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J. Kaner

J. Zuniga

Robert D. Teasdall

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## The Facioscapulohumeral Syndrome: A Report of Two Cases

J. Kaner, DO,\* J. Zuniga, MD,\* and Robert D. Teasdall, MD\*

*Two patients developed progressive muscle weakness in adult life. The initial diagnosis of polymyositis was made in both cases, but subsequent studies and the distribution of muscle involvement suggested facioscapulohumeral myo-*

*pathy. Other investigators have proposed that this syndrome may result from either genetic or acquired causes. In our patients, the disorder was probably a sequela of polymyositis.*

Several investigators have suggested that facioscapulohumeral myopathy may not be a discrete entity but a syndrome with several causes (1-4). Recently, we have seen two patients for whom an initial diagnosis of polymyositis was made. In both, the clinical features were similar to a facioscapulohumeral myopathy.

### Case Reports

#### Case 1

This woman developed difficulty walking and raising her arms when she was in her early 50s. Over the next ten years the condition worsened until she was unable to climb stairs and could walk only by pushing a wheelchair. Her arms became too weak to lift a walker, and she was unable to feed herself. Paresthesias of the hands and feet were noted. She was diagnosed as diabetic and for 11 years required insulin. A cervical and two lumbar laminectomies had been performed for spondylosis. There was no family history of any similar weakness. Two adult daughters and her granddaughter were studied and found to be normal.

Findings from a muscle biopsy of the left deltoid (Fig. 1) and an electromyogram (EMG) were interpreted to indicate a late stage, inflammatory myopathy. Transaminases, aldolase, lactic dehydrogenase (LDH), and creatine phos-

phokinase (CPK) levels were elevated. Urinary creatine was 879 mg per 24 hours (creatinine 1311 mg). The thyroxine level was 12.2 mg/dl, but the 24-hour thyroid uptake of I<sup>131</sup> was 9%, and a scintigram revealed a normal size thyroid gland with uniformly distributed radio-iodine.

On the basis of the clinical, muscle biopsy, and laboratory findings, the diagnosis of polymyositis was made. The patient received several courses of prednisone, methotrexate, cyclophosphamide, and azothioprine, but her muscle weakness did not improve, and the serum muscle enzymes remained elevated. The paresthesias and EMG pattern of denervation from distal muscles were compatible with polyneuropathy secondary to diabetes mellitus.

The patient was 63 years old when she was first referred for neurological consultation. She had a lumbar lordosis and could not walk unaided. Bilateral facial weakness was noted, and she was unable to maintain lid closure. The sternocleidomastoid muscles were thin and the clavicular heads were absent, but no other cranial nerve deficit was detected. Wasting and weakness of the scapular, shoulder girdle, biceps, and triceps muscles were marked, especially on the right side. She was unable to lift either arm at the shoulder and could barely flex the forearms against gravity. Both brachioradialis and sternocostal portions of the pectoralis major muscles were absent, while the flexor and extensor muscles of the forearms were spared. Although the intrinsic muscles of both hands were wasted, their strength was good. In the lower limbs there was profound weakness of hip extensors, flexors, quadriceps, and hamstring groups. Wasting and weakness were not detected in the distal muscles.

The sensory system was intact. The deep tendon reflexes were absent, while both plantar responses were normal.

\*Department of Neurology, Henry Ford Hospital

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Address reprint requests to Dr. Teasdall, Department of Neurology, Henry Ford Hospital, 2799 W Grand Blvd, Detroit, MI 48202





Fig. 1

**Case 1: Biopsy from left deltoid**  
Early clinical phase of illness; changes compatible with a late stage inflammatory myopathy (X90).

Blood tests relative to the diagnosis of myopathy disclosed elevation of the transaminase and aldolase values. LDH ranged from 412 to 580 U/L. Erythrocyte sedimentation rate (ESR) was 21 mm/hr.

