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Surgical treatment of dropped head syndrome secondary to fascioscapulohumeral muscle dystrophy: a case report

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Abstract

Fascioscapulohumeral muscular dystrophy is an uncommon hereditary myopathy which affects mainly the muscle of the face and upper limb girdle. We present a rare case with dropped head syndrome as the prominent manifestation of that disease and successfully treated by surgical management. It was a 25-year-old male patient with the chief complaint of neck pain and inability to maintain his horizontal gaze for long periods and as a result he had to quit his job as a shipper. His mother also had signs and symptoms of fascioscapulohumeral muscle dystrophy. Conservative treatment consisting of physical therapy and hard collar was the first attempt in order to reduce the neck pain and had limited result. We then performed a posterior cervical surgery including C2 to T2 instrumentation and kyphotic correction for the patient. The ten-month postop clinical and radiological results were satisfactory and the patient could return to his previous job. Dropped head syndrome with failed conservative treatment can be surgically treated after considering all clinical and radiographic factors.

Background

Facioscapulohumeral muscular dystrophy (FSHD) is a relatively rare hereditary myopathy which was first reported by Duchenne in 1853 and classified by Landouzy and Dejerine in 1886 (therefore also called as Landouzy-Dejérine muscular dystrophy) [1,2]. After myotonic dystrophy and Duchenne muscular dystrophy, it is the third most common muscular dystrophy with an estimated prevalence of 3.2-4.6 per 100,000 population [3]. FSHD is an autosomal-dominant inherit disorder caused by a heterozygous partial deletion of a subset of D4Z4 macrosatellite repeat units on chromosome 4q35, which led to an abnormal expression of the DUX4 retrogene [4]. Although it is often considered a relatively benign dystrophy, to date there are no pharmacological disease-modifying therapies available for FSHD [5]. It is often described as a slowly progressive muscular dystrophy that manifests between age 15 and 30 [6]. As its name suggests, patients suffer from weakness/paralysis of the facial, shoulder and upper arm muscles (fascio-scapulo-humeral) with possible involvement of trunk and leg muscles [6]. In severe forms, patients might encounter significant physical limitations and approximately 20% of cases become wheelchair-bound [4]. While advanced genetic and epigenetic-targeted treatments are recently proposed in ongoing trials, current treatments only aim at increasing muscle strength or managing disease complications [5].

On the other hand, dropped head syndrome (DHS) was first defined in 1986 as the weakness of the neck extensor muscles when working against gravity and might be accompanied by less pronounced weakness of neck flexor muscles [7]. In a review conducted by Kazakov et al., only 4 out of 200 FSHD patients showed mild to moderate weakness of the neck extensor muscles [8]. FSHD is often considered as a rare cause of DHS, which might lead to the under-diagnosis and inadequate treatment of the disease [9].

In this paper, we present the case of a FSHD male patient with significant dropped head syndrome who was underdiagnosed by several neurologic and orthopedic facilities. After spinal fusion surgery, the patient could return to his normal activities with a good quality of life.

Case Presentation

A 25-year-old male patient was admitted to our hospital due to neck muscle fatigue and difficulty in maintaining the horizontal gaze with moderate to severe pain at the cervicothoracic junction radiating to the interscapular area. Those problems had occurred for over 3 years and had been worsening during the period. The initial symptoms were shoulder weakness and inability to raise his arms over 90 degree with slowly progressive muscle atrophy of the upper trunk, noted by the patient more than five years ago. However, given that his mother had a similar condition, he did not seek treatment until three years ago when neck muscles were affected and this prevented him from working as a shipper. He went to several clinics and hospitals but was only diagnosed with non-specific neck pain. The treatment was only oral medications and the condition progressed. There was no history of cervical spine injury or surgery.

Physical examination of the patient revealed mild facial paralysis with the lips and philtrum were deviated to the right side when patient was talking or smiling (Fig 1A). The patient also has winged-scapular sign, positive poly-hill sign (Fig 1B), positive scapular assistance test and positive Beevor's sign. Significant pectoral, deltoid and humeral muscle dystrophy could be observed (Fig 1C). Flexed cervical deformity with flattening of thoracic spine as compensation, which led to the muscle pain in the interscapular area pain as the chief complain. Neck extensor muscle power was 2/5, other muscle groups had a grade 4/5 power. No other relevant signs or symptoms could be detected during physical examination. The patient was diagnosed as facioscapulohumeral muscle dystrophy based on classic clinical presentations [6,10].

