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Action myoclonus induced by visually guided movement

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Abstract In order to determine what kind of voluntary movement induces action myoclonus, we gave two siblings with sialidosis two kinds of tasks. When the patients were asked to move their index fingers following a smoothly moving target, action myoclonus became prominent. In contrast, when they were asked to perform the same movements with their eyes closed, they could move their index fingers very smoothly. This shows that action myoclonus was induced by visually guided movement, but not by self-paced movement. Our observations might reflect a disorder of the cerebellum, which controls visually guided movement.

Key words Action myoclonus
Visually guided movement
Sialidosis

Introduction

Since Lance and Adams [6] first described action or intention myoclonus in posthypoxic encephalopathy, it has been described in other conditions [2, 4, 7, 10]. Action myoclonus is characterized by its induction by voluntary movement. What type of voluntary movement induces action myoclonus, however, has not yet been established.

We report here the cases of two siblings with sialidosis (type 1), whose chief complaints were action myoclonus. In order to specify what type of voluntary movement induces action myoclonus, we gave two kinds of tasks involving sensory guided movement and self-paced movement [3], and observed which task induced action myoclonus more prominently.

Case reports

Case 1

A 24-year-old Japanese female had been healthy until the age of 15 years, when she began to notice unsteadiness in walking. At the age of 16, she suffered from generalized seizures with loss of consciousness. After this, irregular jerky movements of the extremities occurred when she attempted to make certain movements, and she noticed dysarthria. She could not walk unassisted by the age of 17. These symptoms progressed slowly, despite medications such as clonazepam, phenobarbital, and sodium valproate.

Physical findings

She had macular cherry-red spots. Involuntary movements were not observed at rest. However, on attempting to make certain movements, there were irregular jerky movements in the extremities. They occurred mainly when she made voluntary movements under visual guidance. For instance, it was very difficult for her to grasp a cup on a desk while watching it, because of jerky movements. But she could easily grasp it with her eyes closed, without jerky movements. These jerky movements were not activated by sensory stimulation such as sound and light. She had scanning speech. Dysmetria was not prominent. Intelligence, muscle tone, muscle strength, reflexes, and sensation were normal.

