Facioscapulohumeral muscular dystrophy; A report of three cases from Pakistan

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ABSTRACT

Facioscapulohumeral muscular dystrophy is often described as the third most common form of muscular dystrophy with a slow progression. We present here, three male cases from Pakistan who presented with raised serum creatine kinase. Electrodiagnostic evaluation revealed myopthic findings in muscles and muscle histopathology was consistent with a dystrophy favoring the diagnosis of Facioscapulohumeral muscular dystrophy. The disease was managed through supportive measures involving therapeutic exercises, activity modification and use of orthotics to improve function and mobility. (Rawal Med J 2014;39: 96-99).

Keywords: Facioscapulohumeral muscular dystrophy, nerve conduction studies, electromyography.

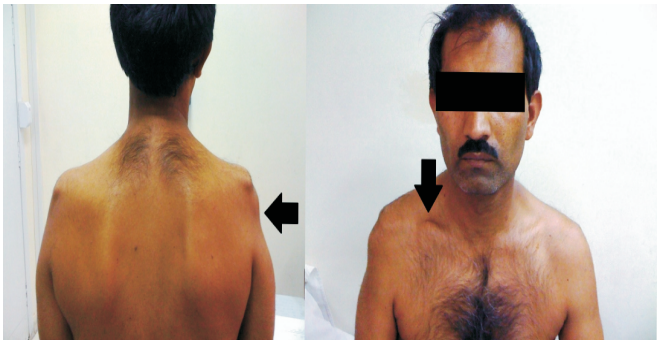
INTRODUCTION

Facioscapulohumeral muscular dystrophy (FSHD) is an established genetic muscular disorder characterized by impairment and weakness of the muscles of face, shoulder blades and upper arms. Its prevalence is about 1 to 5 per 100,000 individuals.[1] It is not considered life threatening due to its slow progression and rare advancement to cardiac abnormalities.[2] Currently, there is no cure for FSHD and all the clinical trials focusing on increasing muscle mass or suppressing inflammatory reactions have been unsatisfactory.[3] Over the past ten years, major advances have happened in the understanding of the genetics of this disorder, however, the exact mechanisms that lead to muscle atrophy and weakness still remain unrecognized.[4] A case of FSHD was reported by us in 2012 for the first time in Pakistan.[5] We report here a series of three cases who were referred to Armed Forces Institute of Rehabilitation Medicine for Nerve Conduction Studies and Electromyography (NCS/EMG).

CASES

Case 1: A 32-year-old male was referred to our institute for NCS/EMG to rule out suspected right Brachial Plexus Injury (BPI). He had weakness of right shoulder and difficulty in over-head activities with right arm. He was right handed and attributed his weakness to heavy manual work involving use of heavy hammer for past one decade. Difficulty in chewing food was also noted. BPI was suspected due to an old fractured clavicle, which occurred ten years ago. Patient had multiple consultations with labels such as BPI, adhesive capsulitis and Post-polio paralysis. On examination, there was wasting of right deltoid, biceps and scapular muscles with right clavicle prominence due to old fracture (Fig. 1). Muscles of face were also wasted. Rest of the clinical examination was unremarkable.

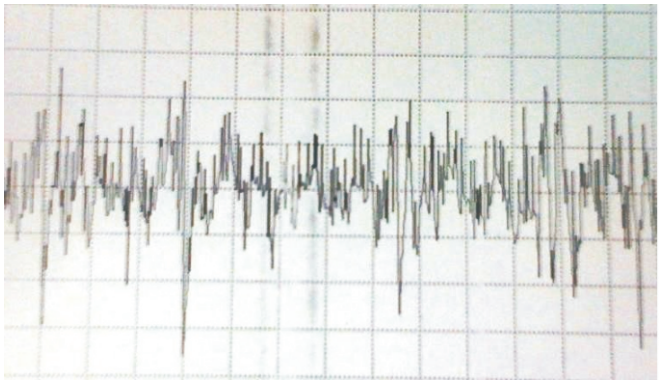
Fig. 1. Wasting of Trapezius, Deltoid, Facial and Scapular muscles and prominence of mid right Clavicle due to old fracture.



The patient's laboratory investigations yielded raised serum creatine kinase (CK) and aldolase levels, (510 U/L and 8.3 U/L, respectively). NCS were normal and EMG of supraspinatus, infraspinatus, buccinator, deltoid and biceps muscles bilaterally, revealed small polyphasic

motor unit action potentials (MUAP) with early recruitment and full interference pattern consistent with a myopathic disorder (Fig. 2). EMG of lower limbs was normal. Muscle biopsy of right deltoid muscle revealed dystrophic findings, which further confirmed the diagnosis of FSHD. Genetic testing for FSHD is not available in Pakistan.

Fig. 2. Polyphasic, small, short MUAP with early recruitment pattern in right deltoid muscle (a typical myopathic EMG pattern.

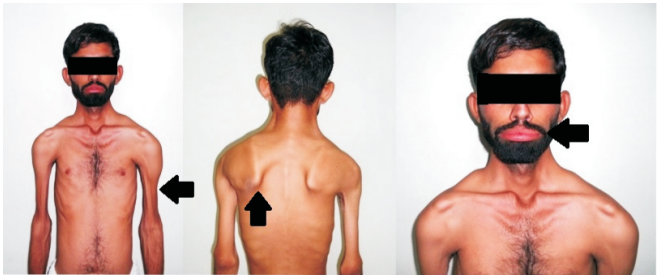


He was counseled about the nature of disease, activity modification, energy conservation techniques and therapeutic exercises. At three months follow up, he had better understanding of his disease and reduced fatigue with a little better functional profile.