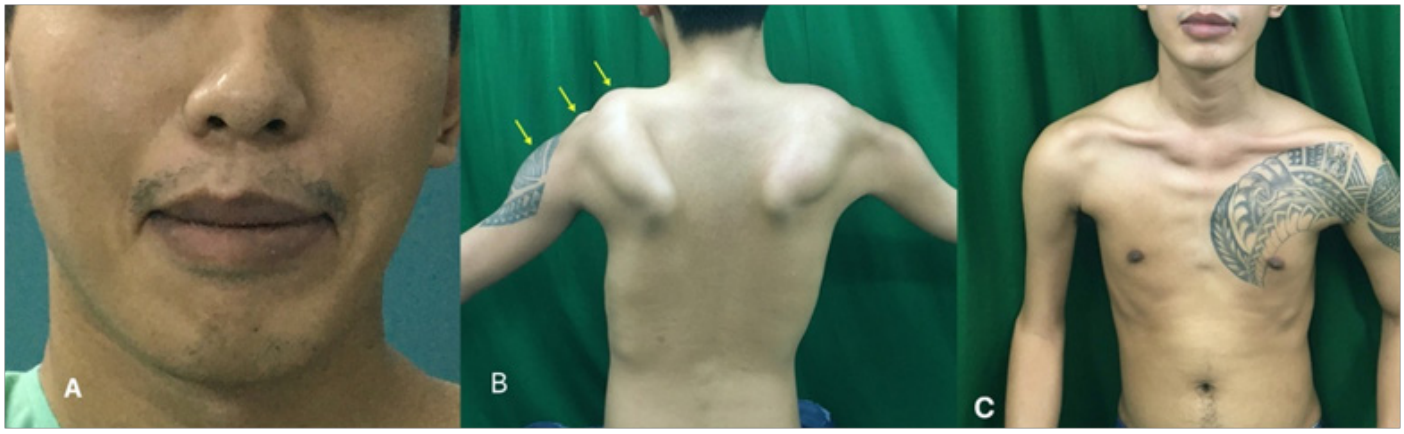

Figure 1: Clinical findings: lips and philtrum deviated to one side when smiling due to asymmetrical fascial muscle weakness (A); winged scapular with poly-hill sign (yellow arrows) (B), deltoid, pectoralis major and biceps muscle atrophy (C).

Investigations

Anterio-posterior and lateral upright radiographs of the entire spine showed spinal deformity including cervical kyphosis, compensated by thoracic hypo-kyphosis and lumbar hyper-lordosis in order to maintain the global sagittal balance (Fig 2A). We also found belly distension due to abdominis rectus paralysis and mild lumbar scoliosis secondary to truncal muscle weakness (Fig 2B). T2 weighted MRI scan depicted the sagging sign of nuchal ligament due to neck muscle atrophy without any spinal cord compressions (Fig 3A). The trapezius shape became a thin line with fatty degeneration and muscle atrophy of the paraspinal muscles could be observed on the T1 weighted axial scan (Fig 3B). Electromyography highly suggested the presence of myopathy. Blood tests showed no sign of inflammation, except two-fold increase of creatinine kinase level above the upper limit value.

