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Physiotherapy and Rehabilitation in Dysferlinopathy

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Abstract

Dysferlinopathy is autosomal recessive neuromuscular diseases resulting from genetic dysferlin deficiency, a protein involved muscle repair. The aim of this study is to present information about evaluation, treatment program of patients with dysferlinopathy. 27-year-old female patient was treated 3 days a week for 7 months. Before and after treatment; it has been assessed that muscle strength with 0-5 Medical Research Council Scale, physical performance with Timed Physical Performance Test, fall worries with Falls Efficacy Scale International; number of falls, body mass index recorded. Kinesiotape, isotonic-isometric-resistant exercises, PNF were applied to increase muscle strength; ear acupuncture was used for weight control. Timed performance durations, worries of falling, the number of falls and body mass index decreased significantly. Compared to pre-treatment muscle strength increased in all muscular groups included. It has been observed that isotonic, isometric, PNF exercises, kinesiotape application increase muscle strength, physical performance; decrease the number of falls, falls worries.

Keywords: Dysferlinopathy; Muscle Strength; Physiotherapy; Pathogenesis

Introduction

Dysferlinopathy is autosomal recessive neuromuscular diseases caused by genetic dysferlin deficiency [1]. The incidence is estimated to be 1 in 100.000-200.000 [1]. Dysferlin known to be present in skeletal, cardiac muscle cells; monocytes, macrophages, plays role in membrane transfer, fusion, repair. Dysferlin loss reduces adhesion, increases cell migration and phagocytosis [2]. When human and mouse are examined, muscles inflammation that cause skeletal dysfunction is characteristic of dysferlinopathy. According to other inflammatory diseases, macrophages accumulate more in the muscles [3]. Fat infiltration was detected in the thigh, leg, arm, forearm, paravertebral and trunk muscles in relation to disease duration, muscle atrophy and decreased motor function [4]. Serum Creatine Kinase-(CK) levels are usually elevated in the early asymptomatic stage (10-100 times normal values) [5].

Principally, Limb Girdle Muscular Dystrophy-(LGMD) Type 2B on the proximal lower extremity, Miyoshi Myopathy-(MM) that holds the gastrocnemius muscle especially in young adulthood, hyper CKemia, distal anterior compartment myopathy, proximodistal myopathy, pseudo-metabolic myopathy, congenital onset myopathy, scapuloperoneal muscular dystrophy diagnosed in only a few cases as the types are defined in different phenotypic manifestations [6]. LGMD2B is usually slow progressive and begins with myalgia. The patients may be dependent on wheelchair within 10-20 years after the disease onset. Usually, cardiac or respiratory involvement is not seen. The shoulder girdle, upper extremity muscles are usually affected less. The face, neck and hand muscles are generally not affected [7]. Dysferlinopathy severity varies from high CK levels to exercise intolerance to severe functional disability [8].

While dysferlinopathy affects muscle strength, cardiac, respiratory functions, walking and Daily Life Activities-(DLA) negatively, according to our knowledge, there is no study about the dysferlinopathy rehabilitation in the world. The aim of this study is to draw a path map in the physiotherapy of PwD-(Patient with dysferlinopathy), to give information about evaluation-treatment methods and results.

Case Report

The complaints of 27-year-old woman who applied to Special Melek Special Education and Rehabilitation Center with dysferlinopathy-LGMD2B diagnosis started 15 years ago as stair climbing, fatigue and muscle weakness. Our work is approved by the Ethics Committee of the Institute of Health Sciences of Okan University. (Protocol number: 10 , The date of approval: 17.01.2018). The paper was held according to the Helsinki Declaration. The patient was informed about the study aim and she signed informed consent forms. Participation in the study was voluntary.

Assessment Methods

MRC-(0-5 Medical Research Council) Scale was used for muscle strength assessment (Brigadier). Timed performance tests were used to assess physical performance. Times for each test were recorded with a stopwatch.

The tests were

1. Rotation time of right to left: Lying on supine position, it is wanted to roll to one side and return to the supine position as soon as possible. The patient repeated the rolling from the supine position to the prone position.
2. Lying / sitting test: Starting from the lying position on the back, the patient was asked to move to the normal sitting position.
3. Sitting / standing test: Patient was asked to sit on a bed at standard height from the ground and to stand up once.
4. Duplicate sitting / lifting: The patient was asked to sit on a bench at standard height and stand up three times.
5. 50-meter walking test: The patient was wanted to walk as fast as possible on condition to go and return 25 meters [9].