Case 2: A 20-year-old male was referred to us for NCS/EMG to look for Radial Nerve injury because of weakness and wasting in right arm suspected due to previous history of intramuscular injection in right arm. He was previously labeled as a case of Poliomyelitis. He also had received physiotherapy sessions but failed to continue due to excessive fatigue. On detailed inquiry, he gave history of progressive muscular weakness, more pronounced in upper limbs and face for last eight years that had involved right arm initially (the dominant side) and then progressed to the left arm. There was a continuing weight loss and constipation. He had difficulty in bringing hands to mouth during eating and doing routine activities that required movements above shoulder level. Patient was unable to blow whistle or drink juices with straws. He was able to eat but had difficulty in breaking hard eatables during mastication. There was no difficulty in lower limb activities.

On examination, there was wasting of arms (Right>left), scapular muscles and quadriceps with relative sparing of deltoid more on the left side with scapular winging and Pectus Excavatum (Fig. 3). Rest of the limbs showed normal appearance. Muscle power was 1-2/5 in proximal muscles of arms according to Medical Research Council Scale and 5/5 in distal muscles of arms and all key muscles of legs. Gower's sign was negative. Biceps and triceps jerks were depressed.

Fig. 3. Wasting of Trapezius, Biceps, Triceps, Supraspinatus and Infraspinatus Muscles, scapular winging and inability to whistle.



His serum CK was raised (641 U/L) while aldolase was normal (6.82 U/L). NCS were normal but EMG of the affected muscles (deltoid, biceps, triceps, quadriceps and masseter) showed myopathic pattern. Muscle biopsy of deltoid muscle showed dystrophic changes. Genetic testing was not done. He was thoroughly counseled about the disease and was managed by energy conservation techniques, activity modifications, simplification of the tasks and therapeutic exercises. The patient did not follow-up later on.

Case 3: A 23-year-old male presented with complaints of wasting of shoulders and arms with mild pain and increasing weakness noticed for the last seven years. He also had difficulty in chewing and above head activities. There was no complaint in lower limbs. On examination, he had scapular winging, wasting of arms (Right>left) and scapular muscles with relative sparing of deltoid more on left side (Fig. 4). Rest of the limbs showed normal

appearance. Biceps jerks were depressed. Serum CK was raised (707 U/L) and aldolase was normal (6.7 U/L). The patient was reviewed by a neurophysician, who suspected FSHD clinically and referred him to us with a request for NCS/EMG, which were consistent with a myopathic disorder. Diagnosis was confirmed on muscle biopsy. He was offered rehabilitation consultation but he opted for therapeutic touch (Placing hands of the healer upon the body part of the person to be cured with the intent of spiritual energetic healing) [6] and prayer therapy under supervision of a religious scholar as per advice of his father. He was lost to follow-up.

DISCUSSION

FSHD is an autosomal dominant inherited disorder with chromosomal linkage of 4q35 locus first described in 1886 by French physicians; Landouzy and Dejerine.[4] The specific mechanism for the disease is not established, however, sporadic mutations are noted in 20-30 %.[1] Clinical criteria for the diagnosis were established by an international consortium in 1991 but the gold standard to confirm the diagnosis is genetic testing.[2,7]

FSHD usually presents at an average age of 20 years.[1] Facial muscles are characteristically involved producing difficulty in pursing lips, whistling, drinking with a straw and smiling. Weakness of shoulder girdle muscles produces winging. Asymmetrical muscular weakness is common but it may be due to overwork. In the lower limbs, the proximal musculature of the hip girdle is typically affected, often late in the disease course.[1] Early involvement of ankle dorsiflexors has been noted in some cases. 20% cases may become wheelchair bound due to the rapid course of the disease in their second or third decade.[1] Contractures and scoliosis are relatively uncommon in FSHD.[1]

Apart from muscular weakness and its sequelae, 32-72% patients experience pain.[4] Cardiac abnormalities are rare and nearly half of FSHD patients develop mild restrictive lung disease, rarely requiring nocturnal ventilatory support.[1] Some patients may develop visual and auditory impairments. Workup for suspected FSHD includes assessment of muscle enzymes, NCS/EMG, muscle biopsy and genetic testing.[1]

Currently, there is no curative genetic or pharmacological treatment for this disease. The mainstay of management is care directed at the symptomatology in order to maximize functional abilities and improve the quality of life. The role of exercise in maintaining or improving strength and function remains controversial because of concerns about over-use weakness and limited number of clinical trials.[4] Orthotics may help in some functional activities. Surgical methods have been developed to provide stability at the shoulder and involve fixing the scapula to the thorax with help of fascial or synthetic slings (scapuloplexy) or by fastening it with wires, screws or plates (scapulodesis).[4,8] Future treatments involving the autologous cell therapy with mesangioblasts, nevertheless, are a strong hope for FSHD community.[4]

CONCLUSION

This case series highlighted the importance of NCS/EMG as a tool in diagnosis. A focused clinical examination, timely referral to concerned specialist physicians and judicious use of investigations can offer an early and correct diagnosis of FSHD. After the diagnosis, the mainstay of treatment is supportive under supervision of a neurophysician and the physiatrist.

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