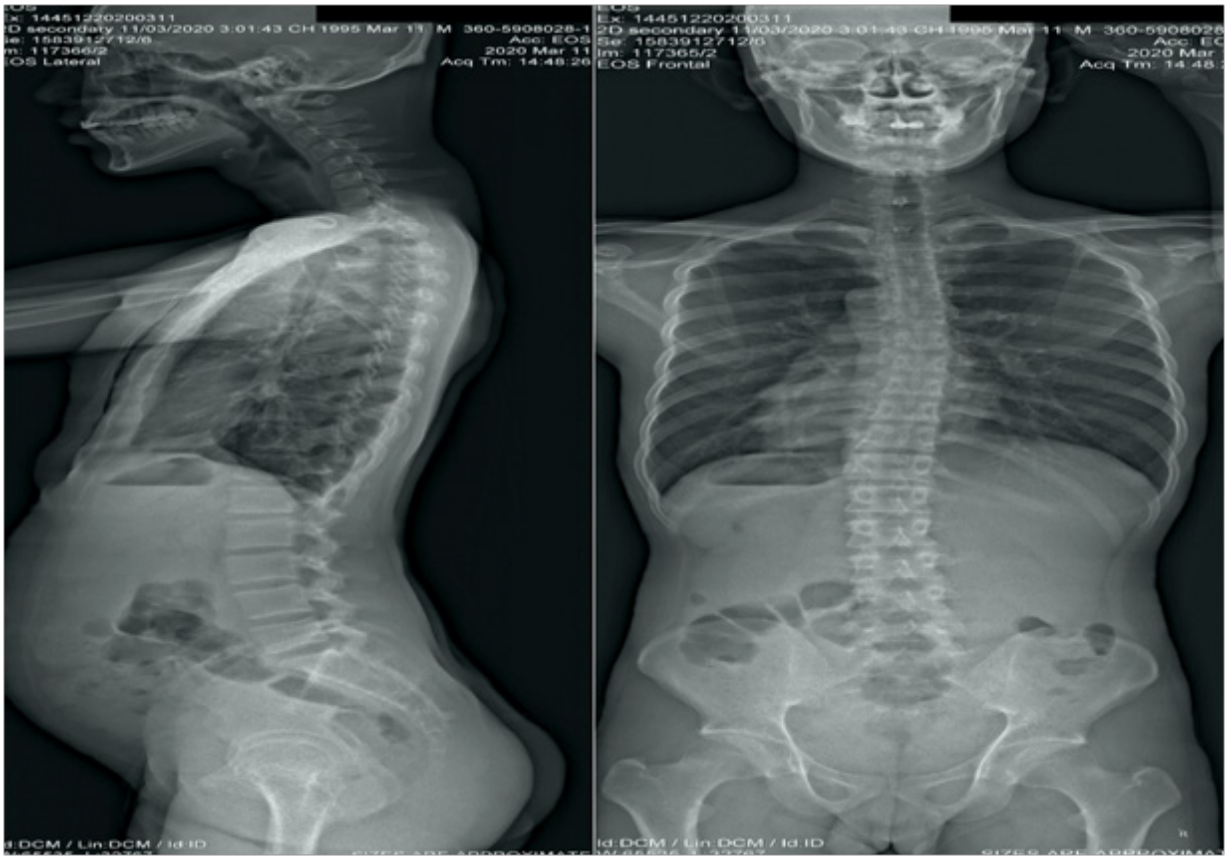

Figure 2: Xray findings: the weakness of the abdominal wall resulted in belly distention and mild scoliosis. Note that due to the flexion deformity of the cervical spine, the thoracic spine has flattened to compensate and maintain the sagittal balance of the whole body.