Falls Efficacy Scale International-(FES-1), consisting of 16 questions, was used to assess the worries about falling [10] and number of falls was learned with anamnesis. Body Mass Index-(BMI) is calculated by weight(kg)/length(m^2) formula. Each assessment was made before and after treatment to compare the treatment efficacy.

Treatment Program

- ✓ The patient was included in a mild violent physiotherapy program- (exercises that allow singing that do not affect breathing and that do not sweat) for three days a week to prevent fatigue and muscle atrophy.
- ✓ Kinesiotape was applied to the major weak muscles (Iliopsoas, quadriceps femoris, hamstring, rectus abdominis, erector spina, deltoid) for facilitation during twelve sessions.
- ✓ For weight control, acupuncture with ear seeds was implemented to five earlobe regions (shenmen, spleen, stomach, endocrine, hunger), unilateral, once a week and eight weeks.
- ✓ Because of major weakness is in limb girdle muscles and trunk, isotonic exercises on these muscles were performed with the patient lying on her back, eliminating gravity. After five months, when the patient began to tolerate gravity, fascilitation-focus-rhythmic initiation technique was applied from PNF techniques.

- Flexor and extensor muscle training was performed gradually. For example for body flexors, the patient was positioned at 100 degrees flexion in the first monthand she asked to sit at 90 degrees (in the neutral seating position). She was asked to sit at 90 degrees from the 110 degree flexion position in the second month and continued decreasing 10 degrees for every step from 0 degrees to 90 degrees. The measurements were done with goniometers.
- The same training was performed for trunk extension and lateral flexion.

- ✓ Other muscles than those mentioned above, were implemented PNF on half-patters for 7 months and resistive exercise program after 5 months.
- ✓ In home program, isometric exercises were added to all the mentioned muscles in addition to the modified clinical exercises.

Highlights

- ✓ Isotonic, isometric, PNF exercises and kinesiotaping in PwD increase muscle strength.
- ✓ Ear acupuncture can be thought to provide weight control in PwD.

Decreased CK may result in decreased muscle damage, increased exercise tolerance.

Results

The treatment effects on Timed Performance Test are given in (Table 1).

