Spinal Progressive Muscular Atrophy with Proximal Muscle Involvement.

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INTRODUCTION

Spinal progressive muscular atrophy (SPMA with distal muscle involvement) is not an unusual disease in middle aged people, but proximal muscles are rarely affected in adult SPMA[1-4], which is at times called, the Vulpian Bernhardt type. In 1899, Bernhardt[5] first reported cases of flaccid muscular atrophy with muscle fasciculation and muscle wasting, occurring around the proximal part of the extremities.

Recently, we have found a patient suffering from muscle wasting and weakness chiefly involving the limb girdle muscles, especially around the right shoulder, with the SPMA type of muscle atrophy. The site of involvement seemed to be only the lower motor neuron.

We shall report on this rare condition and its relation to similar disease entities.

CASE REPORT

Patient, K.O., born July 23, 1898, was in excellent health until Nov. 1973, when he first noted a difficulty in elevating his right arm. Six months later, he also noted muscle weakness in the other arm. He had developed progressive difficulty in washing his face and dressing. In the morning, the symptomes were usually relatively better, but physical exercise aggravated the muscle weakness.

He was first seen at the Neurological Department of Syuto Hospital on June 4, 1974. Family history was negative for any neurological or hereditary diseases. Past history was unremarkable, except for an operation of prostate gland hypertrophy in 1971.

PRESENT ILLNESS

Cardinal pathological signs and symptoms were muscle wasting and weakness of the shoulder girdle of the right side predominant. Mainly involved muscles were the deltoid, serratus anterior, teres major, infra and supraspinatus and biceps and triceps humerus muscles (Photo. 1). The muscles of the thenar and hypothenar were also slightly weak and atrophic (Photo. 2). The muscles of the forearms were relatively spared, so that the patient's grasping power was well preserved. He was unable to hold his hands up for an extended period of time. In the muscle group showing marked atrophy, there were frequent muscle fasciculations, indicating lower motor neuron injury or anterior horn cell damage. The tongue muscle also showed minimal fasciculation, but was not atrophic, and its motility was uninvolved. He showed no sings of bulbar involvement. Intrinsic muscles of the hands showed moderate muscle wasting and his grasping power was 15 kilograms on the right and 18 on the left on June 25, 1974. Circumference of the extremities were: 33.7 cm (right thigh), 33.9 (left thigh), 21.5(right forearm), 21.9(left forearm), 20.5(right upper arm), and 21.7(left upper arm).