Radiograms of the chest and gastrointestinal tract were negative. Cervical films revealed a surgical fusion from C5 to C7, while lumbosacral films demonstrated evidence of a laminectomy at the L5-S1 joint space. Cine-esophagogram was normal. When the electromyogram was performed, polyphasic potentials of low amplitude with rapid recruitment and full interference pattern were recorded from the deltoid, biceps, and triceps muscles. No denervation potentials were recorded. Conduction velocities of median, ulnar, and peroneal nerves were normal, while distal latencies were slightly prolonged.

A biopsy from the left triceps revealed random changes in the muscle fibers (Fig. 2). Many had a rounded configuration with central nuclei, while others were atrophic and had been replaced by bags of nuclei. Occasional sarcoplasmic floccular changes, phagocytosis by histiocytes, and vacuolations were seen. Adipose tissue was notably increased, while the endomysial connective tissue was only slightly increased. An inflammatory response was not detected. Most of the blood vessels were normal. The histochemical preparations did not indicate group atrophy or type grouping. Both Type I and Type II fibers varied in fiber size and were atrophic.

These biopsy findings were considered to be consistent with a noninflammatory myopathy. Indeed, in view of the conspicuous facial weakness, the proximal weakness of the limbs, the asymmetrical involvement, together with the absences of the clavicular heads of sternocleidomastoid,

sternocostal portions of pectoralis major and brachioradialis muscles, we considered the diagnosis of facioscapulohumeral form of myopathy.

Prednisone was discontinued, but therapy with insulin, lasix, and digoxin was maintained. Over a three-year period, the weakness increased, and the patient became bedridden. She died at age 66.

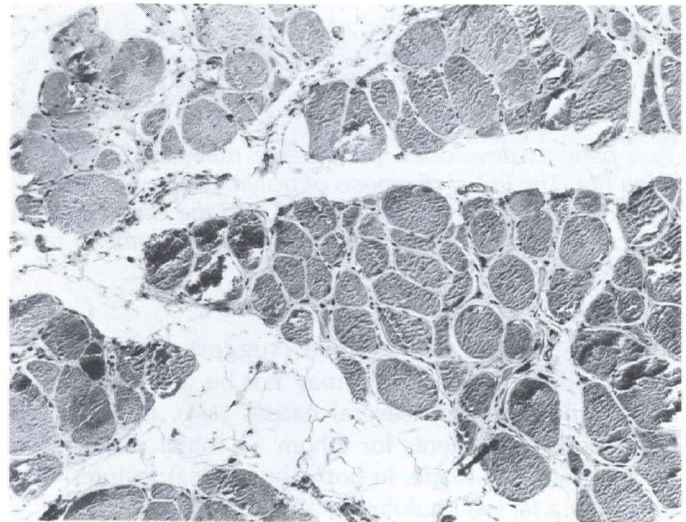


Fig. 2

**Case 1: Biopsy from left triceps**  
Late clinical phase of illness; changes compatible with a noninflammatory myopathy (X150).

## Case 2

This man in his mid 40s began to complain of difficulties in raising his arms and lifting objects with his hands. He developed an unsteady gait, as well as problems rising from chairs and climbing stairs. There was no family history of any similar disorder. Rheumatoid arthritis had been diagnosed several years before the weakness began.

Studies to determine the cause of his muscle weakness disclosed normal values for routine blood determinations, including CPK and transaminases. The LDH was 440 U/L. Thyroid function tests were normal. Median and ulnar nerve conduction was normal, while an electromyogram demonstrated fibrillations in the deltoid and biceps muscles. Voluntary contraction of these muscles produced potentials of short duration but normal amplitude. The interference pattern was slightly reduced. Recordings from the first dorsal interosseous muscle were normal. Based on these findings, but without muscle biopsy, the diagnosis of polymyositis was made and the patient was treated with aspirin and dexamethosone (0.5 mg TID).

