Case Report / Olgu Sunumu

Myotonic Dystrophy

Miyotonik Distrofi

Zeliha Ünlü

Celal Bayar University, Department of Physical Medicine and Rehabilitation, Manisa, Turkey

Abstract

Myotonic dystrophy (MD) is a rare, progressive muscle disease, characterized by autosomal dominant heredity and multisystem involvement. The muscle involvement's being progressive in MD patients leads to negative effects such as dependency of the patients in their daily activities. For this reason, medical treatment and rehabilitation of these patients are very important. We considered 3 patients with MD diagnosis in this series of cases. We made pre-treatment and post-treatment evaluations of 3 patients who were hospitalized in our clinic by giving them a 4 week conventional exercise program and functional electrical stimulation (FES) treatment. The rehabilitation program we applied to the patients contained joint's range of motion exercises, breathing exercises, standing training, gait training, balance coordination exercises, and FES application to ankle and wrist dorsiflexors. We achieved positive changes in the clinical findings that we documented with pre-treatment and post-treatment assessment values.

Keywords
Myotonic dystrophy, rehabilitation, exercise

Öz

Miyotonik distrofi (MD) seyrek görülen, otozomal dominant kalıtım ve multisistemik tutulumla seyreden ilerleyici bir kas hastalığıdır. MD hastalarında kas tutulumunun ilerleyici olması hastaların günlük yaşam aktivitelerinde bağımlılık gibi olumsuz etkiler doğurmaktadır. Bu yüzden bu hastaların medikal tedavi ve rehabilitasyonu oldukça önemlidir. Bu olgu serisinde MD tanısı olan 3 olgu gözden geçirildi. Kliniğimizde yatarak tedavi gören 3 MD hastasına 4 hafta geleneksel egzersiz programı ve fonksiyonel elektrik stimulasyonu (FES) tedavisi verildi, hastalar tedavi öncesi ve sonrası değerlendirildi. Hastalara uygulanan rehabilitasyon programı; eklem hareket açıklığı egzersizleri, solunum egzersizleri, ayakta durma eğitimi, yürüme eğitimi, denge koordinasyon egzersizleri, ayak bileği ve el bileği dorsifleksörlerine FES uygulamasını içeriyordu. Hastaların tedavi öncesi ve tedavi sonrası değerlendirme ölçekleriyle dökümente ettiğimiz klinik bulgularında olumlu değişiklikler bulundu.

Anahtar Kelimeler
Miyotonik distrofi, rehabilitasyon, egzersiz

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Address for Correspondence/Yazışma Adresi:
Zeliha Ünlü MD,
Celal Bayar University, Department of Physical Medicine and Rehabilitation, Manisa, Turkey
Phone : +90 236 236 03 30
E-mail : zelihaunlu@yahoo.com
ORCID ID: orcid.org/0000-0003-2270-0414

Introduction

Myotonic dystrophy (MD) is an autosomal dominant hereditary muscle disease which presenting with progressive muscle weakness. MD is a rare disease with an incidence of 1/8000 (1). This condition is caused by an expanded cytosine-thymine-guanine trinucleotide repeat in the DMPK-gene on chromosome 19q (2,3). MD is not limited to muscle involvement only, it presents with multisystemic symptoms

(4). Along with musculoskeletal system symptoms, such symptoms as cataracts, cardiac pathologies, insulin resistance, alopecia, constipation, interfertility may be present. Progressive muscle weakness, atrophy and myotonia are neuromuscular symptoms of this disease (5). It has 3 sub-groups including myotonic dystrophy-1 (Steinert's disease), MD-2 (proximal myotonic myopathy) and proximal myotonic dystrophy. MD-1 is the most common in adults (6).

The typical involvement pattern in this disease is that it starts with weakness of distal muscles initially and in later stages weakness of proximal muscles is included. Presence of myotonia, as well as muscle weakness, should be considered in these patients. When muscle strength decreases, it also becomes difficult to determine clinical myotonia. Having difficulty in releasing a grasp after making a fist, difficulty in opening eyelids after closing them tightly, occurrence of muscle tightening as a response to hitting muscle with a reflex hammer are clinical symptoms of myotonia.

Although there is no sufficient evidence for the effectiveness of drugs in the treatment of this disease, antidepressants, benzodiazepines, calcium antagonists, taurine (essential amino acids) and prednisone based drugs are among the drugs used (7). Since medical treatment is limited, rehabilitation programs based on exercise are very important in terms of preventing progression of the disease.

Studies conducted with MD patients more often concentrated on the effects of aerobic exercises on progression of the disease and hand skills. In some studies, it was reported that moderate-intensity exercises are not harmful, but their benefit is questionable (8). In this article, it was intended to review MD rehabilitation in the light of recent literature through 3 patients who receive in-patient treatment. The literature concerning MD rehabilitation is limited. It is rather focused on training of subtle skills and aerobic exercise.