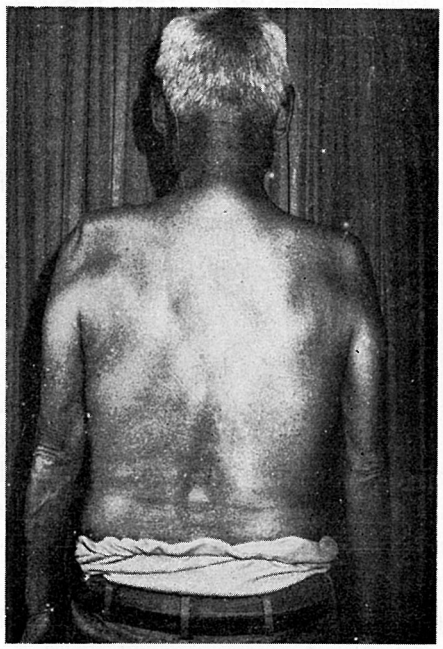

Photo. 1

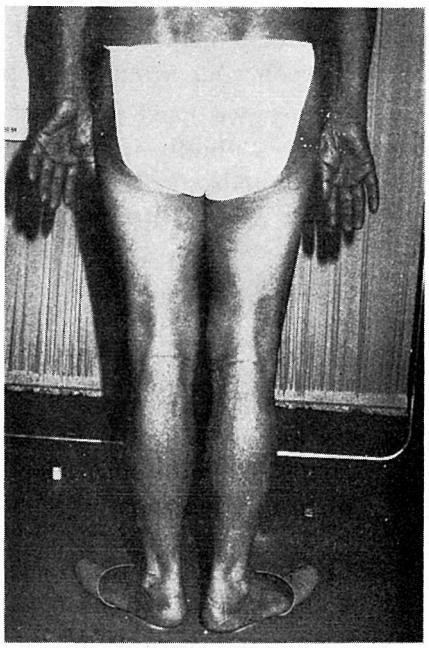

Photo. 2

Photo. 1. Muscle wasting chiefly involving shoulder girdle and upper arm.
Photo. 2. Relative preservation of muscles in forearm, thenar and lower extremity.

There was no evidence of any disturbance in tactile sensation and vibration sense. Abdominal skin reflex was active on both sides. Deep tendon reflexes were markedly hypoactive and symmetrical in each extremity, except for the Achilles tendon reflex, which was relatively active. At the time he was first seen, there was no complaint of muscle weakness in his lower extremities or gait disturbance.

Because of relative sparing of the power of his forearms, he could drive his motor bike when coming to our clinic, but he could not raise his arms, chopsticks, or a wet towel for a long period of time. A pathological reflex was not elicited anywhere in the body. The patient had had high blood pressure, maximal up to 220 mmHg.

Laboratory Examinations:

There was minimal abnormality in serum protein electrophoretic pattern. There were slight increases in alpha 1 and alpha 2 globulins and a slight decrease in beta globulin, and a slight increase in lactic dehydrogenase and creatine phosphokinase activity.

Electromyographic examination revealed a neurogenic change in the affected muscles of the shoulder girdle but unremarkable results in the tongue muscles. X-ray examinations of the cervical spine showed no pathological changes except for the usual aging changes.

Course (Fig. 1) :

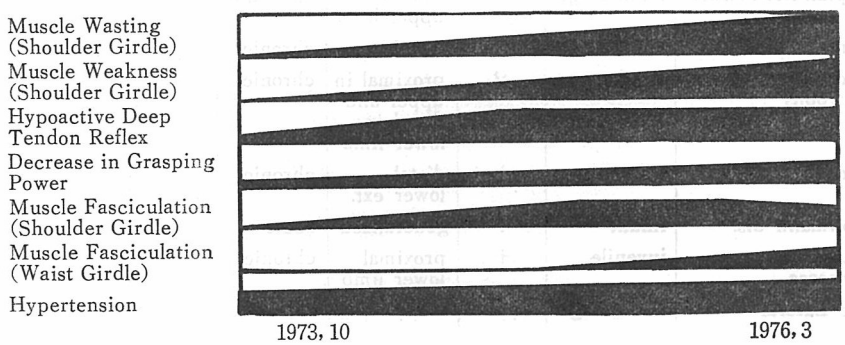

Fig. 1 Pt., K.O. 71 year-old male, SPMA

In spite of oral administration of anabolic steroids, adenosine triphosphate, vitamin B and E derivatives and thiazide for hypertension, the symptomes have been progressively aggravated. Physical therapy, including massage and traction of the cervical spine, was also not beneficial. At the beginning of 1976, deep tendon reflex was only weakly elicited in the Achilles tendon. Muscle fasciculation became weak in the upper

extremities, and more marked in the thigh and waist girdle muscles, although the muscle wasting was not marked in these areas (Photo. 2). He then complained of muscle weakness in the thigh and lumbar region. His grasping power was 7 kg on the right and 8 on the left, which was decreased by one-half compared with that of 20 months before.

COMMENT

For convenience in differential diagnosis, we made a list of relating disorders (Table 1). The table only shows that exact diagnosis is difficult and still obscure. The situation of the proximal type of SPMA seems to be an intermediate condition between Aran's disease[6] and amyotrophic lateral sclerosis (ALS). Considering its progressivity, the prognosis of SPMA in this case, seems to be serious. Differential points of symptoms and signs of ALS are the presense of pyramidal signs and hyperactivity following upper neurone involvement[7]. In this case, the thenar and hypothenar msucles were relatively spared, however, they were obviously damaged.