At the age of 54, ten years after his weakness began, the patient was first examined by one of the authors. Marked



## Facioscapulohumeral Syndrome

rheumatoid arthritis was evident in the hands. Movements of the head were restricted, and a dorsal kyphosis was apparent. He had a shuffling gait. However, he was alert, with normal speech. The cranial nerves were intact, except for slight bilateral facial weakness. The neck flexors and shoulder elevators were weak, wasting of trapezius and sternocleidomastoid muscles was pronounced, and the clavicular heads of the latter were absent.

The scapular and shoulder girdle muscles were markedly wasted, especially on the left. The patient was unable to lift his arms but had full range of motion of the shoulder joints. The upper portions of both pectoralis major muscles were absent, while the sternocostal portions were present. The brachioradialis muscles were absent. Although wasting was evident throughout the upper limbs, conspicuous weakness was present only in the deltoid and biceps muscles (Figs. 3,4). In the lower limbs, wasting was generalized, but weakness was apparent only in hip flexor and extensor muscles.

The sensory system was intact. The deep tendon reflexes were absent in upper limbs, except for the triceps jerks, while the knee and ankle jerks were present and equal. The plantar responses were normal.

Routine blood studies, the transaminases, aldolase, LDH, and CPK determinations were within normal limits. The ESR was 117 mm/hr. The chest radiograph was normal. Spine films revealed slight anterior displacement of C5 on C6, but neural foramina were patent. There was slight compression of the fourth thoracic vertebral body and mild kypho-scoliosis. Esophageal studies were normal except for extrinsic pressure defects from the cervical spine.

Muscle biopsy from the left triceps (Fig. 5) revealed many hypertrophic fibers and central nuclei. Adipose tissue and necrotic fibers were increased, while mononuclear infiltrates, predominantly of plasma cells, were clustered around muscle fibers and blood vessels. Histochemical studies revealed scattered small fibers of Type I and II with no type grouping.

Electrophysiologic studies revealed normal velocities of median, ulnar, peroneal and tibial nerves. The electromyogram from infraspinati, deltoid, and biceps muscles disclosed a full interference pattern with low amplitude, short duration action potentials. At rest, P waves and fibrillations were recorded. Action potentials recorded from distal muscles of the upper limbs were of larger amplitude and longer duration, but the interference pattern was reduced and denervation persisted. In the lower limbs, potentials from quadriceps, tibialis anterior, and gastrocnemius muscles were increased in amplitude and duration with a marked reduction in interference pattern and some denervation activity.

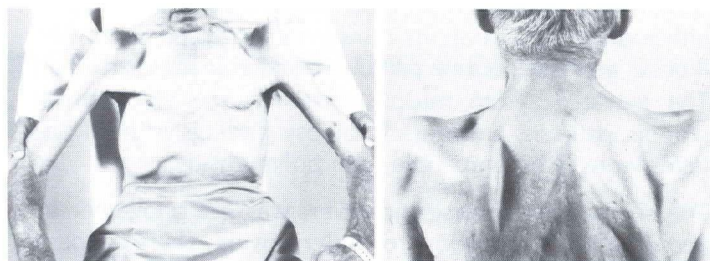


Fig. 3: Left

Case 2

Wasting of shoulder girdles, biceps and absence of clavicular portions of pectoralis major muscles.

Fig. 4: Right

Case 2

Wasting of scapular muscles and trapezii; more profound on left side.

