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Case report

A case of bulbospinal muscular atrophy with large fasciculation manifesting as spinal myoclonus

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ABSTRACT

Objective: This paper reports a patient with bulbospinal muscular atrophy (BSMA) who presented with spinal myoclonus, documented by video and surface electromyography.
Case report: A 66-year-old man had progressive gait disturbance, dysphagia, and easy fatigability of all extremities over a period of 4 years. Neurologically, muscle atrophy, fasciculation, and weakness were observed in the bulbar and limb muscles. When the knees were kept in mild flexion in the supine position, fasciculation of the thigh adductor muscles was so large that it caused shock-like involuntary movements of the legs, corresponding to spinal myoclonus. A genetic test revealed 41 repeats of CAG in the androgen receptor gene, and the diagnosis of BSMA was made.
Significance: The present case suggests that extremely large fasciculation can cause spinal myoclonus.

1. Introduction

Fasciculation is clinically characterized by random and spontaneous twitches of muscle fibers and is commonly observed in patients with involvement of the lower motor neuron. Fasciculation is usually not large enough to be recognized as joint movements, but if it involves small muscles such as the hand intrinsic muscles, it may be recognized as finger movements (Denny-Brown and Pennybacker, 1938; Kimura, 2001; Shibasaki and Hallett, 2016). In a study of fasciculation by using surface electromyography (EMG), fasciculation potentials were of relatively high amplitude (>600 μV), usually formed of two or more phases, and fired at low rates with irregular intervals ranging from less than 1 s to more than 1 min (Howard and Murray, 1992).

Myoclonus is defined as shock-like involuntary movements of face and extremities and classified into cortical, brainstem, and spinal myoclonus according to its estimated site of origin (Fahn, 2005; Shibasaki and Hallett, 2016). Myoclonic jerks of cortical origin are characterized by EMG correlates of short duration, usually less than 50 ms, whereas those of subcortical origin have EMG correlates of a longer duration, with the exception of reticular reflex myoclonus. Cortical myoclonus usually involves multiple muscles, either independently or concurrently, and commonly involves the agonist and antagonist muscles synchronously. In some cases, multichannel EMG recording from an extremity can demonstrate the spread of jerks from the proximal to the distal muscles with the conduction velocity approximately corresponding to that of the alpha motor fibers. Cortical myoclonus usually occurs at irregular intervals, but it may appear rhythmic, particularly when it repeats at high frequency (Shibasaki, 2000; Shibasaki and Hallett, 2016).

So far, little attention has been paid to the clinical manifestation of fasciculation. The primary aim of this case report is to draw attention of neurologists and clinical neurophysiologists to the fact that large fasciculation can clinically manifest itself as spinal myoclonus.

In our patient with bulbospinal muscular atrophy (BSMA), fasciculation was so large that it presented as spinal myoclonus.

2. Case report

A 66-year-old man was admitted because of progressive gait disturbance, special difficulty in climbing upstairs, dysphagia, and easy fatigability of all extremities over a period of 4 years. There was no parental consanguinity and no similar disease in

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the family. The patient's monozygotic twin was reported to be asymptomatic. On general physical examination, gynecomastia was not observed. Neurologically, muscle atrophy of moderate degree and fasciculation were observed in the orbicularis oris muscles and the tongue. There was dysphagia, and voice was nasal and hoarse. There were muscle atrophy and weakness in the neck, chest, proximal part of the upper extremities, hips, and distal lower extremities with some asymmetry.

When the knees were kept in mild flexion in the supine position, shock-like involuntary movements were noted in the thigh adductor muscles (see the Video). The involuntary movements were observed bilaterally and irregularly but were not precisely synchronous between the two sides, and there was no simultaneous contraction of the antagonist muscles. Moreover, the movements were not stimulus sensitive. The muscle twitches in the upper extremities were so small that they did not cause any limb movement. Muscle tone was decreased, and the tendon reflexes were hypoactive throughout. There was no Babinski sign, and sensory function was normal. Gowers sign was mildly positive, but gait was normal. We did not prescribe any drug for the involuntary movements because they did not bother the patient's daily life at least in the resting condition.

In the laboratory test, creatine kinase in the serum was 628 IU/L (normal range: 45–163 IU/L). Testosterone was 6.7 ng/mL in the blood and 31 μg/day in the urine (normal range: 1.9–8.8 ng/mL and 13–160 μg/day, respectively). Genetic test for BSMA showed 41 repeats of CAG in the androgen receptor gene (normal range: 21–26 repeats). Body CT scan showed no testicular atrophy.

Electrophysiologically, motor nerve conduction study of the right upper and lower extremities showed normal conduction velocities. The compound muscle action potentials (CMAPs) of the median nerve and the ulnar nerve both with wrist stimulation were of low amplitude (4.4 and 6.2 mV, respectively). CMAP of the tibial nerve with ankle stimulation was normal (12.6 mV). Sensory nerve conduction study of the right sural nerve showed slightly decreased conduction velocity (37 m/s) with sensory nerve action potential of low amplitude (4.6 μV). Somatosensory evoked potential (SEP) with median nerve stimulation showed low amplitude of the early cortical components. Cortical SEP with tibial nerve stimulation was absent. Needle EMG revealed fibrillation potentials and positive sharp waves in the right deltoid and the right tibialis anterior muscles and polyphasic potentials and high amplitude potentials in the tongue. Muscles involved by the shock-like contraction were not studied by the needle EMG to avoid injury.