Table 1.

Differential Diagnosis	Age of Onset	Heredity	Site of Atrophy	Course	Fasciculation	Pyramidal Sign	Sensory Disturbance
SPMA (Vulpian-Bernhardt)	middle age	?	proximal upper limb	subacute	+	−	−
SPMA (Aran-Duchenne)	middle age	?	distal	chronic	+	−	−
Scapulo-Peroneal Muscular Atrophy	middle age	?	proximal in upper and distal in lower limb	chronic	+	−	+
Charcot-Marie-Tooth dis.	adlescent	+	distal lower ext.	chronic	+	−	+
Werdnig-Hoffmann dis.	infant	+	generalized	acute	−	−	−
Wohrfart-Kugelberg Welander Disease	juvenile	+	proximal lower limb	chronic	+	−	−
Amyotrophic Lateral Sclerosis	middle age	−	distal	subacute	+	+	−
Progressive Muscular Dystrophy	juvenile	+	proximal	chronic	−	−	−

In attempting to label this case, a question arises as to whether SPMA and ALS belong to quite another category of disease, or not. Unexceptionally, ALS shows exaggerated deep tendon reflexes.

SPMA was first described by Aran in 1890[8]. He differentiated SPMA from ALS. Among his 11 cases, proximal flaccid paralysis was

observed in case 3, whose disease was mainly in the shoulder girdle, and was interpreted as ALS retrospectively, a few years later.

From the clinical standpoint, differential diagnosis of the motor neurone disease and its relating disorders still has many obscurities[9]. Müller[3] had reported the fact that the progressive motor neurone disease in adults is usually interpreted as a beginning form of ALS. He described that from 44 cases of SPMA, only 6 were still diagnosed as SPMA after several years. Flaccid paralysis in the proximal muscle in association with muscle fasciculation, called the Vulpian-Bernhardt type of SPMA, seemed to be extremely rare. There is an opinion that SPMA is only an incomplete form of ALS, or a stage prior to the developement of upper motor lesions[10]. In this sense, Aran[8] once claimed that SPMA is a prototype of motor neuron disease in comparison with ALS, which is thought to be the secondary type of motor neuron disease[1].

Proximal neurogenic flaccid atrophy, is a characteristic of Wohlfart-Kugelberg-Wellander's disease (WKW), which occurs in infants, and has a hereditary tendency. However, the adult type, with a non-hereditary tendency, does exist. (Smith et al.[11], Gross[1]) There is an opinion that these are only a subtype of spinal muscular atrophy. (Norris et al.,[6])

In short, the more cases of the intermediate form of motor neuron disease that are reported, the more possible accurate differential diagnosis will become. How could we know the prognosis when we confront a patient with SPMA?

Smith et al.[11] reported the condition of a patient with proximal flaccid paralysis and fasciculation as a WKW disease. No family history for neurological disease was detected. They discuss 2 types of purely motor system disease with anterior horn cell involvement, i.e., Werdnig-Hoffman disease (WH), WKW disease and a proximal type of ALS. They also said that ALS, with the onset of proximal limb weakness early in its course, may be difficult to differentiate from the WKW disease, especially in those who have no family history of the disease, a more prolonged course, and no signs of either bulbar or cortico-spinal tract involvement. In our case, none of the upper motor neuron system was involved, as far as we had examined. Their interpretation is as follows: WKW disease is tentatively classified, together with WH and Duchenne-Aran disease, as a degenerative disease of the anterior horn cell, but with a much more favourable and benign course. Brownell et al.[12], reported the neuropathological findings of the motor neuron disease. They concluded that the motor neuron disease constitutes an ill-defined band in a broad spectrum of multiple system atrophies, i.e., one of the

subacute encephalomyelopathies. They found only unspecific changes with degeneration, loss of nerve cells, nerve fiber tract degeneration and gliosis. They were opposed to the invention of new diagnostic labels for such nonconfirmed cases. The labelling of diseases of unknown etiology is a matter of practical convenience, and the invention of new labels would only be justified if, and when, particular causal agents were identified.

