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CASE REPORT

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Early adulthood onset of muscle weakness in facioscapulohumeral muscular dystrophy and physical therapy management: An unusual case report

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Abstract
Facioscapulohumeral muscular dystrophy is an uncommon and rarely seen progressive myopathic disorder. It is usually seen during periods of rapid growth (4-5 years). Proximal and distal upper extremities characteristically show weakness and atrophy. Face, scapular, shoulder, leg, hip belt muscles are frequently asymmetrically affected. In this paper, a case is presented of this rare, difficult to diagnose disease, not previously reported in this age group, and the findings are discussed in the light of information of earlier cases in Turkey. A 26-year old female patient presented at Hacettepe University Neurology outpatient clinic with complaints of muscle weakness and atrophy in the left arm and leg, which had been ongoing for 4 years. The muscle weakness of left upper, lower extremities and right limb, further cervical cord atrophy, winging scapulae, abnormal spinal curvature were observed. This case report presents information about the differential diagnosis, detailed symptoms, and physical therapy management of a Fascioscapulohumeral patient.

Keywords: Fascioscapulohumeral, Myopathic, Atrophy, Physiotherapy

Introduction

Facioscapulohumeral muscular dystrophy is the third most observed muscular dystrophy and results from autosomal dominant inheritance. And it is known as Landouzy-Dejerine disease [1]. The prevalence of Facioscapulohumeral muscular dystrophy (FSHD) is estimated at 1/20.000. The symptoms of FSHD are dysarthria, wing scapulae, perioral, and peri-orbital facial muscle weakness, upper arm muscle and shoulder girdle muscle weakness, pseudohypertrophy [2]. Also, hearing loss, muscle weakness develops from proximal to distal parts. The onset of symptoms in most patients is before age-20-year old [3]. The patient with FSHD is observed deletion on 3.3 kb chromosome 4, pathologic with hypermethylation of DNA and specific D4Z4. Located a retrogene on the D4Z4 macrosetallic repeat on chromosome 4q35 bring about FSHD [3,4].

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This case report presented a patient, aged 26-year-old, had onset of muscle weakness in the shoulder and pelvic girdle. This is one of the early adults with FSHD onset of impaired upper-limb and pelvic muscles weakness and treated with physical therapy and rehabilitation.

Case Report

Presenting Concerns

A 26-year old female patient, of height 1.66 m and weight of 45 kg, with complaints of shoulder, pelvic girdle and muscle weakness on the left side, which had been ongoing for four years. She was very active until 22-year-old. Firstly, she had walking difficulties because of dorsiflexion restriction of the right foot. After time, she stopped to use of bilateral arms because of shoulder muscle weakness and poor finger control. Also, the asymmetrical muscle weakness of facial, serratus anterior, trapezius, latissimus dorsi, deltoid, pectoralis major and rhomboideus was observed. Medical treatment of this patient included Salbutol, Ventolin, Volmax (albuterol) and Mono-hydrate-kreatin (might be helpful as a muscle performance enhancer). Her family history onset of muscle weakness no observed.

Diagnostic Focus and Assessments

In this patient, psychosocial status, history, background, upper extremity range of motion (ROM) (Table 1), the muscle strength of upper and lower extremity were evaluated (Table 2). The Fatigue Severity Scale (FSS) was used to evaluate the restriction of daily activities. The Box and Block Test (BBT), Nine-Hole Peg Test (NHPT), Bournemouth Neck Survey, Beck Depression Inventory were used to assess hand skills. Anterior and lateral clinical posture analysis was applied. A rough grip (grip meter), and finger grip (pinch meter) were used for distal muscle strength assessment. The evaluation of daily living activities in FSHD patient was applied using the Short Form Health Survey (SF-36).

Table1. Upper Extremity Range of Motion (ROM) Measurement Results

ROM	Pre-treatment (Right)	Post-treatment (Right)	Pre-treatment (Left)	Post-treatment (Left)
Abduction of Shoulder	10^0	15^0	7^0	10^0
Flexion of Shoulder	5^0	10^0	5^0	10^0
Flexion of Elbow	20^0	30^0	15^0	20^0
Flexion of Wrist	65^0	70^0	60^0	65^0
Extension of Wrist	60^0	65^0	50^0	50^0
Supination of Wrist	50^0	55^0	50^0	50^0
Pronation of Wrist	60^0	60^0	45^0	50^0
Ulnar Deviation of Wrist	25^0	25^0	20^0	20^0
Radial Devation of Wrist	30^0	30^0	20^0	20^0
Flexion of MCP	60^0	65^0	50^0	60^0