The surface EMG of the thigh muscles at rest showed occasional small discharges of different waveforms corresponding to fasciculation (Fig. 1). During continuous and gentle muscle contraction, irregular and repetitive discharges of small amplitude, corresponding to fasciculation, and discharges of large amplitude, corresponding to shock-like involuntary movements, were superimposed on the discharges associated with voluntary muscle contraction. After cessation of the voluntary muscle contraction, the involuntary EMG discharges became more distinguishable from the discharges associated with voluntary muscle contraction and remained for a few seconds (Fig. 1).

3. Discussion

The patient reported in this study showed clinical signs indicative of the involvement of the lower motor neuron but not of the upper motor neuron. The needle EMG showed evidence of acute and chronic denervation. The genetic test for BSMA revealed repeats of CAG in the androgen receptor gene. From these results, the diagnosis of BSMA was made (Kennedy et al., 1968; Sobue, 1995).

As seen in the video, our patient showed brisk and gross contractions in the thigh muscles when the knees were kept in flexion in the supine position. Co-contraction of agonist and antagonist muscles was not observed. Some movements were large enough to produce joint movements and appeared shock-like, resembling myoclonus. The surface EMG showed occasional discharges corresponding to fasciculation at rest, which became larger and more frequent during and after voluntary muscle contraction. The duration of the discharges was shorter than 100 ms. The clinical and electrophysiological features of the involuntary movements fulfilled the criteria of myoclonus (Shibasaki, 2000; Shibasaki and Hallett, 2016).

Regarding the source of myoclonus in the present case, involvement of an isolated muscle, lack of co-contraction of agonist and antagonist muscles, and absence of giant SEP suggested myoclonus of spinal origin rather than of cortical origin (Shibasaki, 2000; Shibasaki and Hallett, 2016). Spinal myoclonus is classified into spinal segmental myoclonus and propriospinal myoclonus. Spinal segmental myoclonus is usually symptomatic of a localized lesion or dysfunction of the spinal cord (Hopkins and Michael, 1974; Jankovic and Pardo, 1986). Propriospinal myoclonus is believed to involve propriospinal pathways, in which the myoclonic jerk

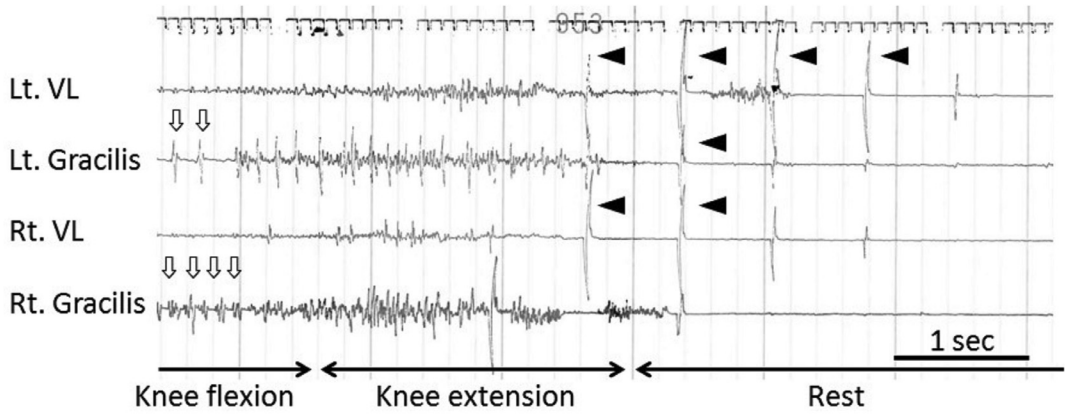

Fig. 1. Surface EMG records of leg muscles during and after voluntary muscle contraction. Note the repetitive EMG discharges of small amplitude, corresponding to fasciculation (white arrow), in the gracilis muscles bilaterally during knee flexion and in the right vastus lateralis (Rt. VL) muscle during knee extension. Immediately following the end of the knee movement, EMG discharges of large amplitude appeared in the VL muscles bilaterally and in the left gracilis muscle and remained for a few seconds, which corresponded to myoclonus (black arrowhead).

starts from a certain thoracic segment and spreads rostrally and caudally with relatively slow conduction velocity (Brown et al., 1991, 1994). The distribution of myoclonus in our patient was compatible with that of spinal segmental myoclonus. As the myoclonic movements were restricted to the thigh muscles, it may be explained as spinal myoclonus originating from the lumbar segments.

In our patient, muscle contractions of various magnitudes were observed in the lips and extremities. Some of them were twitches involving a part of a muscle, which were typical fasciculation, and those of the thigh muscles were large enough to cause joint movements, resembling myoclonus. In fact, many contractions were in between fasciculation and myoclonus in terms of their magnitude. Furthermore, small contractions corresponding to fasciculation were enhanced and became larger and more frequent by volitional contraction of the corresponding muscles. Therefore, it is reasonable to postulate that the spinal myoclonus in this patient can be interpreted as enhanced fasciculation of anterior horn cell origin, and thus, it may be concluded that large fasciculation can be a cause of spinal myoclonus.

Conflict of interest

The authors declare that they have no conflicts of interest and nothing to disclose.

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Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at http://dx.doi.org/10.1016/j.cnp.2017.01.001.

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