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Case Report

Surface electromyogram and muscle ultrasonography for detection of muscle fasciculations in pediatric peripheral neuropathy

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Abstract

A 12-year-old girl presented with talipes equinus of both legs, attenuation of upper and lower limb tendon reflexes, thermal hyperalgesia, and reduction of vibratory sensation. On clinical examination, muscle twitches of fingers of both hands, as well as the abductor halluces and the dorsal interossei muscles of the right foot were observed. Nerve conduction velocity was significantly declined in the upper and lower extremities. Needle electromyography (EMG) was not performed; however, ultrasonography revealed repetitive, semi-regular muscle twitches lasting 0.2–0.4 s, concomitant with muscle discharges on surface EMG in the right foot muscles. These findings were compatible with contraction fasciculation in muscles under chronic reinnervation. Nerve and muscle biopsies were suggestive of chronic motor, sensory, and autonomic neuropathy. This is the first case of pediatric peripheral neuropathy where muscle fasciculation was noninvasively identified by simultaneous surface EMG and ultrasonography.

Keywords: Fasciculation; Peripheral neuropathy; Surface electromyography; Ultrasonography; Autonomic sensory motor neuropathy

1. Introduction

Fasciculations are repetitive brief muscle twitches observed in muscles under denervation and/or chronic reinnervation, usually lasting 0.2–0.5 s [1]. They are commonly found in amyotrophic lateral sclerosis (ALS), spinal muscle atrophy (SMA), and less frequently in hereditary motor and sensory neuropathy [2–4]. Recent progress of ultrasonography (US) has achieved visualization of fascicular muscle contraction and it now plays an indispensable role in the characterization of neuromuscular disease. Generally, needle electromyography (EMG) and surface EMG are used for the detection of fasciculations [5]. However, in the field of pediatric neurology, neither muscle US nor surface EMG has been routinely used for the detection of fasciculations. Since these are noninvasive and easily applicable modalities,

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they could be helpful to evaluate fasciculations in children. We report a case of a girl with peripheral neuropathy for whom fasciculation was detected by simultaneous recording of muscle US and surface EMG.

2. Case report

The patient was a 12-year-old girl who presented with cyclic vomiting since 9 years of age, which necessitated repeated admission for up to 12 days each month. Meanwhile, a steppage gait emerged at age 10 years and 5 months, which progressively worsened. She was admitted to our hospital at the age of 12 years because of persistent abdominal pain and daily vomiting up to 20 times. Clinical examination revealed characteristic features of peripheral neuropathy: talipes equinus of both legs, deep tendon reflex attenuated in the upper limbs and absent in the lower limbs, thermal hyperalgesia, and reduction of vibratory sensation. The sensation of pinprick and cold stimuli appeared to be decreased. The patient had thin lower legs and contractures in the ankles and toes. Otherwise, high arched feet, hammer digit deformities, pyramidal signs, and focal muscle atrophy were not noted. Muscle power of the upper and lower extremities was assessed as good by Manual Muscle Testing. Cerebrospinal fluid (CSF) analysis revealed a white blood cell count of 3, protein level of 21 mg/dL, and glucose level of 62 mg/dL (serum glucose level of 113 mg/dL). Nerve conduction study in an air-conditioned room at a constant temperature of at 28 °C revealed significant decrease in the motor and sensory conduction velocities and amplitude of compound muscle/sensory nerve action potentials of median, tibial, and sural nerves bilaterally (Supplementary Table 1). The F waves of the median and tibial nerves appeared to have prolonged latencies and decreased frequencies of provocation (Supplementary Table 1, Supplementary Fig. 1). Needle EMG was not performed. At the age of 12 years and 10 months, muscle twitches of the fingers of both hands and right abductor halluces muscle and the right dorsal interossei of the right foot were noticed at rest. Surface EMG, with electrodes placed on the skin over the right abductor halluces muscle and the right dorsal interossei of foot, showed repetitive focal muscle twitches at the same time as US identified muscle contractions (Fig. 1A). The maximum amplitude of the right abductor hallucis muscle twitches was 192 μV without artifacts. US recorded muscle twitches at rest, revealing repetitive, semi-regular twitches of an identical muscle fascicle lasting 0.2–0.4 s (Fig. 1B and C, and Video 1).

A biopsy of the left biceps muscle showed marked fiber-type grouping, suggestive of neuropathic changes with chronic denervating and reinnervating process (Supplementary Fig. 2). A biopsy of the left sural nerve showed a reduction in the number of myelinated fibers with a loss of bimodal distribution in diameter, and severe reduction of unmyelinated fibers (Supplementary Fig. 3). Hereditary motor and sensory neuropathy gene analyses showed normal results (see Acknowledgments).

Intravenous hyperalimentation was initiated soon after admission, but cyclic vomiting remained refractory to medications of phenobarbital, valproate, and amitriptyline, and was accompanied by sustained miosis of the pupils of 2 mm in diameter, decreased coefficient of variation of R-R intervals, and absence of sympathetic skin response (Supplementary Table 1). With a suspicion of autoimmune neuropathy, we treated the patient with intravenous immunoglobulins, which showed no effect. Methylprednisolone (mPSL) pulse therapy was effective in terminating the recurrent vomiting and increasing coefficient of variation of R-R intervals (Supplementary Table 1). However, cyclic vomiting recurred within a few months with a maintenance dose of 0.8 mg/kg/d prednisone. After repeated mPSL pulse therapy; gastrostomy; and treatment with valproate, amitriptyline, and topiramate, the abdominal pain resolved and vomiting was significantly ameliorated at 13 years. However, nerve conduction studies showed a progressive decrease in conduction velocities and evoked potential amplitudes (Supplementary Table 1). The patient has been admission-free for >1 year without a steroid therapy. Atrophy of the thenar and lower leg muscles became evident by 14 years of age. Romberg sign was negative. The largest circumference of lower leg was 24.5 and 24 cm on the left and right side, respectively. Orthostatic hypotension was confirmed at this period. We tentatively diagnosed the patient as having chronic motor, sensory, and autonomic neuropathy probably involving both axonal and demyelinating pathology, which is not well established in the literature to date.

3. Discussion

Generalized muscle fasciculations result from disorders involving the spinal motor neurons. US detected tongue fasciculations in 60% of 81 patients with ALS [2]. In addition, muscle US detected fasciculations in 100% of 10 children with SMA and 96% of 25 patients with hereditary motor and sensory neuropathy [3,4]. In addition, fasciculations can also accompany certain peripheral neuropathies through involvement of either distal axons, synaptic terminals, or presynaptic terminals [6]. In general, these fasciculations in peripheral neuropathy show semi-regular appearance and called as "contraction fasciculation" under chronic reinnervation, which represents an action to sustain certain posture and disappears at completely resting state [1,7]. In contrast, "true" fasciculations in ALS and SMA [3] typically show irregular, random appearance at rest. In addition, "contraction fasciculation" can also be