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A CASE OF PRIMARY SPINAL MYOCLONUS

Clinical presentation and possible mechanisms involved

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ABSTRACT - Spinal myoclonus is a rare movement disorder characterized by myoclonic involvement of a group of muscles supplied by a few contiguous segments of the spinal cord. Structural lesions are usually the cause, but in primary spinal myoclonus the etiology remains unknown. We present the case of a 26-year-old woman with cervical spinal myoclonus in which both clinical and electromyographic findings pointed to the segment C1-C3 as the origin of the myoclonus. Laboratorial examinations were normal and no structural lesion was found in magnetic resonance imaging (MRI). Botulinum toxin type A was injected in infrahyoid muscles and cervical paraspinal musculature. The patient remained free of symptoms for almost five months. The pathophysiology of spinal myoclonus remains speculative, but there is evidence that various possible mechanisms can be involved: loss of inhibitory function of local dorsal horn interneurons, abnormal hyperactivity of local anterior horn neurons, aberrant local axons re-excitations and loss of inhibition from suprasegmentar descending pathways.

KEY WORDS: spinal myoclonus, segmental myoclonus, primary myoclonus.

Um caso de mioclonia espinhal primária: apresentação clínica e possíveis mecanismos envolvidos

RESUMO - A mioclonia espinhal é um raro distúrbio do movimento, caracterizado pelo envolvimento mioclônico de um grupo de músculos inervados por segmentos medulares contíguos. Lesões estruturais costumam ser a causa, mas na mioclonia espinhal primária a etiologia não é definida. Apresentamos o caso de uma mulher de 26 anos com mioclonia cervical espinhal em quem os achados clínicos e eletrofisiológicos apontaram o segmento C1-C3 como origem das mioclonias. Os exames laboratoriais foram normais e nenhuma lesão estrutural foi encontrada à ressonância. A toxina botulínica tipo A foi injetada nos músculos infrahioideos e na musculatura paraespinhal cervical. A paciente permaneceu assintomática por cinco meses. A patofisiologia da mioclonia espinhal continua especulativa, mas há evidências de que vários mecanismos possam estar envolvidos: perda da função inibitória de interneurônios da coluna dorsal, hiperatividade anormal de neurônios do corno anterior da medula, re-excitações axonais locais aberrantes e perda do efeito inibitório de vias descendentes suprasegmentares.

PALAVRAS-CHAVE: mioclonia espinhal, mioclonia segmentar, mioclonia primária.

Myoclonus is defined as a sudden, brief, shock-like, involuntary movement due to either active muscular contraction (positive myoclonus) or inhibition of muscle activity (negative myoclonus)[1]. Segmental myoclonus is a rare movement disorder characterized by myoclonic involvement of a muscle or a group of muscles supplied by a few contiguous segments of the brain stem or spinal cord[2]. Several causes of spinal myoclonus have already been described including spinal tumors, infections, vascular lesions, spinal anesthesia, AIDS and demyelinating diseases, but in a few cases the etiology remains unknown[2-5].

We present a case of spinal cord myoclonus in which no structural lesion was found in magnetic resonance imaging (MRI) and review some possible pathophysiological mechanisms.

CASE

A 26-year-old woman came for evaluation with a history of rapid onset of brief involuntary periodic movements on her neck over the past four weeks. There was no significant previous medical history nor family history of neurological illness. She had not used any drugs in the past or recently. She referred that the severity and frequency of

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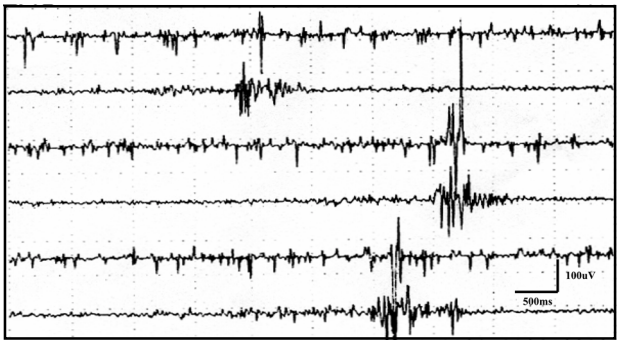

Fig 1. EMG of a single spontaneous jerk registered before injection of Botulinum toxin A in the paraspinal cervical musculature, geniohyoid muscle and neck anterior wall muscles of right (A, C, E) and left (B, D, F) sides, respectively.

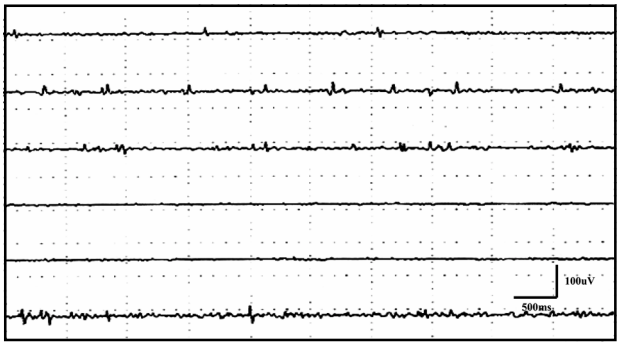

Fig 2. EMG registered in the same muscles described in Figure 1 (A, C, E – right; B, D, F – left), two weeks after Botulinum toxin A. There was reduction in both rate and magnitude of the burst myoclonic discharges.

movements increased rapidly in the first few days and became gradually worse in the last three weeks. General physical examination was normal. On neurological examination, there were involuntary spontaneous synchronous myoclonic jerks of the anterior wall of her neck, submandibular region and nape musculature, which resulted in slight extension of her neck. The contractions were rhythimic, bilateral and with a rate of approximately 1 Hz. No myoclonus was observed in the tongue. There was no vocalization. These movements could not be controlled by her will or effort. The rate and amplitude of involuntary movements were increased by emotional stress and neck extension and did not ceased during sleep. MRI of the brain and spinal cord, cerebrospinal fluid, routine hematological and immunological examinations (including HIV, HTLV I/II, hepatitis B and C, and syphilis), calcium, magnesium, copper, ceruloplasmin concentrations, and routine electroencephalography were normal.

