons for using lower extremity orthotics include plantarflexion weakness during stance phase to provide stability, and, nighttime stretching of the Achilles tendon to minimize muscle imbalances at the ankle.

Several factors may play a role in the effectiveness of orthotic use in the lower extremities. The additional weight that may be

added to the lower extremity by a brace can significantly alter the person's ability to ambulate, hence it is important that the orthotics are made of the lightest materials available. It is important to consider the person's ability to don and doff the orthotic devices, especially in the presence of hand weakness and decreased hand function. Orthotic fit is often difficult because people with DM have muscular wasting and bony landmarks, thus are more susceptible to skin irritation and breakdown. Comfort and satisfaction are important in promoting the use of the prescribed device. Compliance suffers if the prescribed orthotic device is uncomfortable or too difficult for the client to get on and off independently. Patients who are prescribed orthotics should see a physical therapist for gait training after receiving new braces. Furthermore, there has been very limited research on the effect of orthotic use on energy expenditure during walking, thus this area needs further investigation to prescribe appropriate orthotics to this patient population.

In cases where the neck muscles are also affected, neck braces may also be beneficial. Many of these braces are off the shelf and can be fit by an orthotist. Consultation with an occupational or physical therapist is highly recommended prior to purchase of these braces so that they fit individualized needs.

### **Assistive Devices/Adaptive Equipment**

Individuals with myotonic dystrophy may be at a higher risk for falls.<sup>56</sup> Decreased visual acuity, lower extremity weakness and depression can play a role in increasing the risk for stumbles and falls.<sup>12</sup> The use of canes, walkers, wheelchairs, and powered mobility devices can be used to allow a person to continue to be safe and independent in mobility. The use of mobility devices helps individuals continue with their societal roles and responsibilities, while maintaining and improving quality of life. Adaptive equipment such as long handled sponges, foam buildups on silverware and pens, and button hooks can make bathing and dressing easier and allow individuals to be more independent in caring for themselves. When assessing for adaptive equipment, a referral to an Occupational Therapist (OT) may also be beneficial.

## Children with Myotonic Dystrophy

Even though DM1 is considered to be the most common of the adult muscular dystrophies, congenital (present at birth) and childhood presentations are also recognized.<sup>60, 61</sup> Congenital myotonic dystrophy (CDM) tends to be more severe than the childhood form and is often associated with hypotonia, respiratory insufficiency and feeding problems.<sup>3, 60, 61</sup> When symptoms arise during the childhood years, the progression is similar to that in adult onset DM, however since the symptoms start earlier, they may be more severe later in life.<sup>62</sup> Despite the early onset of muscle weakness, CDM may represent a developmental disorder where children show some improvement in strength and functional abilities yet with severe motor delays.<sup>63</sup> This muscle maturation with age may compensate for some of the underlying pathophysiology of the disease.<sup>64</sup>

Cognitive impairment is also present in the pediatric phenotypes, with higher severity in the congenital form.<sup>3, 61</sup> The need for physical therapy services can be highly variable and individualized based on the type and severity of symptoms. Initially, therapy services may focus on motor skill attainment with future services being directed towards areas similar to that of adults with DM including exercise recommendations, orthotics, and adaptive equipment.

## Early Intervention and Facilitation of Motor Skills

The child will be developing motor skills and should be referred to early intervention services for gross and fine motor development. Speech delays may require more intensive hands-on therapy to facilitate motor development and attainment of motor milestones. Hands on physical therapy services can be provided in several different settings and formats ranging from traditional home or clinic-based activities, to aquatic therapy, to equine movement called hippotherapy. Traditional home or clinic-based services utilize the child's natural environment, including daycare, preschool, or playground to address specific developmental or functional needs. Parent and caregiver training is essential

in pediatric therapy to ensure that the handling skills and facilitation of movement is carried across to performance at home during activities of daily living.

## Aquatic Therapy

Aquatic therapy uses the physical properties of water to perform exercise. The buoyancy provides support and facilitates movements. The viscosity or resistive properties of the water also allow for strengthening of the postural and limb muscles. These qualities of the aquatic environment have been shown to be beneficial in improving functional mobility of children with motor limitations.<sup>65, 66</sup>

## Hippotherapy

Hippotherapy is a treatment strategy in which the movement of a horse is used to address impairments and functional limitations in people with neuromuscular dysfunction. Hippotherapy has been shown to improve upright posture with trunk stabilization, pelvic mobility and weight shifting of the body to positively impact gross motor function and walking ability in children with developmental delays.<sup>67-69</sup>

## Summary

There are no reports of any studies that have looked specifically at using the above interventions in children with myotonic dystrophy. It is difficult to document the specific impact of these interventions versus the natural gains that occur with development since there are very few appropriately controlled longitudinal case studies reported in the literature. Further research is needed to determine the appropriate type, frequency, intensity, and duration of physical therapy services for children with DM.

Currently, the frequency and intensity of the hands-on services vary depending on the individual child's needs. These services may be followed by episodic care where the physical therapist will play a more consultative-educator role, monitoring the child's development and working with the family and school to set up a home-based program of daily activities and



exercises to maximize the child's functional abilities. Within the school system, the physical therapist will work with the school team, including classroom teachers, gym teachers, school nurse, and counselors to educate regarding the condition and the appropriate activities and supports within the school environment to assure safety, mobility and maximize the learning opportunities.

## CONCLUSION

In these guidelines, we have attempted to meet the needs of physical therapists who may rarely encounter patients with myotonic dystrophy, thus may not have much knowledge of the condition. We hope that the information and the references we have provided will help physical therapists get started in meeting the needs of their patients. For individuals with DM who may be reading these guidelines, we hope that we have given you useful information about the role of physical therapists in your care, so that you are better prepared to partner with them to improve function and independence. We appreciate your feedback about how we might make these guidelines more responsive to your needs. We appreciate the opportunity and support provided by the Myotonic Dystrophy Foundation to share this information with you. The authors want to recognize their mentor Shree Pandya, PT, DPT, MS for her contributions to previous versions of this guide.

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Myotonic's mission is to enhance the quality of life of people living with myotonic dystrophy and accelerate research focused on treatments and a cure.



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