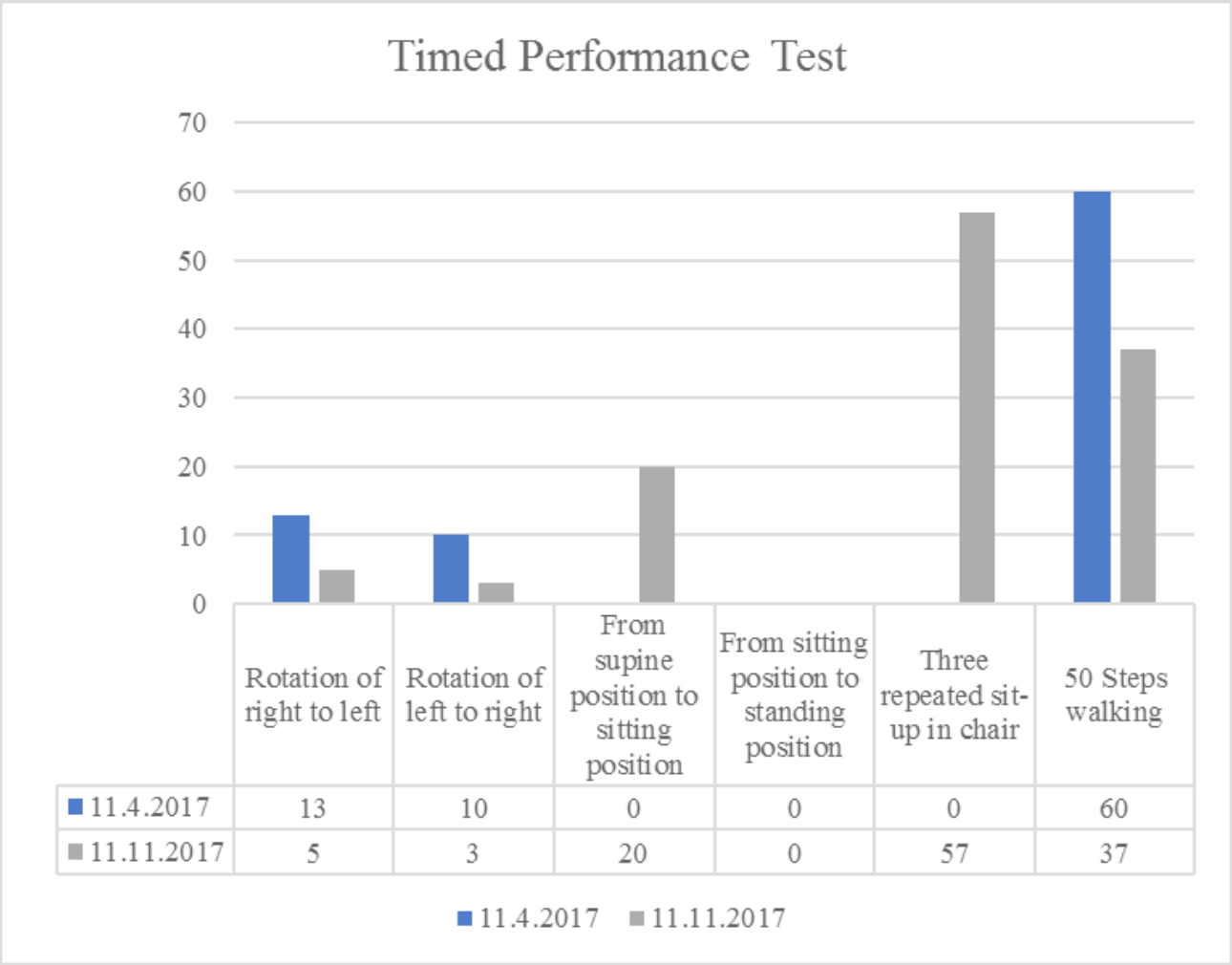

Table 1: Times of performing Timed Performance Test tasks are as shown in the table.

After the treatment concerns about falling were significantly reduced as seen in (Table 2).

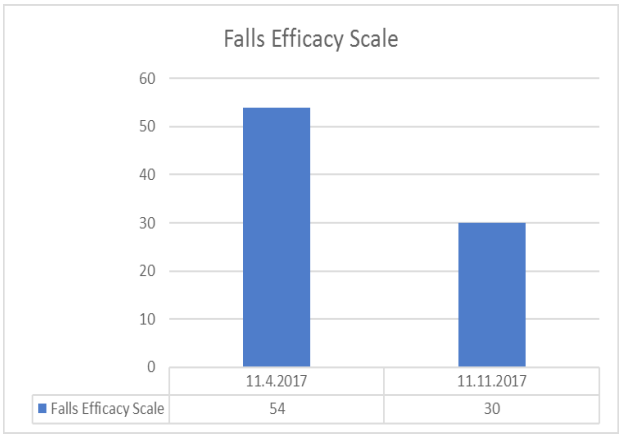

Table 2: Falling worry scores are shown in the table.

Also, 38 drops in the previous 7 months of treatment; 7 drops after the treatment were recorded. Manual muscle test results were evaluated over 10 points. When compared top pre-treatment, there was a significant increase in muscular strength around the trunk, hip and shoulder girdle, knee and ankle, as seen in (Table 3).