Still the question remains open as to what is the deciding factor on how the involvement occurs in proximal, or distal muscle groups, and how the primary, the secondary, or both, neurons are affected.

The collection and precise analysis of the symptoms and signs of borderline cases will be essential for solving these problems. These classifications tentatively seem to be useful for evaluation of prognosis, but not for treatment.

More extensive study for detecting the pathogenesis of these diseases will be requested.

SUMMARY

A case of the Vulpian Bernhardt type of spinal progressive muscular atrophy was reported. The patient was 65-year-old male, who developed progressive difficulty in washing his face and dressing because of weakness of shoulder girdle muscles.

Physical examination revealed muscle weakness and wasting in the proximal part of the upper extremities. The muscles of the forearms were relatively spared. The affected muscls showed muscle fasciculation and hypoactive deep reflexes. His family history was completely negative for any neurological diseases. The mainly involved site was suspected to be the anterior horn cell or secondary motor neuron in the spinal cord.

This case was interpreted as an intermediate form between spinal progressive muscular atrophy, proximal type of amyotrophic lateral sclerosis and adult type of Wohlfart-Kugelberg-Wellander's disease.

The problems in differential diagnosis of these motor neuron diseases were discussed.

REFERENCES

1) Gross, M.: Proximal spinal muscular atrophy. *J. Neurol. Neurosurg. Psychiat.*, 29 : 29–34, 1966.
2) Hurwitz, L., Lapresle, J. et Garcin, R.: Atrophie musculaire neurogine de topographie proximale et symetrique simulant une myopathie. Etude de deux observatione. *Revue Neurol.*, 104 : 97–107, 1961.

3) Müller, R.: Progressive motor neurone disease in adults. *Acta Psychiat. Neurol.*, 27 : 137–156, 1952.
4) Namba, R., Aberfield, D.C. and Grob, D.: Chronic proximal spinal muscular atrophy. *J. Neurol. Sci.*, 11 : 401–423, 1970.
5) Bernhardt, M.: Über eine hereditäre Form der progressiven spinalen mit Bulbarparalyse complizierten Muskelatrophie. *Arch. Path. Anat. Phys. Klin. Med.*, 115 : 197–216, 1889.
6) Norris, Jr. F.H. and Farbes, H.: Adult spinal motor neurone disease. Progressive muscular atrophy (Aran's disease) in relation to amyotrophic lateral sclerosis. In *Hdb. Clin. Neurol.*, Vol. 22, North Holland Pub. Co., Amsterdam, 1968. p. 1.
7) Metcalf, C.W. and Hirano, A.: Amyotrophic lateral sclerosis. *Arch. Neurol.*, 24 : 518–523, 1971.
8) Aran, F.A.: Recherches sur une maladie non encore decrite du systeme musculaire (atrophie musculaire progressive). *Arch. Gen. Med.*, 24 : 15–35, 1850.
9) Friedman, A.P. and Freedman, D.: Amyotrophic lateral sclerosis. *J. Nerv. Ment. Dis.*, 3 : 1–18, 1950.
10) Shindo, M., Tsukagoshi, H.: Historical review of the concept of amyotrophic lateral sclerosis. *Adv. Neurol. Sci.*, 21 : 281–292, 1977. (in Japanese)
11) Smith, J.B. and Patel, A.: Wohlfart-Kugelberg-Wellander disease. *Neurology*, 15 : 469–473, 1965.
12) Brownell, B., Oppenheimer, D.R. and Hughes, J.T.: The central nervous system in motor neurone disease. *J. Neurol. Neurosurg. Psychiat.*, 33 : 338–357, 1970.