MYOTONIC DYSTROPHY FOUNDATION

ROLE OF PHYSICAL THERAPY IN THE ASSESSMENT AND MANAGEMENT OF INDIVIDUALS WITH MYOTONIC DYSTROPHY

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Physical therapists are healthcare professionals who hold a post-baccalaureate graduate degree (MPT, DPT) from a 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 care, and schools.[1] Most individuals with myotonic dystrophy (DM) will probably first encounter a physical therapist in the multidisciplinary clinic where they receive care for their muscular dystrophy 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 as necessary. Some common areas that will be addressed by physical therapists are related to exercise/activities, pain and/or fatigue management, orthotics/braces and assistive/adaptive equipment. The goals of physical therapy management are to maximize functional ability, delay secondary complications 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, which means 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 and endocrine systems. Currently 2 variants of DM are recognized – DM1 which arises from a defect on chromosome 19 and DM2 which results from a defect on chromosome 3.[2] DM1 was first described in the early 1900's and hence is a much better studied entity while DM2 was only described in the past decade and hence there is a lot to learn regarding this phenotype.[3] 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 and thus four clinical phenotypes are described in the literature. Congenital phenotypes have not been described in DM2 yet and most patients present in adulthood. Weakness and 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 - weakness, wasting and functional problems - are very often the concerns that lead individuals to seek attention and help from physical therapists. However, DM is a multi systemic disorder and hence it is essential to understand all the systemic complaints and help manage the muscle related symptoms in the overall context of concerns for an individual.[2, 3, 4] Congenital and childhood onset DM1 have unique features, and therefore, we have chosen to address the physical therapy management of these conditions separately later is this section.

PHYSICAL THERAPY ASSESSMENT

During an initial evaluation a physical therapist will obtain a detailed history of the 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 stated before, myotonic dystrophy is a systemic condition. It is therefore important for the physical therapist to perform a systems review according to the Guide to Physical Therapy[5] 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.[2, 3, 4.] Symptoms include, somnolence, apathy, specific personality traits, deficit in executive functions, depression and fatigue. These cognitive deficits may impact a person's ability to comply 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 (masseter and temporalis) muscles, neck muscles (sternocleidomastoids), long finger flexors of the hand and ankle dorsiflexors and/or plantarflexors[4]. 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 and also slowly progressive, beginning in the 'mid-adult' life [2]. It is critical that physical therapists are knowledgeable in 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 expensive systems such as a 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 as well as adult populations. [6-10]

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 creates an embarrassing social situation. Complaints of myotonia are also reported in the jaw and tongue leading to difficulties with speech, swallowing and chewing [2, 3, 4]. Myotonia in the leg muscles may lead to difficulty with movements like climbing stairs, running 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 has been managed with medications such as Mexilitene.[11]

DM1 is a slowly progressive disease and as strength decreases, individuals may become adept in substituting less affected muscles to perform movement. Hence it is important to assess simple functional activities, including the ability to get up from a chair, ambulate and climb stairs. These functional tasks can also be timed and used as outcome measures to document benefits of interventions or to monitor the progression of the disease. 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.

The cardiovascular system can be compromised by the presence of cardiac arrhythmias and conduction defects as well as involvement of the cardiac muscle itself [2]. Insufficiency of the respiratory system may be a result of both myotonia and weakness in the muscles that control respiration.[2] Respiratory muscle involvement often leads to a reduced vital capacity later in the disease. Individuals with DM1who have reduced respiratory function are often at more risk for pulmonary complications such as pneumonia[4]. When making exercise recommendations for a home program, it is essential to educate individuals about how to monitor their cardio respiratory responses with simple tools like pulse monitors, Borg scale, etc. It is essential that individuals report their responses to exercise to the person overseeing and/or monitoring the home program. Depending on the progression of their disease, individuals with myotonic dystrophy may have limited exercise tolerance and will need to be monitored carefully.

The integumentary system is not usually involved as the sensory system is spared in myotonic dystrophy. However, if poor mobility is demonstrated and bony prominences are exposed secondary to muscle wasting, the integumentary system may require attention.

Pain and Fatigue are common complaints among individuals with DM1 and DM2 [12-16]. In a study by Jensen et al.[14], complaints of pain were reported most commonly in the low back and legs. More than 60% of patients with neuromuscular disorders complain of fatigue. Fatigue can have a major impact on the employment status of patients with DM. Therefore, pain and fatigue should be assessed and addressed in the treatment plan as necessary.