Case Report

In this study, we made pre-treatment and post-treatment evaluations of 3 patients who were hospitalized in our clinic by giving them a 4 week conventional exercise program and functional electrical stimulation (FES) treatment. The rehabilitation program we applied to the patients contained joint's range of motion (ROM) exercises, breathing exercises, standing training, gait training, balance coordination exercises, and FES application to ankle and wrist dorsiflexors and moderate-intensity lower extremity and upper extremity strengthening exercises for 2 patients. Strengthening exercises were not given to patient 1 because creatine kinase (CK) values were above normal.

Patient 1 (N.Y.): Forty-eight-year old female patient diagnosed with MD 20 years ago. She could be mobilized with electric-powered wheelchair. She used unarticulated, solid ankle foot orthosis (AFO). She received outpatient treatment in a physical therapy and rehabilitation center one year ago. She had no family history. In her physical examination findings; she couldn't walk, she couldn't stand. Movements in bed were present; she couldn't take sitting position from lying position.

PHASE: It was stage 1. (functional ambulation scale stage 1: physical aid dependent, continuous manual support of a person is required for the patient's ambulation).

Manual muscle strength examination: shoulder flexors-abductors: 4/5, biceps: 3-3+/5, flexor carpi ulnaris - flexor carpi radialis (FCU-FCR): 1/5, finger flexors: 2+/5, hip flexors on the left: 3-/5, on the right: 3+/5, hamstrings: 3+/5, ankle dorsiflexors: 1/5, plantar flexors: 1/5. ROM (joint's ROM) limitations; wrist and ankle active-passive ROM was limited. Deep tendon reflexes (DTR): hypoactive was detected in 4 extremities. There were no pathological reflexes.

Patient 2 (M.Ç.): Fifty-eight-year old patient diagnosed with myotonic dystrophy 20 years ago, walk without support was unstable and short distant. Standing, taking sitting position from lying position was present.

PHASE: Stage 2 (functional ambulation scale stage 2: physical aid dependent, manual support was to assist balance and coordination). The patient hadn't received a physical therapy program nor had any orthesis or AFO to support walking. It was learned that in the family history, daughter of her uncle, her sister and daughter (3rd patient: B.Ç) had the same disease. In her medical history; there was bilateral operated cataracts, a history of arrhythmia and a history of thyroid nodules existed.

In the physical examination; manual muscle strength examination: shoulder flexors-abductors: 4/5, biceps: 4/5, FCU-FCR: 3+/5, finger flexors: 3-/5,

hip flexors: 4/5, hamstrings: 5/5, ankle dorsiflexors on the left: 3+/5, on the right: 3-/5 plantar flexors on the left: 3+/5, on the right: 3-/5. ROM (joint's ROM) limitations; wrist and ankle active ROM was limited, passive was natural. DTR; hypoactive was detected on 4 extremities. There were no pathological reflexes.

Patient 3 (B.Ç): The patient, daughter of the patient 2, diagnosed with MD could walk without support and go up and down the stairs.

PHASE: It was stage 4-5. (functional ambulation scale stage 4-5: patient can walk on flat ground without support, and may need support in inclined surfaces such as stairs). In the physical examination; manual muscle strength examination; wrist extensor-flexors: 4+/5, ankle dorsiflexors/plantar flexors: 4+/5, other muscles: 5/5. No ROM limitation was detected. Active passive electro-hydrostatic actuator was natural. DTR normoactive and there were no pathologic reflexes.

In this study, we made evaluations of patients with examinations and measurements before and after 4-week rehabilitation program. The conducted evaluation forms are as follows:

1) Daily living activities assessment (DLA), health assessment questionnaire; this is a test where patients' daily activities are examined in 8 categories such as dressing, general self-care, eating, sitting standing etc. Evaluation is made as follows: 0 (I do not have any difficulty), 1 (I have a little difficulty), 2 (I have a lot of difficulty) and 3 (I can't perform it). Higher the score in the survey means worse the health situation (9).

2) Posture analysis; it is a measure where head, chest, shoulder, scapula, spine foot posture and leg length are assessed.

3) Gait analysis [functional ambulation classification (FAC)]; patients' walking levels are categorized in 6 groups. (PHASE Level 0: non-functional ambulation, level 5, independent ambulatory) (10).

4) Manual muscle strength assessment; muscle strength is rated from 0 to 5 (muscle strength 0; no movement, muscle strength 5; normal) (11).

5) Balance - coordination tests; get up and go test, functional reach test and romberg test (12).

6) Hand skills test (bimanual fine motor function); both hand functions are rated on 5 levels (13).

7) Goniometric ROM measurement.

According to the results of the evaluation forms obtained before and after the treatment of the patients, we observed improvement in DLA test, FAC, manual muscle strength assessment and goniometric ROM measurements. We did not detect a difference in hand skills test, posture analysis and balance - coordination tests (except patient 2). In patient 2, there was improvement in 'get up and go test' and functional reach test among balance - coordination tests. Patients' pre-treatment and post-treatment DLA and FAC values are stated in Table 1.

Improved muscle strength values of patient 1 and patient 2, whose muscle strength measurements are stated in the case report section in detail, is shown in Table 2. For Patient 3, left big toe flexors improved from 4 /5 to 5/5, other muscle strength measurements were assessed as 5/5.