Needle electromyography (EMG) showed rhythmic irregular burst discharges of motor units with a rate of 1-3 Hz in the muscles of anterior wall of the neck and cervical paraspinal muscles bilaterally (Fig 1). Trapezius and sternocleidomastoid muscles had no abnormal contractions. The bursts of activity were increased by neck extension and almost disappeared with neck flexion.

Sodium valproate 750mg/day was prescribed for two weeks but no improvement was observed. Botulinum toxin type A (Botox ®, Allergan) was injected: 20 units in each side of cervical paraspinal musculature, 20 units in each splenius capitis, 10 units in each geniohyoid muscle and 10 units in the infrahyoid musculature on the left side. Bilateral injection of infrahyoid muscles was avoided in order to prevent disphagia. Two weeks later, there was complete cessation of myoclonus, and electromyographic reduction in both rate and magnitude of the burst discharges of myoclonus (Fig 2). Patient was videotaped pre and post toxin injection. She remained free of symptoms for almost five months.

DISCUSSION

Pathophysiologically, myoclonus can be broadly classified as cortical, subcortical, cortical-subcortical, segmental, or peripheral[6]. In the segmental type, lesions placed at different locations along the neuraxis may be the cause. When the presumed cause is in the spinal cord, it is called spinal myoclonus. The responsible site is usually estimated by clinical observation and electrophysiological examination. This rare kind of myoclonus involves only the musculature innervated by a few adjacent spinal levels and is usually rhythmic and slow (<4Hz). The presence of myoclonus in the mouth floor of our patient and the lack of involvement of her tongue suggested that the hypoglossus nuclei themselves were not involved. The affected muscles in the mouth floor belongs to a group called infrahyoid muscles. The infrahyoid muscles (geniohyoid, thyrohyoid, sternothyroid, sternohyoid and omohyoid) are supplied by nerve fibers derived from the first cervical nerve (C1) and from the junction of the second and third cervical nerves (C2-C3)[7]. The EMG showed the involvement of muscles of the anterior wall of the neck known as infrahyoid muscles and the simultaneous myoclonic jerks of cervical paraspinal muscles bilaterally. The main muscles of this site include semispinalis capitis, rectus capitis posterior, obliquus capitis superior and splenius capitis bilaterally, which also depend of C1 and C2 nerves[8]. Trapezius and sternocleidomastoid muscles were free. Both clinical and electromyographic findings pointed to the segment C1-C3 as the site of segmental spinal myoclonus.

The pathophysiology of spinal myoclonus remains speculative. In 1979, Howell et al. suggested that focal myoclonus could be caused by loss of local inhibitory spinal interneuronal function, which allowed the spontaneous repetitive discharge of local segmental anterior horn cell pools[9]. In 1981 Davis et al. provided histological evidence for this hypothesis in

a case in which the number of small and medium sized neurons in the posterior horns of the lumbar cord was reduced along with relative sparing of large neurons in the anterior horns[10]. On the other hand, another study showed that physiological suppression of dorsal horn interneurons failed to occur in segmental myoclonus, indicating that dorsal horn interneurons could be abnormally hyperactive[11]. Jankovic hypothesized that the rhythmical contractions could be the expression of spontaneous spinal neuronal discharge due to suppression of physiologic inhibition from suprasegmentar levels but experimental evidence for this hypothesis is lacking[2].

Electrophysiological studies in hemifacial spasm, which is a form of segmental myoclonus, may contribute to elucidate the possible mechanisms of spinal myoclonus. Axono-axonal ephatic transmission among injured fibers due to compression and demyelination of the intracranial segment of the facial nerve[12,13] or abnormal central hyperexcitability of the facial motor nucleus or both[14,17] have been considered as possible mechanisms. Neurophysiological studies in patients with cryptogenic hemifacial spasm showed that the self-sustained repetitive firing in facial nerve axons could result from re-excitations occurring both at the ephapse site on the axon and at the cellular level[18] or from a permanent antidromic stimulation from a peripheral ectopic center of excitation[19]. In summary, there is evidence that various possible mechanisms can be involved: loss of inhibitory function of local dorsal horn interneurons, abnormal hyperactivity of local anterior horn neurons, aberrant local axons re-excitations and loss of inhibition from suprasegmentar descending pathways.

The worsening of myoclonus in relation to changes in position has been described[3,20]. In our patient, the worsening of myoclonus during neck extension suggests that peripheral stimulus may modulate the abnormal movement, but the symmetrical pattern of muscle activity points that local spinal generators could be the source of myoclonus. Our patient remained asymptomatic for five months after botulinun toxin injection. This longer duration of benefit as compared to that observed in focal dystonias have been described in similar cases and points to a higher degree of susceptibility of this type of movement to chemical denervation with botulinum toxin[4].

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