Facio-Scapulo-Humeral Muscular Dystrophy - A Case Report

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Introduction

Muscular dystrophy is a group of unrelated diseases, each transmitted by a different trait and each differing in its clinical course and expression[1]. The term dystrophy derives from the Greek trephein meaning nourishment[2]. It means abnormal growth. These degenerative diseases are generally progressive where degeneration and death of muscle fibres occurs at some stages. Many types of muscular dystrophy have been reported, Facio-scapulo-humeral (FSHD) type being the third most common following Duchenne and Myotonic Muscular Dystrophy having an insidious onset and slow progression[3,4]. It is also called Landouzy-dejerine type of muscular dystrophy. The gene responsible for FSHD is has been localized to chromosome 4q35-qter, although a few 4q unlinked families are known. As many other autosomal dominant conditions, FSHD may result from a new mutation, which can explain the absence of similar disease among the family members. Worldwide prevalence of FSHD is 1 in 20,000[5].

Cases of Duchenne muscular dystrophy are fairly common in our clinical practice but FSHD is relatively rare. Muscles of facial expression, scapular and humeral muscles are involved primarily whereas pelvic girdle muscles are involved in the later stages. Recently some atypical phenotype (e.g., facial sparing variety) of FSHD have been reported[6-8]. A patient of FSHD (facial sparing type) has been diagnosed recently leading to report this case after adequate explanation to the parents and taking written consent for the publication.

Case Report

An 11 years old boy from a non-consanguinous parents of lower middle class family hailing from a village of Feni district reported with the complaints of weakness and difficulty in movement of shoulder muscles, gradual thinning of shoulder and arm muscles for the last 2 years, outward protrusion of both the scapulae for 1 year and generalized weakness for 6 months. He complained of weakness during upper limb activities like picking of objects from the ground, lifting objects upwards. Though initially he had no difficulty in standing, walking and running but for the last 6 months he had been suffering from generalized weakness. His facial muscles were functioning normally, eye closure was normal while asleep. Thinning involved the muscles of shoulder and arm while the muscles of other regions showed no changes. He had no history of recurrent RTI nor dyspnoea. His birth history was uneventful and he was immunized against EPI diseases. On examination, the child was ill looking, apathetic, mildly pale, BCG mark was present. His height was 138 cm, weight 28 kg, both being within normal range. His muscles of facial expression were found to be functioning normally like smiling, blowing, wrinkling of forehead, eye closure. His shoulders, arms and scapular regions were found grossly wasted with markedly diminished muscle power and tone. His gait and joints were found to be normal. He had scoliosis to the right while standing. There was no cuff hypertrophy, Gower sign was negative. There was gross winging of both scapulae particularly while pushing forward. His blood picture was normal, serum CPK was 154U/L and ECG tracing was within normal range. Electromyogram was not done. Biopsy was taken from left Triceps muscle under local anaesthesia. Histopathological examination revealed muscle fibre with minimal variation in diameter. Small number of necrotic muscle fibres were present. Internalized sarcolemal nuclei were present in moderate amount. Mild perivascular infiltrate of lymphocytes in the perimysial blood vessels which is consistent with facio-scapulo-humeral muscular dystrophy. Considering history, clinical examinations and investigation reports the case was diagnosed as a case of Facio-scapulo-humeral type of muscular dystrophy (facial sparing type).

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As there is no specific treatment of muscular dystrophy, counseling was done with the parents regarding the aetiology, progression and possible outcome of the disease. Advice was given regarding adequate nutrition, to avoid heavy physical work and to take immediate treatment for infections particularly RTI. He was also advised regular follow-up for progression of the condition.

Discussion

Muscular dystrophy covers a group of primary muscle diseases that are genetically determined and are usually progressive[9]. The term dystrophy suggests a degenerative process of muscle. Muscle fibres form normally and then undergo degeneration, die and are removed by phagocytes and finally replaced by either fat or connective tissue.

In FSHD the earliest and most severe weakness is in the facial and shoulder girdle muscles. Inability to close the eye completely during sleep that means weakness of the extra-ocular muscles is the facial manifestation[1]. However, recently an increasing number of atypical cases have been reported with sparing facial muscles[6-8]. Here in this case there is no involvement of facial muscles suggesting the subtype of facial sparing.

Scapular winging is prominent, often even in infants[1], as in this case. Flattening or even concavity of the deltoid contour is seen and biceps and triceps muscles are wasted and weak. Muscles of the hip girdle and thigh also eventually loose strength and undergo atrophy[1]. Here though there was involvement of shouldergirdle muscles, later the child developed generalized weakness as well. Gait disturbance is more commonly associated with early onset FSHD[10]. Hence the normal gait can be explained in this case as the age at onset of this patient was 9 years. Cuff hypertrophy is not a feature of FSHD as is in this case. Lumber lordosis and kyphoscoliosis are common complications of axial muscle involvement[1]. This patient was found to have scoliosis to the right owing to weakness of the axial muscles.

Serum levels of CPK and other enzymes vary greatly from normal or near normal to several thousand[1,5]. Here CPK level was found normal which cannot be considered as a point against FSHD. ECG should be performed in every case but is usually normal as it is in this case. EMG is often unhelpful and the diagnosis depends upon the clinical feature, family history and histopathology findings[9].

In hisopathology, extensive proliferation of connective tissue, between muscle fibres, extreme variation in fiber size with many hypertrophic as well as atrophic myofibers and scattered degenerating and regenerating fibers. Mononuclear infiltrates are also found in some cases of the FSHD[11]. Histopathology findings of this case were quite consistent with the above descriptions.

As other muscular dystrophies no specific therapy other than supportive is available so far[1]. Overall long term prognosis is not that discouraging. Life expectancy is normal or near normal with some disability with the upper extremities.

Conclusion

Muscular dystrophy is not uncommon in our region though facio-scapulo-humeral type is relatively rare. Unlike many other types these cases have better outcome and near normal life expectancy. Fairly good number of FSHD-facial sparing type has been reported by authors from different parts of the world. This report would simulate at least some of the clinicians to be suspicious about the diagnosis.

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