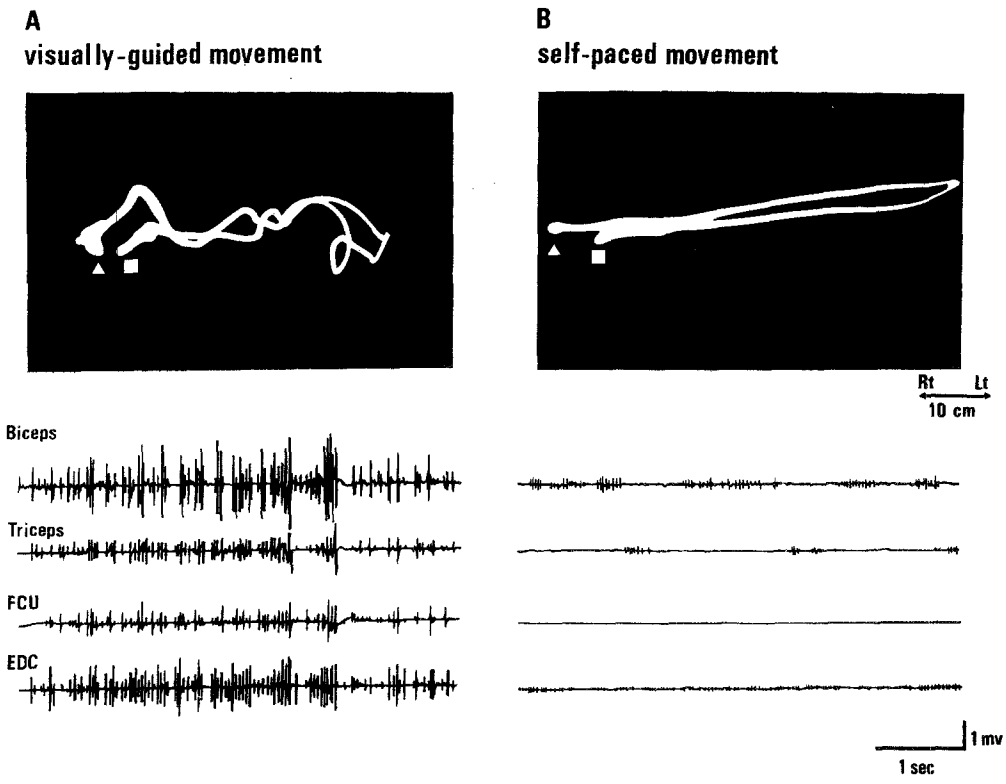

Fig. 1 The traces of the index finger (*upper*) and surface electromyography (*lower*) during visually guided movement (**A**) and self-paced movement (**B**), in case 1. The trace was recorded in one cycle of the movement. The *triangle* shows the beginning of the movement, and the *square* shows the terminal of the movement in one cycle. The mediolateral direction of the movement is shown. Surface electromyography was continuously recorded on biceps brachii (*Biceps*), triceps brachii (*Triceps*), flexor carpi ulnaris (*FCU*) and extensor digitorum communis (*EDC*)

Laboratory data

Surface electromyography (EMG) showed irregular discharges, which were synchronized between the antagonistic muscles (Fig. 1 A). α-Neuraminidase activity in leucocytes was reduced (0.18 nmol/h per mg protein; normal range 1–2 nmol/h per mg protein), whereas β-galactosidase activity in leucocytes was preserved (98.2 nmol/h per mg protein; normal range 90–200 nmol/h per mg protein). Cranial magnetic resonance imaging was normal. Electroencephalography showed frequent diffuse multiple spike-and-slow wave complexes. Somatosensory evoked potentials by stimulation of the median nerve showed a large potential with middle latency (N_{20}–P_{25} 20.4 μV, P_{25}–N_{30} 30.8 μV).

Case 2

A 22-year-old Japanese male, the younger brother of case 1, had been healthy until the age of 17 years, when he began to notice unsteadiness in walking. At the age of 19, he suffered from generalized seizures with loss of consciousness. This unsteady walking progressed slowly, despite medication with clonazepam. At the age of 21, he noticed irregular jerky movements in the upper extremities when he attempted to use them.

Physical findings and laboratory data were essentially the same as his sister's, except that dysmetria was more prominent than jerky movements.

Clinical diagnosis

Both of these siblings had macular cherry-red spots and progressive jerky movements, induced by action, in the extremities. Enzymological studies confirmed the clinical diagnosis of sialidosis type 1 [8].

Methods

We gave the patients two kinds of tasks and observed how action myoclonus was different. The patients were seated on a chair in a dimly lit quiet room. First, the patients were asked to move their index fingers following a target, at shoulder height, with the elbow fixed at about 90°. The examiner moved the target horizontally in front of the patients with a range of about 60 cm and a cycle of about 2 s. Second, the patients were asked to mimic the same movements as closely as possible with their eyes closed.

We analysed these movements through their traces and surface EMG. A penlight was attached to the index finger and the trace was recorded on photographs. At the same time, surface EMG was recorded from the muscles of biceps brachii, triceps brachii, extensor digitorum communis, and flexor carpi ulnaris.

Results

Figure 1 shows the findings in case 1. When the patient moved her index finger following the target, rapid and coarse movements were observed and the trace of her index finger was irregular. Surface EMG, which was recorded at the same time, showed irregular discharges. The discharges were synchronized, not reciprocal, between the antagonistic muscles (Fig. 1 A). In contrast, when she mimicked the same movements with her eyes closed, she could skillfully move her index finger and the trace was very smooth. Surface EMG showed reg-