Table2. Evaluation of Muscle Strength of the Upper and Lower Extremities

ROM	Pre-treatment (Right)	Post-treatment (Right)	Pre-treatment (Left)	Post-treatment (Left)
Abduction of Shoulder	2	2	1	2
Flexion of Shoulder	2	2	1	2
Adduction of Shoulder	1	2	1	1
External Rotation of Shoulder	2	2	2	2
Medial Rotation of Shoulder	1	2	1	2
Flexion of Elbow	3	4	3	3
Extension of Elbow	3	3	2	3
Flexion of Wrist	4	4	3	4
Extension of Wrist	4	4	3	4
Radial Deviation of Wrist	3	4	3	4
Ulnar Deviation of Wrist	3	4	3	4
Flexion of MCP	3	4	3	4
Adduction of Hip	3	3	2	2
Flexion of Hip	3	4	2	3
External Rotation of Hip	2	3	2	2
Medial Rotation of Hip	2	3	2	2
Extension of Hip	2	3	2	3
Flexion of Knee	3	4	2	3
Extension of Knee	2	3	2	3
Dorsiflexion of Ankle	3	4	2	3
Plantar flexion of Ankle	3	4	3	3
Pinch Grip Strength (kg)	2.33	4.33	1.33	3.66
Rough Grip Strength (kg)	13	15	11.33	14.33

Clinical Findings

In the neurological examination performed, low amplitude of BCAP and prolongation in distal latencies in the left musculocutaneous were recorded. Typically the scapular adduction angle is 17º while it was determined as 14^0 in the current patient, and patient's pulmonary function testing was unremarkable (FVC %96 predicted, FEV1 %90 predicted). The patient stopped using Ventolin because of having a normal pulmonary function. Prednisone and cortisone not prescribed through her treatment. Blood testing including erythrocyte sedimentation rate, complete blood count, antinuclear antibodies, rheumatoid factor, parathyroid hormone, HgA1c, TSH, pyruvate, and immunofixation was observed normal out of creatine kinase. The level of creatine kinase was found as 747 U/L, typically ranged from 30 to 170 U/L.

In the anterior postural analysis of the current patient, the right shoulder was higher, and the left-side lateral flexion was detected. In the posterior postural analysis, winging scapulae dropped left shoulder, and pseudohypertrophic gastrocnemius muscle was observed. When she performed shoulder abduction or anterior elevation, winging scapulae were observed (Figure 1a). In the lateral postural analysis, decreased neck flexion increased total kyphosis, more increased lumbar lordosis, and anterior pelvic tilt was observed (Figure 1b). In the measurement of neck pain, 60 points from a maximum of 70 points were recorded on the Bournemouth Neck Survey. A total of 38 points were recorded on the Neck Disability Questionnaire, in which ≥35 points is explained as a complete disability. With a score of 40 points on the Beck Depression Inventory, the patient with FSHD was diagnosed as having a severe depressive mood.

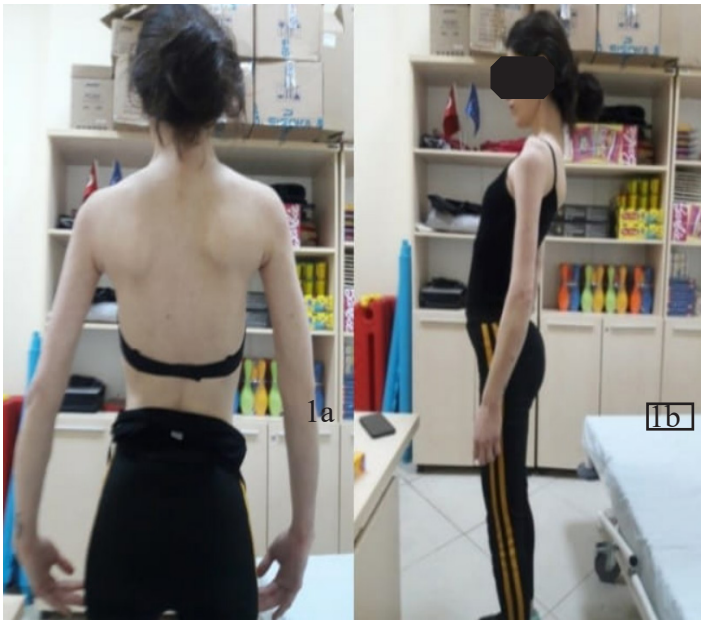

Figure 1. Winging scapulae (1.a), increased lomber lordosis and kyphosis posture (1.b)