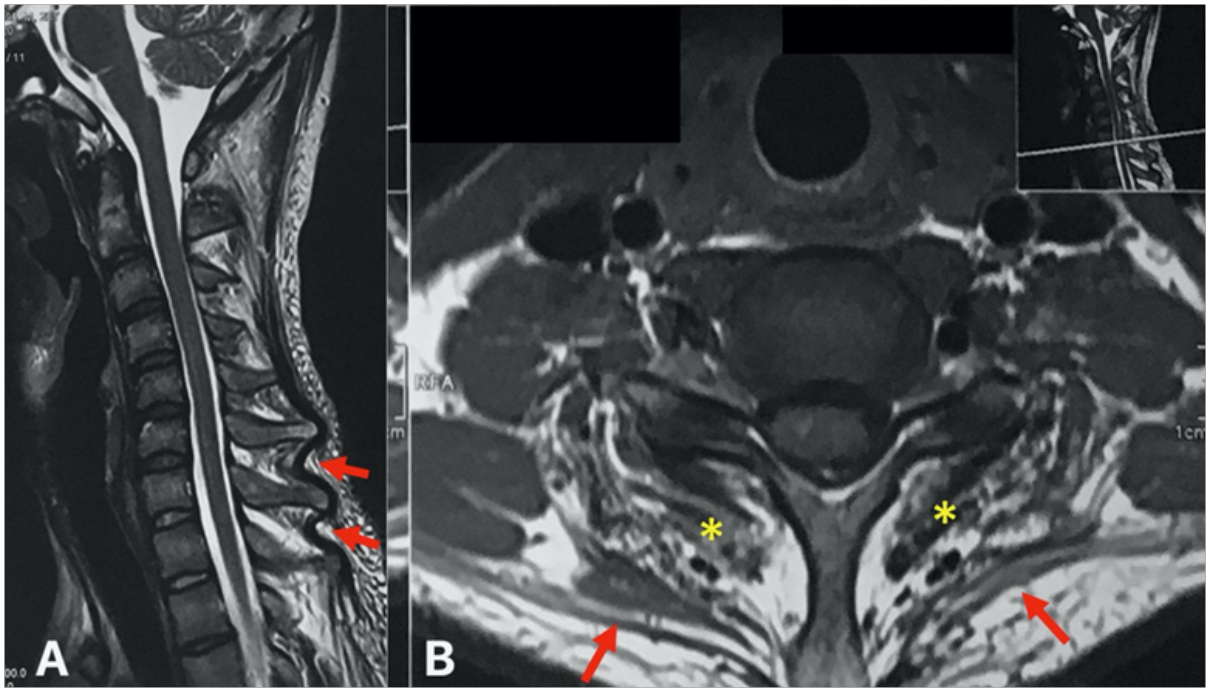

Figure 3: MRI findings: T2 weighted sagittal MRI of this patient revealed the sagging sign of the nuchal ligament (red arrows) (A) and T1 weighted axial MRI revealed fatty infiltration of the cervical multifidus (yellow asterisks) and trapezius (red arrows). Note that the trapezius became a thin line due to muscle dystrophy.

Treatment

Conservative treatment including hard collar and physiotherapy was initially indicated for this patient in six weeks. The hard collar did help him to maintain his head in the upright position and decrease the pain in the interscapular area, however the compliance with cervical collar was poor due to tropical climate in Vietnam. Physiotherapy, however did not have any clear positive effect on the condition. Therefore, corrective surgery was performed in May 2020. Conventional posterior approach was applied with C2 to T2 instrumentation (lateral mass screw fixation from C2 to C7 and pedicle fixation from T1 to T2) with posterior bone graft and no neurological decompression. Hard collar was applied in three weeks post-operatively, continued by physiotherapy in order to partially restore range of motion. After the surgery, full spine xray showed that the entire spine including cervical spine was properly realigned and stabilized in the optimal position, the thoracic spine regained its physiological kyphosis as patient did not have to compensate the sagittal imbalance because of the dropped head condition (Fig 4).

