Chronic spinal muscular atrophy of facioscapulohumeral type

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Summary. Chronic spinal muscular atrophy of FSH type affecting a mother and her son and daughter is reported. The relevant literature is reviewed and the relation between this condition and Kugelberg-Welander (K-W) disease is discussed. Chronic spinal muscular atrophy of FSH type is considered to be a different entity from the eponymous K-W disease. Each type of muscular dystrophy, e.g. limb-girdle, FSH, distal, ocular, or oculopharyngeal type, has its counterpart of nuclear origin. A classification of the chronic spinal muscular atrophies is suggested following the classification of muscular dystrophy.

Chronic spinal muscular atrophies can be defined as a group of chronic disorders, in which the primary defect is degeneration of the anterior horn cells of the spinal cord and sometimes of the bulbar motor nuclei, but with no evidence of peripheral nerve or pyramidal tract involvement.

Since the reports of Wohlfart, Fex, and Eliasson (1955) and Kugelberg and Welander (1956) on chronic spinal muscular atrophy simulating muscular dystrophy, interest has been focused on this group of disorders. Several attempts have been made to classify the chronic spinal muscular atrophies (Becker, 1963; Zellweger et al., 1969; Emery, 1971); however, there is still much confusion.

In this communication familial cases of chronic spinal muscular atrophy of facioscapulohumeral (FSH) type are reported. The relevant literature is reviewed and the relation between our cases and Kugelberg-Welander disease (K-W disease) is discussed. The classification of these clinically and genetically heterogeneous disorders will be presented.

Case reports

Case 1. The proband was a 49-year-old housewife (Fig. 1). From the age of 15 years she suffered from muscular atrophy of shoulders and upper arms. Muscle twitchings were frequently observed. She was diagnosed as FSH muscular dystrophy in another hospital. When she visited us in 1973, she had myopathic facies. She could not close her eyes properly. Muscle atrophy was noted not only in her face, but also in the neck, shoulders, and upper arms bilaterally. The tongue was normal. Bilateral winging of the scapulae was remarkable. She could not raise her arms over her shoulders. Muscle power was almost half of normal in proximal muscles of the upper extremities. Deep tendon reflexes were abolished in the upper extremities, but normally preserved in the lower extremities. Pathological reflexes were not elicited. Sensation was intact. The distribution of atrophic muscles was similar to that in FSH muscular dystrophy, but profuse fasciculations of muscles were seen in the shoulder-girdles.

At biopsy of the left deltoid muscle nearly 10 years ago she was diagnosed as FSH muscular dystrophy in another hospital. However, our electromyographic (EMG) studies showed action potentials of high amplitude and

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Case 1. Proband with muscular atrophy of facioscapulohumeral region and winging of scapulae.

The so-called myogenic patterns were also seen. Serum creatine kinase (CK) activity was normal. Motor nerve conduction velocities of peripheral nerves were normal. These data were suggestive of the degeneration of the anterior horn cells.

Case 2. A daughter of Case 1 was 21 years old (Fig. 3, left). A few years ago she noticed that her face had become thin. Muscles in the neck and shoulder-girdles were also atrophic. The muscle atrophy was slightly progressive. Fasciculation of muscles was observed in the shoulder-girdles. EMG showed neurogenic patterns as well as myogenic. Serum CK was normal.

Case 3. A son of the proband (Case 1) was 17 years old (Fig. 3, right). From the age of 10 years he had suffered from muscular atrophy of face, neck, shoulder-girdles, and recently of pelvic girdles. At the time of our

long duration in various muscles of shoulders and upper extremities as shown in Fig. 2. The so-called myogenic patterns were also seen. Serum creatine kinase (CK) activity was normal. Motor nerve conduction velocities of peripheral nerves were normal. These data were suggestive of the degeneration of the anterior horn cells.

Fig. 1. Case 1. Proband with muscular atrophy of facioscapulohumeral region and winging of scapulae.

Fig. 2. Action potentials of high amplitude and long duration recorded from extensor carpi radialis muscle of the proband. Myogenic pattern was also observed in deltoid muscle.
examination muscular atrophy was noted in the face, neck, shoulder-girdles, and also in the hips and thighs. Lumber lordosis was accentuated. He could not raise his arms at all, and Gowen's sign was present. He was more severely affected than his mother and sister. Profuse fasciculation of muscles was also seen in this patient. Serum CK was normal. EMG showed the same patterns as in his mother.

The diagnosis was chronic spinal muscular atrophy of FSH type affecting the mother and her son and daughter, probably transmitted by an autosomal dominant gene.

**Discussion**

Wohlfart *et al* (1955) described 'hereditary proximal spinal muscular atrophy' and considered the disorder as an entity simulating progressive muscular dystrophy. In 1956 Kugelberg and Welander published an article entitled 'Heredo-familial juvenile muscular atrophy simulating muscular dystrophy'. Thus, K-W disease has been delineated from muscular dystrophy and characterized by spinal muscular atrophy simulating muscular dystrophy, with a slowly progressive course over several decades. However, in all of their cases muscular atrophy simulated the limb girdle type of muscular dystrophy. Though with the increasing number of reports various atypical cases of K-W disease have been described, the cardinal feature of the disease resembles limb-girdle type. In this respect, the present cases were different from the eponymous K-W disease, because muscular atrophy was of the FSH type. However, histological and electrophysiological findings could not distinguish between the two conditions.

Concerning chronic spinal muscular atrophy of FSH type, Mares *et al* (1964) reported a family in which 4 members of 2 generations showed the FSH pattern of muscular atrophy, with evidence of both neurogenic and myogenic changes. In 1967 Fenichel, Emery, and Hunt reported the same disorder affecting a mother and her daughter. We reported similar cases as a variant of K-W disease (Furukawa *et al*, 1969). Recently the same condition was described by some investigators (Patel and Swami, 1969; Igata and Watanabe, 1973; Hosokawa *et al*, 1974).

A search of the earlier papers shows probable cases of this disorder. In 1885 Landouzy and Dejerine collected many cases of FSH muscular dystrophy, and FSH dystrophy is, therefore, called Landouzy-Dejerine type after their names. In their report, a 25-year-old man was described who had a chronic course and remarkable fasciculation of the muscles. "The authors' comment on this case was as follows: 'L'amélioration durant de si longues années, c'est en faveur de la myopathie, mais les contractions fibrillaires des muscles plaident pour la myélopathie (amelioration lasting so many years favours myopathy, but fibrillary contractions of muscles favour myelopathy)." It is worth noting that Landouzy and Dejerine suggested the presence of this type of chronic spinal muscular atrophy as early as in 1885. In 1953, Benassi and Canestrari reported an unusual form of muscular dystrophy of Landouzy-Dejerine type. Muscular atrophy was observed in the face and shoulder-girdles, with remarkable fasciculation of muscles. Tendon reflexes were abolished and the tongue was slightly atrophic.

These reports suggest that chronic spinal muscular atrophy of FSH type constitutes a distinct entity, which is transmitted as an autosomal dominant trait.

In the present family the male patient was more severely affected than his female sib. This phenomenon, a sex-influenced manifestation, is usually observed in K-W disease (Furukawa *et al*, 1968); however, it must await further accumulation of cases to confirm whether this is also the case with this type of chronic spinal muscular atrophy.

According to the Research Group on Neuromuscular Disorders of the World Federation of Neurology (1968), muscular dystrophy is classified as follows: (1) Duchenne (pseudohypertrophic)
Although final classification awaits further investigation of underlying metabolic disorders, an attempt at classification will facilitate the understanding of this group of disorders.

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