In the measurement of ambulation level, 7 points from a maximum of 34 points were recorded on the North Star Ambulation Scale. In the measurement of functional motor scale, 21 points from a maximum of 100 points were recorded on the Gross Motor Functional Measurement. In the analysis of the activity-based measure of functional exercise capacity, 300 m was recorded on the 6 min walk test. Also, 12 seconds was recorded on the 10 m walk test.12 sn was recorded on the climbing stairs with helping Also, seven sn was observed on the stepping downstairs with assisting. 18 sn was recorded on the climbing stairs with no help; while 15 sn was recorded on the stepping down with no help before taking the physiotherapy treatment.

The patient with FSHD was diagnosed as having chronic fatigue syndrome, with a score of 6.22 points on the Fatigue Severity Scale (FSS). With a score of physical functioning 20/100, role limitations because of physical health 0/100, role limitations because of emotional problems 0/100, energy and vitality 30/100, emotional well-being 24/100, social functioning 12.5/100, pain 22.5/100, general health 15/100, health change 50/100 points on the SF-36, the patient with FSHD was diagnosed as having a low quality of the life. In the Box-Block Test, which measures hand grip ability, the number of box blocks in 1 minute is recorded. The test was repeated three times, and the highest scores recorded were 17 for the right extremity and 11 for the left.

The Nine-Hole Peg Test (NHPT), which measures manual dexterity, consists of 9 wooden bars of equal dimensions and a woodblock with nine holes. The patient places the bars in the holes and the time is recorded. The average of 3 trials was used for the evaluation. The current patient had no movement in the left and arm in regardless of wrist and finger movement and the average placement time of the blocks was 40 seconds.

Neurological Examination (DTR, pathological reflex), mental status, speaking difficulties, facial appearance were evaluated before treatment. Normal patellar reflex, biceps, and brachioradialis reflexes were observed.

As right side facial muscle strength was lower than the left side, asymmetrical facial muscle weakness (right side the muscle weakness of orbicularis oris, bilateral muscle weakness of orbicularis oculi) observed. Also, orbicularis oculi, levator palpebrae, levator labii superior and buccinator muscles were assessed 3/5 on the Medical Research Council (MRC) scale. Weakness was observed predominantly proximal upper extremity, while muscle weakness was observed distal lower extremity. Also, muscle weakness of neck, especially sternal part of sternocleidomastoideus (SCM), was found. Rough and finger grip strength (pinch) values were measured (Table 2) using a Jamar hand dynamometer to measure rough grip strength and a pinch meter to measure the pinch finger grip strength.

Treatment Protocol for FSHD

Physical treatment of upper and lower extremity included symmetrical and reciprocal training included stretching, strength (cervical smoothing exercises, neck splenius and respiratory muscle strength, augmentation and neck flexion, extension) exercises, endurance, aerobic exercises followed by 3 sets of 10 repeat three days per week in 4 months and kinesiotape once per a week during 4 months. Kinesiotape was used for improving foot drop, balance, and walking capacity (Figure 2).