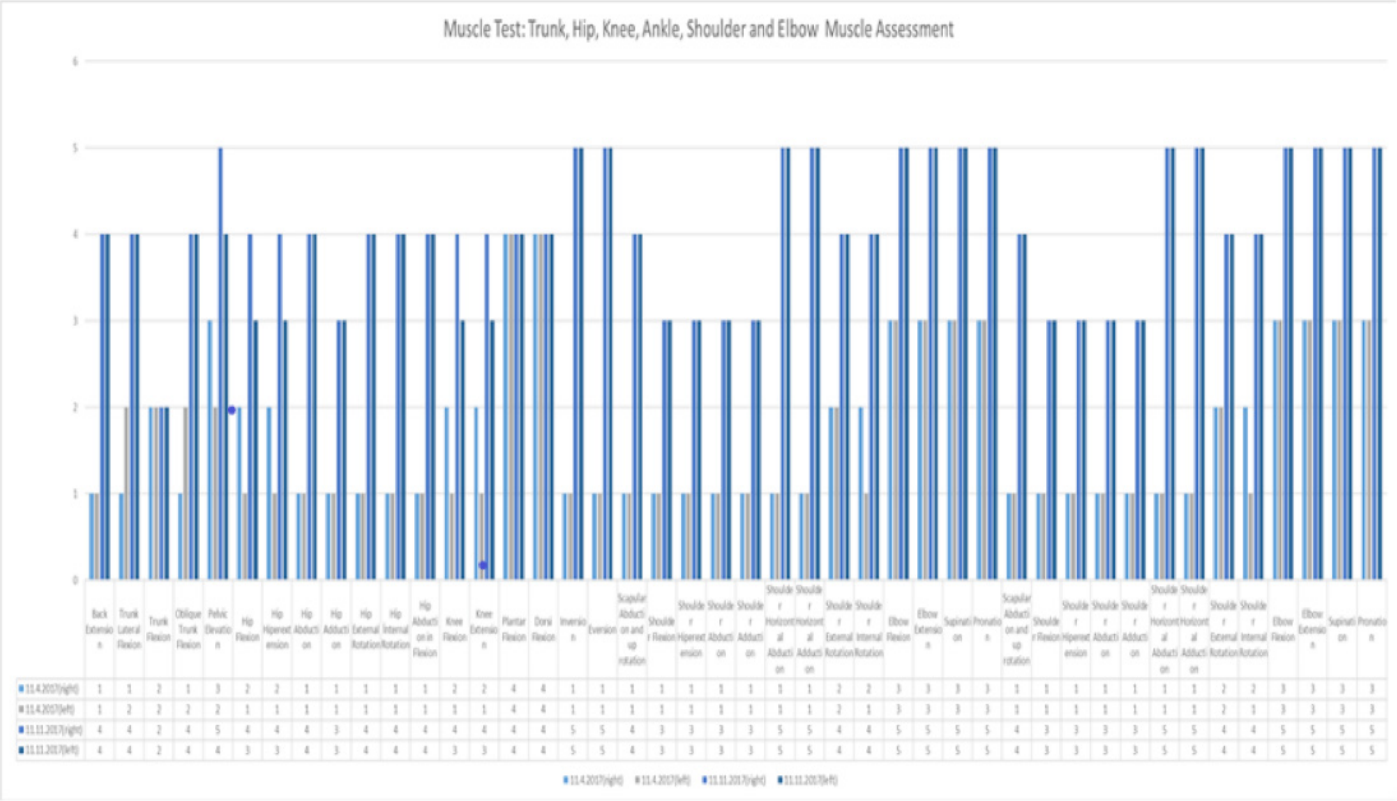

Table 3: The changes of muscle strength values according to treatment is as shown in the table.

Patient gave 5 kg in 7 months with ear acupuncture application (BMI Before Treatment:26,6 kg/m^2, BMI After Treatment:24 kg/m^2). The patient's CK level was 700 IU/L in 2002, whereas in November 2018, decreased to 393 IU/L (The CK reference range is 0-145 IU/L).

Discussion

It is very important to closely monitor muscle weakness and weight in PwD. Muscle strength is the basis of DLA, affecting ambulance and balance. Muscle weakness-related falls also increase. With decreased muscle strength, increased immobilization and muscle tissue turns into adipose tissue. According to a study about the physical activity effects on dysferlinopathy progression; 18 patients who had active sports before the diagnosis showed that progression was faster [7]. So, in our treatment program, the exercises were implemented at mild severity and intensity. In a study by Biondi and colleagues on dysferlinopathic rats, concentric/isometric exercises increased muscular strength and eased muscle dystrophy by limiting the membrane lesions accumulation; eccentric exercises, especially with age, have worsened membrane lesions and increased myofibril damage [11].