Lastly, it should also be mentioned that many of these individuals have gastrointestinal manifestations that may be present anywhere along the digestive tract. Symptoms reported span the spectrum of dysphasia and heartburn to abdominal pain and changes in bowel function.[17, 18] Involvement of the GI system may be very disabling to the individual and again, may impact the person's ability to participate in exercise programs.

PHYSICAL THERAPY MANAGEMENT

Exercise

Individuals with myotonic dystrophy often have questions about exercise. Exercise, including range of motion, strengthening and cardiovascular (aerobic) exercise, is important for the management of the musculoskeletal and cardiorespiratory manifestations of myotonic dystrophy. Range of motion exercises are important in maintaining joint function and muscular balance and may play a role in reducing pain that is caused by muscular imbalance or tightness. As muscles atrophy resulting in weakness, gravitational pull may limit a person's ability to move a body part through its entire range of motion and 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 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 gravity is eliminated. Individuals may also participate in range of motion exercises that are more dynamic in nature. This includes Yoga and Pilates based activities that can either be done individually or in a class setting. Education regarding range of motion exercise is essential to the management of the symptoms related to the musculoskeletal system.

Weakness occurs as part of the disease process; however, weakness may also develop secondary to disuse. Strengthening exercises may help to minimize the disuse weakness; but there is also a concern that too much exercise or inappropriate exercise may hasten disease progression, and hence finding the right balance for each individual is important. The evidence available regarding the role of exercise in myotonic dystrophy is limited. In a Cochrane review published in 2010.[19], the authors examined the safety and efficacy of strength and aerobic training in neuromuscular diseases. They identified a total of 36 studies; however, there were only three randomized controlled trials that fulfilled their inclusion criteria. Based on these studies the authors concluded that strengthening exercises at a moderate intensity did not worsen the disease progression in persons with myotonic dystrophy.[19, 20.] Many of the studies involving individuals with myotonic dystrophy were excluded from the review because they lacked randomization. Many of these studies also grouped different neuromuscular diseases together, making it difficult to draw conclusions about the individual's response to exercise in a specific disease like DM. Disorders like DM are difficult to study as they are rare diseases and it is difficult to enroll enough patients to carry out a well powered randomized control trial. Other problems cited with the reviewed studies included lack of detailed descriptions of the exact exercise protocols used and short durations of the exercise trials. Orngreen and colleagues studied the benefits of aerobic exercise using bicycle ergo meters in patients with DM1 and concluded that aerobic exercise is safe and improves fitness in patients with DM1. [21]. Cup et al[22.] chose to look at 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 the evidence suggests that strengthening exercises in combination with aerobic exercises are "likely to be effective". Given the evidence from the 2 major reviews that exercise may be effective and that moderate exercise does not worsen disease progression, some general recommendations regarding exercise can be made to guide clinicians and individuals with myotonic dystrophy.

Depending on the activity level of the individuals, they may benefit from a strengthening program. Individuals who lead an active lifestyle may not have much disuse weakness, and further activity may be fatiguing to them. However, others who lead a more sedentary lifestyle may benefit from a

strengthening program. Strengthening exercise can be accomplished in several ways with resistance provided by gravity, water – in a pool - or equipment such as elastic bands, free weights and machines. Yoga and Pilates types of exercises may also be recommended as part of a strengthening program, but there are no studies reported that have examined the effects of these specific interventions in patients with DM. It is essential that individuals with myotonic dystrophy work with providers knowledgeable about their condition; have proper baseline evaluation and appropriate follow-up to monitor and modify the program as necessary.

Cardiovascular exercise performed at a low to moderate intensity has been found to be safe in people with myotonic dystrophy. Cup et al.[22] also concluded that there was "indication of effectiveness" for aerobic exercises in individuals with muscle disorders. However, because of the cardiac involvement that can occur in persons with myotonic dystrophy, it is essential that individuals have a physical, appropriate cardiac evaluations and clearance from their primary care physicians prior to initiating an aerobic exercise program.

