Stiff Person Syndrome with Pyramidal Signs

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Dear Editor,

Stiff person syndrome (SPS) is a rare disorder characterized by continuous muscle activity causing severe rigidity and episodic spasms in axial and limb muscles (1). According to the diagnostic criteria, normal motor and sensory examination is the rule. Hyperactive deep tendon reflexes may be observed, but extensor plantar reflexes are rare (1,2,3,4). Here, by presenting this case with atypical features like pyramidal signs, we aimed to review the clinical and electrophysiological signs of this rare syndrome.

A 39-year-old woman was admitted to our clinic with contractions induced by psychosocial stress in her legs and lumbar region since 2 years. Permission was obtained from the patient for this report. Neurological examination revealed increased lumbar lordosis, mildly decrease strength of proximal thigh muscles, hyperactive deep tendon reflexes, and extensor plantar reflexes.

Brain and whole spinal MRI were unremarkable. All biochemical and rheumatologic tests were normal. The cerebrospinal fluid (CSF) biochemistry was normal, and there were no atypical cells. The anti-glutamic acid decarboxylase (GAD) antibody serum level was high as 272 U/mL (<1.0 U/mL positive).

We performed electrophysiological examinations with the preliminary diagnosis of SPS. Needle EMG showed continuous activity in dorsal paraspinal and thigh muscles. Nociceptive flexor reflex, recorded after sural nerve stimuli was in a continuous pattern in anterior tibial (AT), biceps femoris, and lumbar paraspinal (LP) muscles, was recorded by four consecutive stimulations administered from the foot base (Figure

Figure 1. a, b. Flexor reflex (a): Our patient's record. Channels: right lumbar paraspinal, right biceps femoris, right anterior tibial, and left anterior tibial muscles. Obtained with four repetitive stimuli from foot base. Stimulus intensity 7.6 mA (b): Normal sample. Channels: anterior tibial and biceps femoris. Stimulus intensity 32 mA

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1a). Latency of the soleus H-reflex was normal, and reciprocal suppression by dorsal flexion of the foot remained. Auditory startle response was more easily elicited after a low volume without any habituation. All responses over the orbicularis oculi, sternocleidomastoid, biceps brachii, LP, and AT muscles were very high in amplitude with a very long duration (Figure 2a, b: normal sample). Somatosensory and trigeminal startle responses were found to be enhanced (Figure 3a, b). Blink and masseter inhibitory reflexes were normal (Figure 3b, 4a, 4b). In summary, increase in the excitability of polysynaptic spinal reflexes and startle responses was recorded, whereas other segmental and brain stem reflexes were normal. These features were consistent with the diagnosis of SPS.

Stiff person syndrome is a rare disorder, and the presence of atypical findings provides difficulty in diagnosing SPS. The diagnosis of SPS can be suspected based on clinical features and is supported by serological and electrophysiological tests. According to the diagnostic criteria defined by Gordon, normal motor and sensory examination is the rule (2). However, our patient had hyperactive deep tendon reflexes in all extremities

and plantar skin reflex responses were extensor. MRI examinations performed for investigating the etiology of pyramidal signs were all normal.

Anti-GAD antibodies are associated with several autoimmune diseases and are diagnostic markers in SPS (1,5). GABAergic pathways serve as one of the types of inhibitory pathways by inhibiting spontaneous discharges from spinal motor neurons. Impairment of the GABAergic pathways with deficiencies in GABA leads to continuous firing of the spinal motor neurons, with resultant stiffness and spasms (6).

Normality of motor unit morphology distinguishes SPS from other abnormalities that may be associated with stiffness. The rigidity and continuous motor unit activity decreases or even disappears during sleep and after spinal or general anesthesia, indicating a central source (7,8). Nerve conduction studies are normal; however, following discharges are seen after direct muscle response. Monosynaptic reflexes are hyperactive, H-reflex and reciprocal inhibition are sustained. Reduced vibration-induced suppression of the soleus H-reflex suggests a disorder of presynaptic inhi-

bition of Ia terminals in the spinal cord. Latencies of polysynaptic reflex responses are normal (50–80 ms), but responses are very long in duration as continuous crescendo responses (7,8,9). Exaggerated, non-habituating, exteroceptive or cutaneomuscular reflexes are characteristic features and contribute to the jerks and spasms (10). Startle responses in the cranial muscles are normal; however, they are exacerbated in the muscles of the trunk and lower sides. Exaggerated startle in SPS probably reflects segmental hyperexcitability of axial and lumbar spinal motor neurons (9,10,11). Here, by presenting this case, which meets these electrophysiological criteria defined for SPS, we aimed to review the clinical and electrophysiological signs of this rare syndrome.

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