According to anamnesis, the hip and shoulder girdle muscle weakness occurred before the limb muscle weakness, and the muscular strength at the limbs decreased as the mobility decreased. When the diagnosis is considered, it is thought that distal limb muscle weakness is a consequence of proximal muscle weakness. It supports this idea that the lack of weakness in the fingers, hand, wrist, and facial muscles; distal muscle strength was higher than proximal before treatment; distal muscle strength increased more than proximal during the treatment. The hand and wrist muscle strength was 4+ and over found; so these muscles are not included in the treatment. Research in recent years, participants' CK levels were examined-(exercises with eccentric contractions); CK levels increased after exercise [12-14] .In a study of exceptionally Australian footballers, CK level was found to be lower after training [14]. It was thought to be delayed recovery and chronic muscle damage because of this [14]. In the result of these study, CK level increased with muscle injury, fatigue and in this context it may be considered to decrease muscle damage and increase excercise

tolerance as the cause of the fall of the CK level in the long term. It is foreseen that this is the reason why patient's CK level decreases over the years. More research can be done to support this idea. As a result of our study, we observed that suitable for muscular performance isotonic, isometric, PNF exercises and kinesiotape application in PwD increased muscle strength, improved physical performance, decreased falls number and worry about falling. Ear acupuncture can be thought to provide weight control.

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References

1. Moore SA, Shilling CJ, Westra S, Wall C, Wicklund MP, et al. (2006) LGMD in the United States. J Neuropathol Exp Neurol 65: 995-1003.
2. Bansal D, Campbell KP (2004) Dysferlin and the plasma membrane repair in muscular dystrophy. Trends Cell Biol 14: 206-213.
3. Bansal D, Miyake K, Vogel SS, Groh S, Chen CC, et al. (2003) Defective membrane repair in dysferlin-deficient muscular dystrophy. Nature 423: 168-172.
4. Díaz J, Woudt L, Suazo L, Garrido C, Caviedes P, et al. (2016) et al. Broadening the imaging phenotype of dysferlinopathy at different disease stages. Muscle Nerve 54: 203-210.
5. Angelini C, Nardetto L, Borsato C, Padoan R, Fanin M, (2010) et al. The clinical course of calpainopathy (LGMD2A) and dysferlinopathy (LGMD2B). Neurol Res 32: 41-46.
6. Jin SQ, Yu M, Zhang W, Lyu H, Yuan Y, et al. (2016) Dysferlin Gene Mutation Spectrum in a Large Cohort of Chinese Patients with Dysferlinopathy. Chin Med J (Engl) 129: 2287-2293.
7. Angelini C, Peterle E, Gaiani A, Bortolussi L, Borsato C, et al. (2011) Dysferlinopathy course and sportive activity: clues for possible treatment. Acta Myol 30: 127-32.
8. Nguyen K, Bassez G, Krahn M, Bernard R, Laforêt P, et al. (2007) Phenotypic study in 40 patients with dysferlin gene mutations: High frequency of atypical phenotypes. Arch Neurol 64: 1176-182.
9. Coote S, Garrett M, Hogan N, Larkin A, Saunders J, et al. (2009) Getting the balance right: a randomised controlled trial of physiotherapy and Exercise Interventions for ambulatory people with multiple sclerosis. BMC Neurol 9: 34.
10. Yardley L, Beyer N, Hauer K, Kempen G, Piot-Ziegler C, et al. (2005) Development and initial validation of the FES-I. Age Ageing 34: 614-619.
11. Biondi O, Villemeur M, Marchand A, Chretien F, Bourg N, et al. (2013) Dual effects of exercise in dysferlinopathy. Am J Pathol 182: 2298-2309.
12. Hicks KM, Onambele-Pearson GL, Winwood K, Morse CI (2017) Muscle-Tendon Unit Properties during Eccentric Exercise Correlate with the CK Response. Front Physiol 8: 657.
13. Kearns AK, Bilbie CL, Clarkson PM, White CM, Sewright KA, et al. (2008) The creatine kinase response to eccentric exercise with atorvastatin 10mg or 80mg. Atherosclerosis 200: 121-125.
14. Hunkin SL, Fahrner B, Gastin PB (2014) CK and its relationship with match performance in elite Australian Rules football. J Sci Med Sport 17: 332-336.