Those who showed improvement in joint's range of movement measurements are shown in Table 3. In Patient 3, no joint movement limitation was detected pre-treatment or post-treatment. We did not find any difference in patients' other joint range of movement.

Discussion

Medically, rehabilitationally and socially, a multidisciplinary approach is needed to reduce morbidity and mortality rates in MD patients (14). There is a limited literature concerning implementation of rehabilitation programs in MD and their results. There is not sufficient information about rehabilitation protocol to be applied to these patients. Therefore, the rehabilitation practices applied to these patients are focused on ROM exercises, strengthening exercises, aerobic exercises and relation techniques, which underlie conventional

Table 1. Pre-treatment and post-treatment evaluation form results

Patients	HAQ - PreT	HAQ - PostT	FAC - PreT	FAC - PostT
Patient 1	46	41	Stage 0	Stage I 1
Patient 2	31	29	Stage 2-3	Stage 3
Patient 3	6	4	Stage 5	Stage 5

PreT: Pre-treatment, PostT: Post-treatment, HAQ: Health Assessment Questionnaire, FAC: Functional Ambulation Classification

rehabilitation approaches. Low-to-moderate- intensity strength and aerobic exercise training, and an active lifestyle are have beneficial evidences in patients with a slowly progressive neuromuscular disease, such as MD. A hand isometric exercise program applied to 35 MD patients for 12 weeks achieved positive outcomes (15). Studies indicate that there is insufficient evidence whether or not strengthening exercises and aerobic exercise rehabilitation programs in muscle diseases (16). Kierkegaard et al. (17) evaluated a comprehensive group exercise programme supported by music (Friskis&Svettis Open Doors Programme) in patients with MD for 14 weeks. They were not revealed evidently beneficial or harmful effects in intention-to-treat programme. The harmful effects are an important manner when the patient has a progressive disease. They were not detected gross improvements in the outcome measures but a majority of participants in the exercise group felt more subjective positive effects and reported increasing in fitness, strength, flexibility and excessive daytime sleepiness levels.

Due to lack of information and protocol in these patients, we also applied conventional methods to our patients. Considering CK level in this implementation, we avoided overloading during muscle strengthening as in patient 1. We applied moderate-intensity strengthening program to patient 2 and patient 3. At the end of 4-week rehabilitation program, we observed positive developments in our patients as shown in the tables. These observations were documented with measurements. One of the most significant factors for the increased fall frequency in patients with MD is foot drop due to ankle dorsiflexor weakness. Also drop foot is considered a factor decreasing quality of life in these patients. FES might be suggested a safe and effective tool to improve muscle function. We applied FES to stimulate ankle dorsiflexors during one month. Tibialis anterior muscle strength increased all of the cases at the end of the treatment. A pilot study

Table 2. Pre-treatment and post-treatment, comparative muscle strength values of patient 1 and patient 2

Manual muscle strength measurement	Patient 1		Patient 2	
	PreT	PostT	PreT	PostT
Hand finger flexion	2+/5	3/5	3/5	3/5
Thumb flexion	2+/5	3/5	3/5	3/5
Ankle plantar flexion (M. gastrocnemius - M. soleus)	1/5	3/5	3/5	4/5
Ankle dorsiflexion	1/5	1/5	3/5	4/5
Ankle inversion (M. tibialis anterior and posterior)	1/5	2/5	3/5	4/5
Ankle aversion (M. peroneus brevis and longus)	1/5	2/5	3/5	4/5
Toe flexion	3/5	3/5	2/5	3/5

PreT: Pre-treatment, PostT: Post-treatment,

Table 3. Goniometric measurement of joint's range of movement of patient 1 and patient 2

	Patient 1				Patient 2			
	Active		Passive		Active		Passive	
EHA goniometric measurement	PreT	PostT	PreT	PostT	PreT	PostT	PreT	PostT
Wrist extension	5	10	50	70	50	55	60	60
Wrist flexion	0	0	0	0	60	60	90	90
Ankle plantar flexion	5	5	45	45	30	35	45	45
Ankle dorsiflexion	0	0	0	0	10	15	20	25

EHA: Electro-hydrostatic actuator, PreT: Pre-treatment, PostT: Post-treatment

showed that FES lower extremity training (extensor and flexor muscles of the knee as well as ankle dorsiflexor and plantarflexor muscles) has a greater effect on improving muscle strength and endurance when compared with conventional training (18). Recently, improvement of mild tibialis anterior muscle weakness was reported the useful effect of electrical stimulation in 4 patients with MD (19).

Although it is our limitation that we could not provide rehabilitation programs or protocols for these patients due to small number of patients, our observations will constitute preliminary information for wider series of studies to be conducted in the future. MD is a rather neglected disease and new treatment approaches are needed to improve clinical practice in the management of these patients. In this respect, we thing our observations will contribute to the literature.

Since MD might be familial, genetic counseling and guidance in these patients are also important. Patient 2 (M.Ç.) and patient 3 (B.Ç.) are noteworthy in this regard. Clinical follow-up of our patients currently continues.

Ethics

Informed Consent: Patients were informed and they consented to the study.

Peer-review: Internally peer-reviewed.

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