

**LETTER TO THE EDITOR**

Pseudodystonia and Neuropathic Tremor in a Patient With Monomelic Amyotrophy

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Dear Editor,

Dystonia is a complex movement disorder characterized by sustained or intermittent muscle contractions causing patterned, repetitive movements or abnormal posture.¹ The anatomical causes of dystonia have not been fully elucidated; however, recent studies have suggested that it is a network disorder involving several brain regions, including the basal ganglia, cerebellum, thalamus, brainstem, and cortex.² However, determining the pathological substrates of dystonic movements is difficult because of its heterogeneous manifestations and etiologies. Furthermore, a condition mimicking dystonia, referred to as pseudodystonia, should also be considered during differential diagnosis. Pseudodystonia is also characterized by abnormal muscle contraction, resulting in patterned movements or abnormal postures, but is not compatible with dystonia.³ We report the case of a patient with monomelic amyotrophy, a form of focal motor neuron disease (MND), who presented with pseudodystonia with neuropathic tremor.

A 22-year-old male presenting with tremors of both hands that began at 18 years of age was referred to the Department of Neurology at Asan Medical Center. Bilateral hand tremors persisted in resting and action and were aggravated while grabbing a cup or extending the forearms. Occasionally, his left fingers, wrist, and elbow would involuntarily contract with pain. The tremors had worsened over recent years, and as a result of progressive weakness, the patient had avoided lifting objects with his left hand. No family history of abnormal movement was reported. Neurological examination revealed asymmetric (left side dominant) amyotrophy in the intrinsic hand muscles (Figure 1). Abnormal posture was observed in the left hand with repetitive and

patterned flexion of all five fingers but predominantly in the interphalangeal joint of the thumb while his hands were extended (Figure 1 and Supplementary Video 1 in the online-only data supplement). Finger abductors were assessed as Grade 3/5 on the Medical Research Council Scale on the left and 4/5 on the right, while finger flexion was 4+/5 on the left and 5/5 on the right. He had an irregular tremor with a small amplitude and high frequency in the left thumb in the resting state and postural tremors in both hands (Supplementary Video 1 in the online-only data supplement). A mild kinetic tremor was observed in the left hand during the Archimedes spiral test. A nerve conduction study (NCS) indicated normal distal latencies and conduction velocities in the bilateral median and ulnar nerves but decreased compound motor action potential amplitude in the left ulnar nerve. Electromyography (EMG) findings suggested acute and chronic denervation in the muscles of the left distal upper limb belonging to the C8-T1 roots (Supplementary Figure 1 in the online-only data supplement). Motor evoked potential study results were normal. There was no indication of compressive cord lesions or abnormal cord flattening upon cervical magnetic resonance imaging (MRI) in a neutral position. A cervical MRI on flexion was not performed. The patient was subsequently diagnosed with monomelic amyotrophy. After 6 months of clinical observation, follow-up EMG and spinal MRI showed no significant changes. His symptoms remained stable.

Monomelic amyotrophy, also termed Hirayama disease, is a benign form of juvenile-onset lower MND.⁴ Neurological manifestations of monomelic amyotrophy include insidious onset of asymmetric weakness and muscular atrophy of the distal upper limb in the C7, C8, and T1 myotome distribution, followed by

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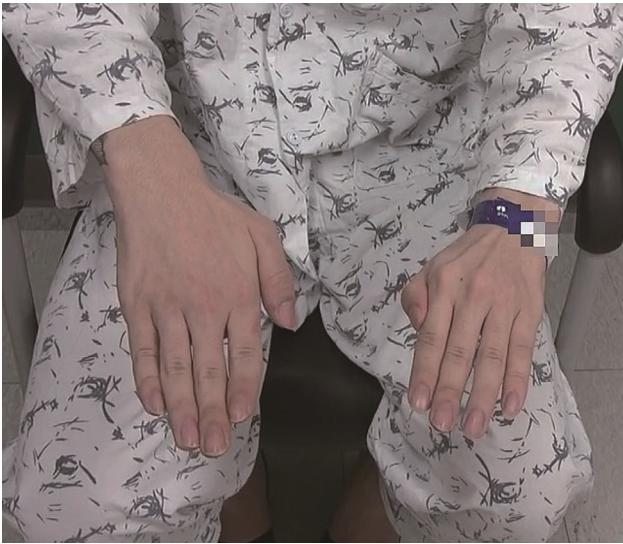