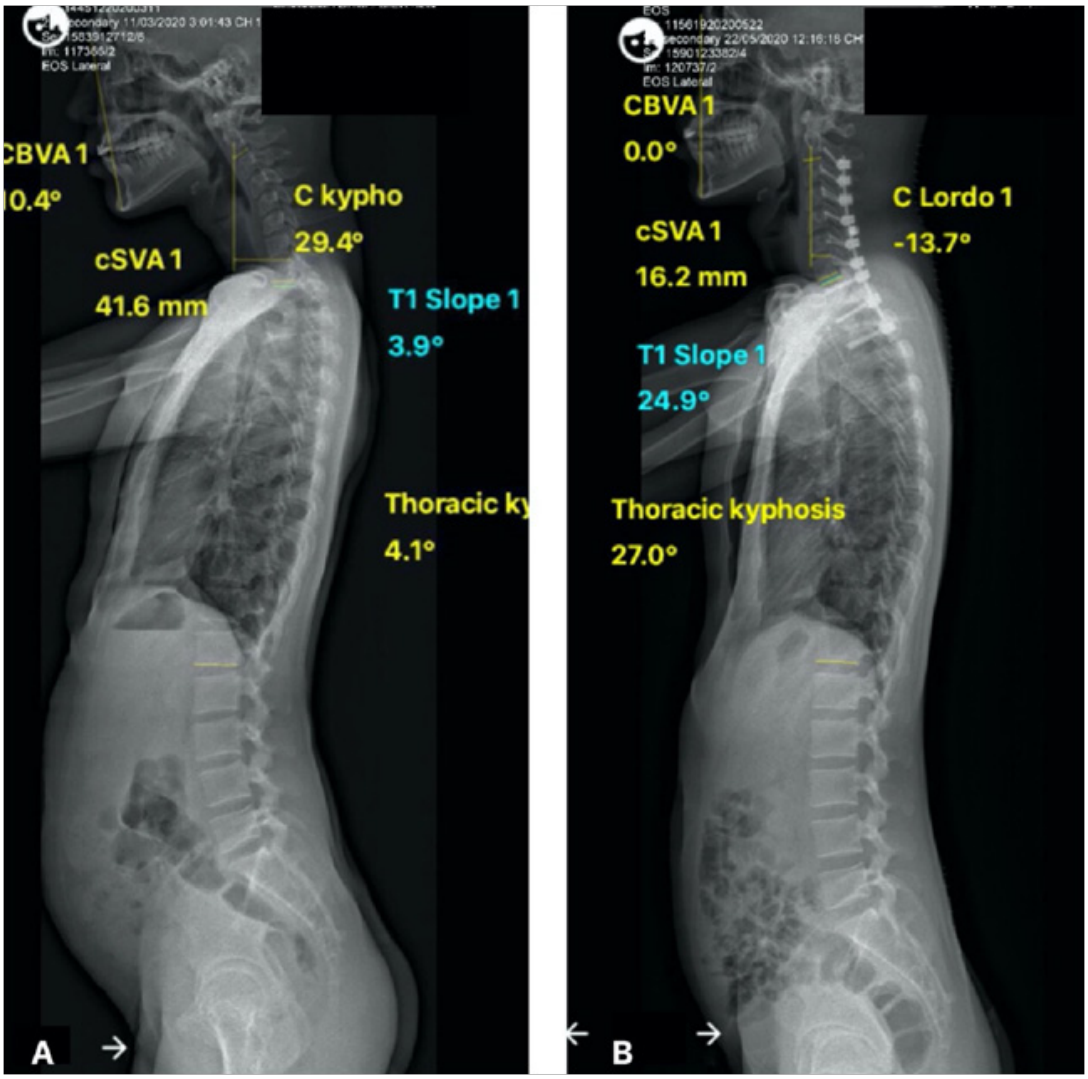

Figure 4: Comparison between preop and postop full spine Xrays: preop Xray (A) with flexed cervical deformity and flattening of thoracic spine as compensation, postoperative Xray (B) with optimal sagittal balance parameters.

Outcome and Follow-Up

The patient was followed up monthly during the first three months, then after every three-month interval. The wound condition, clinical symptoms (neck pain, neck stiffness), neurological symptoms, the shape and range of motion of the cervical spine were monitored at each visit. Cervical Xray was taken to assess the shape of the cervical spine and to detect hardware-related complications, if any. Final Xray (taken 10 months after the operation) did not reveal any correction loss or sign of implant failure (Fig 5). The patient could return to his previous job as a shipper.

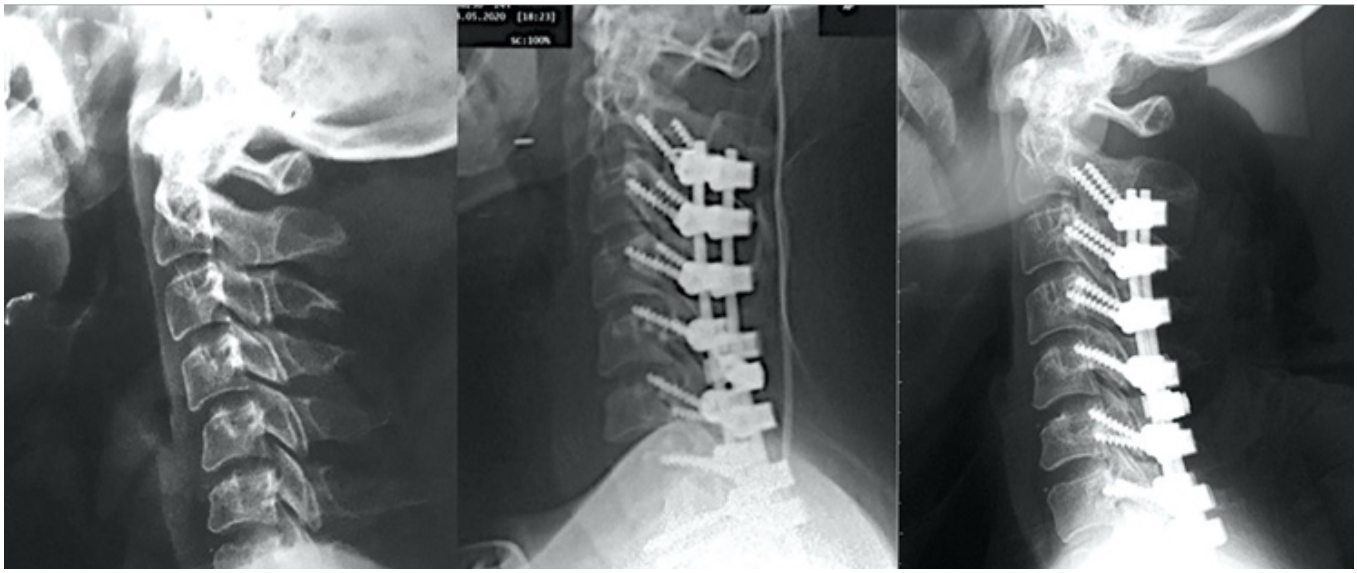

Figure 5: Comparison between preop, immediate postop and final cervical Xrays: preoperative cervical Xray (A) with flexed deformity, immediate postoperative Xray (B) with deformity correction and final cervical Xray (C) with good cervical lordosis and no signs of implant failure.