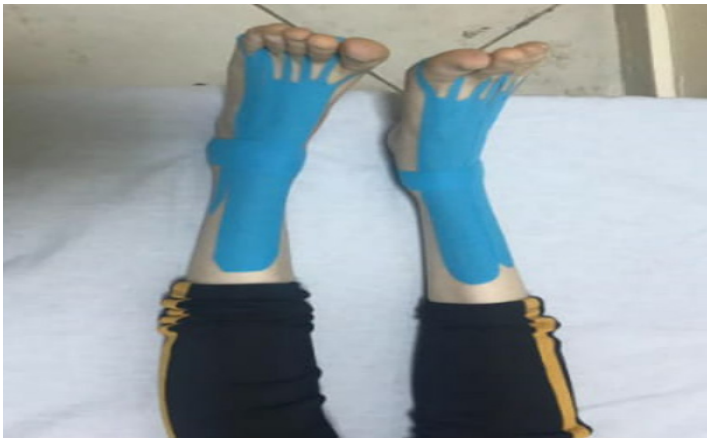

Figure 2. Kinesio Tape Method for Foot DropIn the Post-treatment Outcomes

In the measurement of ambulation level, 22 points from a maximum of 34 points were recorded on the North Star Ambulation Scale. In the measurement of functional motor scale, 48 points from a maximum of 100 points were recorded on the Gross Motor Functional Measurement. In the measurement of activity-based measure of functional exercise capacity, 360 m was recorded on the 6 min walk test. Also, 8.6 s was recorded on the 10 m walk test. In the measurement of neck pain, 50 points from a maximum of 70 points were recorded on the Bournemouth Neck Survey. A total of 30 points were recorded on the Neck Disability Questionnaire, in which ≥35 points is explained as complete disability. With a score of 25 points on the Beck Depression Inventory, the patient with FSHD was diagnosed as having a middle depressive mood. The muscle strength and ROM of distal and proximal upper extremities increased.With a score of 3.88 points on the Fatigue Severity Scale (FSS), patient was diagnosed as having a high quality of life. The most top scores of Box-Block Test recorded were 21 for the right extremity and 15 for the left. In the Nine-Hole Peg Test, the average placement time of the blocks was 34 seconds. Ten s was recorded on the climbing stairs with helping. Also, 5.60 s was observed on the stepping downstairs with assisting. 13.07 s was recorded on the climbing stairs with no help; while 5.60 s was recorded on the stepping down with no assist after the physiotherapy treatment.

Discussion

This paper presents the detailed symptoms and treatment methods of physical therapy applied to a patient with Facioscapulohumeral dystrophy disease, and information about the differential diagnosis. To the best of our knowledge, this is the first study to have investigated detailed symptoms, the differential diagnosis, and physical therapy methods used in the rehabilitation of Facioscapulohumeral dystrophy disease.

Early detection of the disease is critical as the prevention of foot dorsiflexion muscle significantly reduce the progression of the disease [5,6). As the current patient presented at the polyclinic after eight years of disease progression, almost complete findings of the disease late period was observed.

Van der Kool et al. investigated the effects of training and albuterol on pain, functional status, psychological distress, and fatigue in facioscapulohumeral muscular dystrophy. Their study included 65 patients with FSHD. They divided four group as non-training+placebogroup (n:16), non-training+albuterol group (n:15), training+placebo group (n:19), training+albuterol group (n:15). Albuterol (8 mg twice a day) and moderate-intensity strength training were given at 52 weeks. They did not observe significant differences in changes between the training and non-training groups with or without albuterol.

In contrast to their expectation, no synergistic effects between strength training and albuterol were found. Conclusively, they observed that strength training and albuterol failed to have a significant impact on pain, fatigue, functional status and psychological distress [7). This study found that symmetrical and reciprocal training included stretching, strength, aerobic exercises had a significant difference in fatigue, muscle strength, quality of life, functional status and psychological level of patients with FSHD.

Olsen et al. experience that exercise programs have been observed to increase strength and endurance in patients with FSHD. They investigated the effect of aerobic training in patients with facioscapulohumeral dystrophy (FSHD). Twelve weeks of low-intense aerobic exercise provided maximal oxygen uptake and improved workload with no signs of muscle damage. They found that aerobic exercise is a safe method to increase exercise performance in patients with FSHD [8). This conclusion was similar to the completion of the current study in terms of the effect of aerobic training on muscle weakness.

In physiotherapy applications, the exercise program should aim to prevent movement inhibition of muscular weakness, proximal muscular atrophy, reduce distal and proximal joint contractures, and increase muscle strength. Also, aerobic training with or without post-exercise protein-carbohydrate supplementation improves muscle strength and workload in patients with FSHD (9). In parallel with literature, the current patient with FSHD was applied with late-period exercises improve muscle strength, stretching, endurance, fatigue, the level of depression, manual dexterity.

Conclusion

This paper can be considered to contribute to literature as it reports in detail the orthopedic evaluations and physical therapy and rehabilitation of the 26-year old patient with Facioscapulohumeral dystrophy. Detailed neurological and orthopedic assessment should be made of these patients for the implementation of appropriate physiotherapy and rehabilitation programs. Late period physical therapy and rehabilitation for FSHD prevents movement inhibition of muscular weakness, proximal muscular atrophy, reduce distal and proximal joint contractures, fatigue, the level of depression and increase muscle strength, timing performance, exercise capacity, endurance, walking ability.

Competing interests
The authors declare that they have no competing interests.

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