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FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY WITHOUT FACIAL SYMPTOMS

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ABSTRACT

Introduction: Facioscapulohumeral muscular dystrophy (FSHD) is a progressive muscular dystrophy which starts with shoulder and face muscles involvement and rarely spreads to pelvic muscles. It has autosomal dominant inheritance pattern, serum CK levels are normal or mildly high, and EMG is myopathic.

Case report: We present the 43 year old male patient with pain, numbness and weakness in cervical area and both arms for 1 year. His mother has similar symptoms as well. His cervical spinal nerve EMG results were found normal.

Results: EMG results were found normal. Our patient had the defect of closing eyelids or pressing them based on faced muscles involvement just like regular FSHD cases. Because of the atrophy of lower face muscles, lips seemed to be fuller. On the other hand, there was a no whistling or blowing defects related to orbicularis oris muscle and face involvement. We presented this study as this muscle disease represents itself with weakness and numbness in muscle is seen rarely in literature and our patient did not have one of its components (facial).

Key Words: : Facioscapulohumeral muscular dystrophy, neuromuscular disorder, muscle imaging, muscular dystrophy, facioscapulohumeral/diagnosis

INTRODUCTION

Muscle dystrophies are genetic diseases which has muscle fibers' histopathologically repetitive both destruction (necrosis), repair (regeneration). In the end, because of necrosis, muscle lose is seen and endomicial and perimicial fat and connective tissue takes muscles' part. (1)

Facioscapulohumeral muscular dystrophy (FSHD) is a progressive muscle dystrophy starts with shoulder and face muscles, rarely spreads to pelvic muscles and is fist described by Landouzy and Dejerine in 1886. Prevalence is between %0.005-%0.05. It usually starts in 2nd decade and is very rare to start before the age of 5. In familial cases, the most seen pattern in autosomal dominant (2). 4q35 gene deletion is the most frequent gene pathology (3). Scoliosis is rare. Pectus ekskavatum deformity is seen more frequent. Cardiomyopathy is not seen. Some patients experience loss of hearing for high frequency that can be introduced only by eudiometric tests. FSHD with retinal vascular diseases is described Coats disease. Clinical symptoms are usually diagnostic. Serum CK levels are normal or mildly high, EMG is myopathic rarely normal. Muscle biopsy is used only in suspicious cases for differential diagnosis. (1) In this study, a case of facioscapulohumeral muscular dystrophy with normal EMG and no facial symptoms.

CASE REPORT

43 year old male, TIR driver patient presented to Trakya University Medical School Physical Therapy and Rehabilitation clinics with restriction of shoulder movings started almost 20 years ago, has been increasing for 1 year and witness, pain, numbness in upper extremity. It was found out that his mum also suffered

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from restrictions of shoulder movings. In his physical examination scapula was found 4 cm lateral from medial line (scapular abduction). In neurological examination, consciousness was open, cooperated and place-time-person orientation was complete. Mimic muscles were seen normal. There was no loss in mimic movements or facial asymmetry. Both shoulders were restricted in active 90degrees flexion and abduction. Passive range of motion was full open. In motor examination, right side muscles were intact, left side C4,C4 myotomes were 4/5,C7 myotom was -4/5 muscle strength. In sensory examination, hypoesthesia was found on the left C3, C4 and C6. Bilateral Deep tendon reflex was normal, Hoffman Test was bilateral negative. Spurling test was positive on the left. According to the laboratory test results, total biochemistry and full blood count values were normal. CRP 0,8 mg/dl, erythrocyte sedimentation rate: 6mm per hour, and CK level were 357 IU/l (mildly high). (Normal values for men; CRP: 0-0.5 mg/dl. Erythrocyte sedimentation rate: 0-20mm, CK: 38-234 IU/L.) The patient with a normal EMG result, had an MRI of cervical and detailed bilateral shoulder. According to the cervical MRI results, C3-4: left foraminal protrusion, root pressure C4-5: ventral root pressure C5-6: osteophyte + central protrusion C6-7 left foraminal protrusion were found. According to the bilateral shoulder and upper thoracal MRI: notable atrophy on the left triceps and bilateral latissimus dorsi; and the atrophy of serratus anterior, bilateral pectoralis major and minor, anterior of bilateral deltoid muscle were seen. Even though there were no pathologies of the face muscles according to these data and clinic of the patient, the patient was diagnosed as FSHD. The patient was given NSAID as for analgesic and resistive strengthening on upper extremities, isometric strengthening of the muscles around the neck and also isometric strengthening for the muscles that are atrophic.

DISCUSSION

Facioscapulohumeral muscular dystrophy (FSHD) is a progressive muscular dystrophy which starts with shoulder and face muscles involvement and rarely spreads to pelvic muscles. Generally, in FSHD cases, serum CK is normal or mildly high; EMG myopathic or rarely normal. But, in our case, serum CK level is high (CK:357) EMG results was found normal. Our patient had the defect of closing eyelids or pressing them based on faced muscles involvement just like regular FSHD cases. Because of the atrophy of lower face muscles, lips seemed to be fuller. On the other hand, there was a no whistling or blowing defects related to orbicularis oris muscle and face involvement.

Atrophies of both scapula and shoulder muscles (atrophy on the left triceps and bilateral latissimus dorsi; and the atrophy of serratus anterior, bilateral pectoralis major and minor, anterior of bilateral deltoid muscle) that are seen often were also seen on our patient. Cardiac examination of the patient was normal and there were no hearing disorders or visual disturbances. Scoliosis wasn't found. The patient is planned to be sent to the department of genetics for 4q35 deletion. We presented this study as this muscle disease represents itself with weakness and numbness in muscle is seen rarely in literature and our patient did not have one of its components (facial).

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