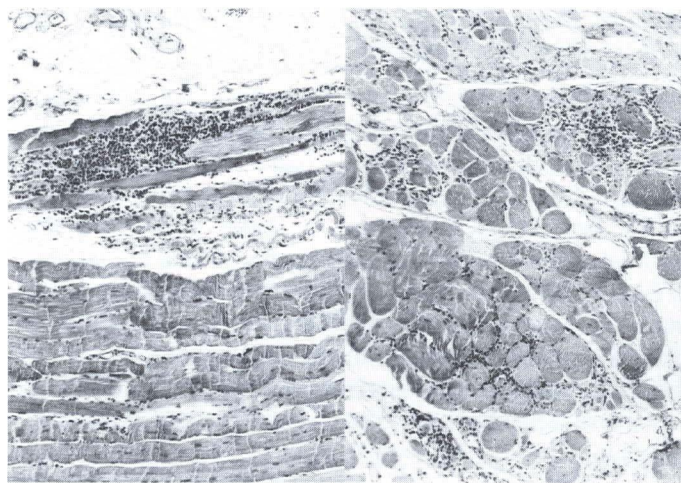


Fig. 5

Case 2: Biopsy from left triceps

Late clinical phase of illness; changes compatible with an active inflammatory myopathy (X90).

In this patient, the distribution of the wasting and weakness was compatible with a facioscapulohumeral myopathy. The muscle biopsy and electromyographic findings support this diagnosis, although the inflammatory changes noted on the muscle biopsy and the denervation pattern suggested the possibility of polymyositis.

## Discussion

Although the presentation of polymyositis may vary, the diagnosis can usually be made on the basis of clinical features (5-7). The clinical characteristics of facioscapulohumeral myopathy have also been defined (8-10).

In our patients, the absence of family history and onset of proximal muscle weakness in middle life supported the diagnosis of polymyositis, and the presence of collagen vascular disease provided further evidence for this diag-



nosis. The first patient had several positive lupus erythematosus preparations during the early phase of her illness, and the second patient had rheumatoid arthritis. The elevated serum muscle enzymes, the electromyographic patterns, and muscle biopsy findings in both patients were also compatible with polymyositis.

The subsequent course, however, was not altogether in accord with the diagnosis of polymyositis. The weakness progressed slowly and failed to respond to steroids. Finally, the conspicuous involvement of facial, scapular, and proximal muscles of the arms and legs prompted the diagnosis of facioscapulohumeral myopathy. Supporting features were the asymmetrical involvement along with the absences of the clavicular heads of sternocleidomastoid and brachioradialis muscles and, in the first case, the sternocostal portions of the pectoralis major muscles. The wasting of the clavicular portions of pectoralis major muscles in the second case was unusual for facioscapulohumeral myopathy.

Conceivably, other processes might have contributed to the clinical disorder. Both patients had cervical spondylosis that could account for involvement of scapular, shoulder girdle, and proximal muscles of upper limbs. The first patient had mild peripheral neuropathy related to diabetes. In both patients, electromyographic recordings from proximal muscles revealed a myopathic pattern, with additional evidence of denervation in the second patient. In spite of the progression of weakness, these findings remained essentially unchanged over several years in both patients.

In contrast, changes in the muscle biopsy findings seemed to depend on the duration of the illness. In the first patient,

the biopsy obtained during the early clinical phase of her disease was interpreted as a late stage, inflammatory myopathy, while the muscle histology five years later was compatible with a noninflammatory myopathy. In the second patient, the muscle biopsy performed ten years after onset disclosed inflammation that consisted primarily of plasma cells. This inflammatory reaction could be a manifestation of the rheumatoid arthritis. The myopathic features, as well as the reduced inflammatory response, might be attributed to the steroid drugs that both patients received (11).

Munsat has suggested that the course of facioscapulohumeral dystrophy may consist of two phases. Initially, the progression of weakness is acute with an accompanying polymyositis, which is followed by a more quiescent phase with myopathic features (4). This sequence seems to have occurred in our first case, while in the second case inflammatory changes in muscle have persisted.

Although a genetically determined facioscapulohumeral dystrophy does exist and sporadic cases of this type possibly do occur, other, acquired cases may well result from myogenic (3,4) and neurogenic disorders (1,2). In our patients, the facioscapulohumeral syndrome appeared to evolve from an inflammatory disease of muscle resembling polymyositis.

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