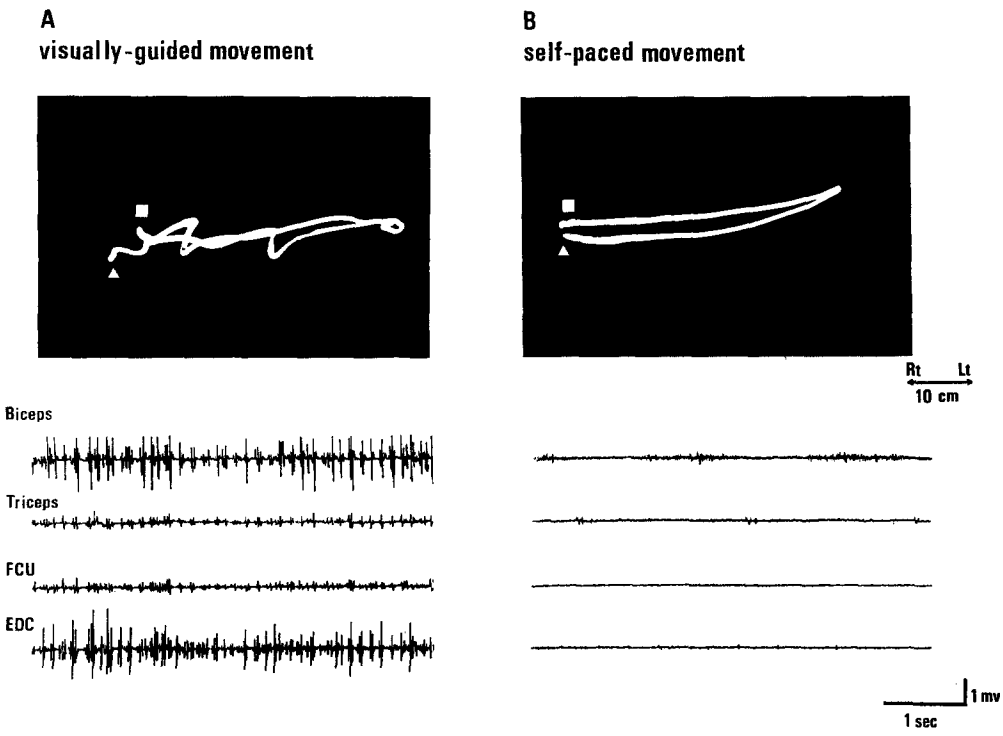

Fig. 2 The traces of the index finger (*upper*) and surface electromyography (*lower*) during visually guided movement (**A**) and self-paced movement (**B**), in case 2. The abbreviations and symbols are the same as in Fig.1

ular discharges accompanied by cyclic movement (Fig. 1 B).

The results in case 2 were similar to those in case 1 (Fig.2). Irregular jerky movements were easily induced by the movement of following the target, although in daily life such jerking was not so prominent. These movements were not observed when he imitated the same movements with his eyes closed.

Discussion

Action myoclonus and visually guided movement

The patients developed irregular jerky movements, which interfered with skilled voluntary movements. This involuntary movement was clinically considered to be action myoclonus, not ataxia, because of its jerky nature. Synchronized discharges between the antagonistic muscles on surface EMG confirmed this observation. We attempted to elucidate which movement induced this action myoclonus more prominently: sensory guided movement or self-paced movement. We designed these tasks to differentiate each movement independently. The task of having the patients move their index fingers following the target was considered visually guided movement, whereas the task of imitating these movements from memory was considered self-paced movement.

In self-paced movement, surface EMG showed small discharges in each muscle at a particular phase of the cyclic movement, but not synchronized between the antagonistic muscles. Then, this action myoclonus, which interfered with the skilled pursuing movements under visual guidance, disappeared in self-paced movement. These results show that this action myoclonus was induced by visually guided movement, and not by self-paced movement. We could also observe similar phenomena in daily life. For example, the patient in case 1 had difficulty manipulating objects under visual guidance because of action myoclonus, but she had no such difficulty with her eyes closed.

One might think that action myoclonus would be diminished in a task of self-paced movement because of its simplicity. However, in this task, we asked the patients to mimic precisely the movements they had seen, which required effort to achieve fine adjustments with stored memory information. Thus we conclude that the task of self-paced movement, which we chose, was not easy for the patients, and that the act of utilizing visual information to trigger and guide the movement exacerbated action myoclonus.

Possible mechanism

Goldberg [3] proposed that the cerebello-cerebral system controlled sensory guided movement. There are some

physiological data that support this hypothesis in some aspects, even though this hypothesis is over-simplified. Beppu et al. [1] showed that patients with cerebellar ataxia could not do well a visuomotor tracking movement using elbow flexion. At their own pace, however, they could do the elbow flexion movement fairly smoothly [1]. In addition, Mano et al. [9] directly observed that complex spikes in cerebellar Purkinje cells discharged only preceding visually guided movement, but not self-paced movement, in trained monkeys.

Some clinical studies have shown abnormalities within the cerebellar efferent system in cases showing action myoclonus [4, 6]. In fact, in sialidosis itself, Koga et al. [5] reported degenerative changes in the dentate nucleus. The mechanisms for action myoclonus must be more complicated. For example, it may result from hyperexcitability of the cerebral cortex, in the case of cortical myoclonus. However, there is no doubt that this cerebellar disorder is one aspect of complex mechanisms. Our observations, which have characters similar to Goldberg's hypothesis and physiological data, might reflect this cerebellar nature.

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