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. [23.] They recommend that children, adolescents, adults (ages 18-64) and older adults follow the appropriate guidelines to the best of their ability. Individuals with chronic conditions perform as much activity and/or exercise as their condition allows. These include about 2 hours and 30 minutes a week of moderate intensity exercise. Aerobic exercise should be performed in episodes of at least 10 minutes preferably spread throughout the week. Muscle strengthening activities that involve all major muscle groups should be performed at least 2-3 days a week.

Examples of moderate intensity activities include – walking briskly, biking on level ground or on a stationary bicycle, ballroom and line dancing, general gardening, household activities, canoeing, using hand cycles, using a manual wheelchair and water aerobics. Moderate exercises are activities that you can perform while still continuing a conversation –without having to stop to catch your breath.

Pain

A wide variety of methods have been used in the treatment of pain in individuals with myotonic dystrophy. 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. [16] Individuals should consult their physician for recommendations regarding the use of medication for pain relief.

Fatigue

Currently there are no reports of specific interventions and their impact on management of fatigue in patients with DM. Interventions may need to be individualized based on specific factors contributing to the complaint 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 a foot drop and decreased foot clearance during the swing phase of gait. Some individuals may compensate for the ankle dorsiflexion weakness by using a steppage gait pattern, i.e. lifting their knees higher to help the foot clear the ground. The use of ankle-foot-orthotics can help to correct the foot drop; however, care must be taken in prescribing an AFO. 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, and hence it is important that the orthotics are made of the lightest materials available. It is also 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 myotonic dystrophy have muscular wasting, and bony landmarks often become more prominent and 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. Furthermore, there has been very limited research on the effect of orthotic use on energy expenditure during walking and is definitely an area that needs further investigation to prescribe appropriate orthotics to this patient population.[24.] 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.

Assistive Devices/Adaptive Equipment

Individuals with myotonic dystrophy are at a higher risk for falls. Decreased visual acuity, lower extremity weakness and depression can play a role in increasing the risk for stumbles and falls. [25.] 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. Adaptive equipment, such as long handled sponges, foam buildups on silverware and pens, and button hooks can make performing 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 may also be beneficial.

Children with Myotonic Dystrophy

Even though DM1 is considered the most common of the adult muscular dystrophies, congenital (present at birth) and childhood presentations are recognized. Congenital myotonic dystrophy tends to be more severe than the childhood form and is often associated with hypotonia, respiratory insufficiency and feeding problems.[4, 26.] When symptoms arise during the childhood years, the progression is similar to that in adult onset myotonic dystrophy, however since the symptoms start earlier, they may be more severe later in life.[2.] Cognitive impairment is also present in these phenotypes, with the involvement being more severe in the congenital form.[27, 28.] The need for physical therapy services can be highly variable and individualized based on the type and severity of the symptoms. The areas addressed by physical therapists will be the same as in the adult population, including recommendations regarding exercise, orthotics, and adaptive equipment. Additionally, the child will be developing motor skills, and there may be a need for short episodes of 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 including home, daycare, school, playground and clinic depending on the goals of the therapy session. In addition to typical interventions such as range of motion and strengthening exercises,

practice of activities of daily living, motor skill development, therapy may include aquatic therapy or hippotherapy - the utilization of equine movement.

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 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 mobility limitations.[29, 30] Hippotherapy is another 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, and balance therefore positively impacting gross motor function and walking ability in children with developmental delay.[31, 32]

There are no reports of any studies that have looked specifically at using aquatherapy or hippotherapy 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. Hence further research is needed to determine the appropriate type, frequency, intensity, and duration of physical therapy services in children with myotonic dystrophy.

Currently, the frequency and intensity of the hands on services vary depending on the individual child's needs. These may be followed by more limited episodes where the physical therapist will play a more consultative role, monitoring the child's development and working with the family 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 – classroom teachers, gym teachers, school nurse, and counselors etc educating them regarding the condition and the appropriate activities and supports within the school environment to assure safety, mobility and maximize the learning opportunities.

In this section we have attempted to meet the needs of therapists who may rarely encounter patients with DM and hence may not have much knowledge of the condition. We hope that the information and the references we have provided will help them get started in meeting the needs of their patients. For individuals with myotonic dystrophy who may be reading this section – we hope we have given you enough information about the role of physical therapists in your care, so that you are better prepared to partner with them in meeting your needs. We would appreciate any feedback from all readers about how we might make this section more responsive to their needs. We appreciate the opportunity and support provided by the Myotonic Dystrophy Foundation to share this information with you.

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