Discussion

According to the literature, currently there are no specific therapies for FSHD although a developments in the understanding of the pathological mechanisms may lead to therapeutic targets [5]. The current disease management consists of corticosteroid therapy and physiotherapy for muscle strength enhancement. The natural history of this condition is relatively benign as the disease is self-limiting in most cases but fatal evolution with total disability can occur in 20% of patients. Prior to surgery, we tried conservative treatment with a hard cervical collar for six weeks which was helpful but compliance was hindered due to the climate in our tropical country. Physiotherapy with neck extensor strengthening did not have any beneficial effect on the DHS. Based on this we believed that surgery including kyphosis correction and instrumentation was a reasonable option for this patient.

The surgery was carefully planned. Considering his young age, non-smoker, non-diabetic, non-obese patient, we decided that posterior approach alone with instrumentation from C2 to T2 could provide adequate stabilization until bone fusion occurred. Other authors have recommended to extend the instrumentation to the thoracic spine in order to avoid implant failure or junctional instability when stopping at C7 [11-14]. In calculating the amount of correction, we estimated the ideal postop C2-C7 lordotic angle to be 12.5o after considering all the sagittal parameters of our patient (ie T1 slope, thoracic kyphosis and chin-brow vertical angle) [14,15]. The preoperative T2 weighted sagittal MRI revealed no sign of compression of the spinal cord and therefore decompression was not indicated.

During the operation, we also took two samples of muscle for biopsy in two different area: 1. the trapezius sample (for the lower cervical spine area) and 2. the splenius capitis sample (for the upper cervical spine area). The trapezius sample stained with Hematoxylin and Eosin showed diffused muscle atrophy and necrosis with interstitial fibrosis. There was a low level of lymphocyte infiltration that could be detected (Fig 4). The splenius sample, however, was normal and did not reveal any sign of muscular dystrophy. The biopsy result was in line with that of the MRI scan, showing fatty infiltration and atrophy of the trapezius (Fig 6). In a study published in 2015, Gerevini et al with the help of full body MRI scan also confirmed that FSHD mainly affected the trapezius, teres major and serratus anterior [16]. Those findings reinforced our decision to spare the occipito-cervical junction in order to preserve some head motion, as the suboccipital muscles were unaffected.

The immediate post-operative and final Xray revealed good lordotic angle of the cervical spine as planned. Despite a restriction of movement due to instrumentation, our patient was satisfied with the outcome as the horizontal gaze was restored and the neck pain due to muscle fatigue disappeared.

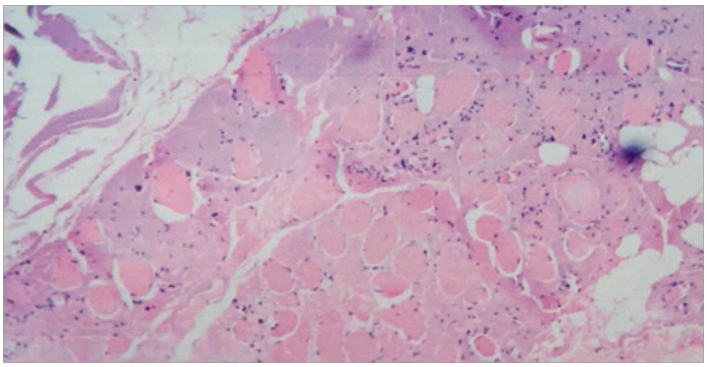

Figure 6: Neck extensor muscle biopsy: the trapezius sample stained with Hematoxylin and Eosin showed diffused muscle atrophy and necrosis with interstitial fibrosis and low-level of lymphocyte infiltration.

Learning Points

Fascioscapulohumeral muscle dystrophy is a rare condition that affect mainly the skeletal muscles of the head, shoulder and arm as its name suggests.

- Dropped head syndrome is an uncommon complication of fascioscapulohumeral muscle dystrophy, which can adversely affect the quality of life of the patient particularly when horizontal gaze is affected.
- When all conservative treatments fail, corrective surgery can be considered a reasonable option to restore the horizontal gaze of dropped head patients.

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