Figure 1. Asymmetric (left side dominant) atrophy in the intrinsic hand muscles and abnormal posture in the left hand with the extension of both hands.

spontaneous stabilization.^{4,5} Neurophysiologic studies have revealed chronic denervation of the affected muscles, with or without acute denervation. Radiologic findings of forward displacement of the cervical dural sac and flattening of the lower cervical cord on neck flexion are characteristic features.⁴ However, such findings might not be observed in neutral position MRI and may be confined to only the progressive stages. Conservative management using a neck collar to avoid neck flexion is recommended for this self-limiting disease to prevent progression.⁵ Decompression surgery is indicated only for patients with persistent neurologic deficits.

A fixed posture with a “claw hand” has been reported in patients with monomelic amyotrophy⁵ and is known to be caused by weakness of the intrinsic muscles as a result of peripheral nerve damage in the arm. However, to our knowledge, an abnormal posture with repetitive muscle contractions as seen in our patient, rather than a fixed claw posture, has not been described in the literature. This abnormal posture mimicking dystonia of the patient is plausible for pseudodystonia, which presents as abnormal postures with superimposed patterned movements and usually features additional findings in the affected body part.³ Our patient showed atrophy and weakness in the affected hand muscles, which are important features for distinguishing pseudodystonia from dystonia. The etiology of pseudodystonia differs from that of dystonia and includes disorders of the peripheral nervous system, spinal cord, or musculoskeletal diseases.³ Pseudodystonia has been reported in various disorders of motor pathways, which are related to hyperexcitation of the pyramidal tracts, peripheral motor nerves, or spinal interneuronal networks.³ In this case, clinical features and neurophysiologic findings suggested

focal loss at the anterior horn cell level. Despite the uncertainty of the underlying mechanism of involuntary movements mimicking dystonia in monomelic amyotrophy, dysfunction in lower motor neurons (LMNs) is postulated to be involved in abnormal muscle contractions. The LMNs in the spinal cord project axons that innervate skeletal muscle fibers, which simultaneously contract after depolarization of the LMN.⁶ Loss of motor neurons and increased motor neuronal excitability are pathophysiologic markers of MND.⁶ Motor axonal hyperexcitability is associated with the generation of fasciculations and muscle cramps in the peripheral nervous system in amyotrophic lateral sclerosis (ALS), the most common MND.⁶

Irregular, small-amplitude, jerky movements in the fingers known as ‘minipolymyoclonus’ have been described in MND; however, the pathophysiology of these tremulous movements remains unclear. A recent study reported that frequent fasciculations originating from anterior horn cells in MND could clinically manifest minipolymyoclonus.⁷ In monomelic amyotrophy, focal fasciculations arise from damage to the anterior horn cells, likely related to this neuropathy-induced tremor.

In conclusion, our patient with monomelic amyotrophy presented with pseudodystonia and neuropathic tremor. Our case highlights that careful inspection and examination are essential for the differential diagnosis of rare causes of pseudodystonia.

Ethics Statement

All procedures performed in studies involving human participants were in accordance with the Institutional Review Board of Asan Medical Center and with the 1975 Declaration of Helsinki and its later amendments or comparable ethical standards. Written informed consent was obtained from the patient.

Supplementary Video Legends

Video 1. The patient shows irregular, small-amplitude, and high-frequency tremors in the left thumb in the resting state and bilateral hand tremors while extending his arms. A kinetic tremor in the left hand is seen during the Archimedes spiral test. An abnormal posture with repetitive and patterned movements in the left hand induced by extension of the hand can be observed. Asymmetric (left side dominant) hand weakness with finger abduction was also observed.

Supplementary Materials

The online-only Data Supplement is available with this article at <https://doi.org/10.14802/jmd.21138>.

Conflicts of Interest

The authors have no financial conflicts of interest.

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Author Contributions

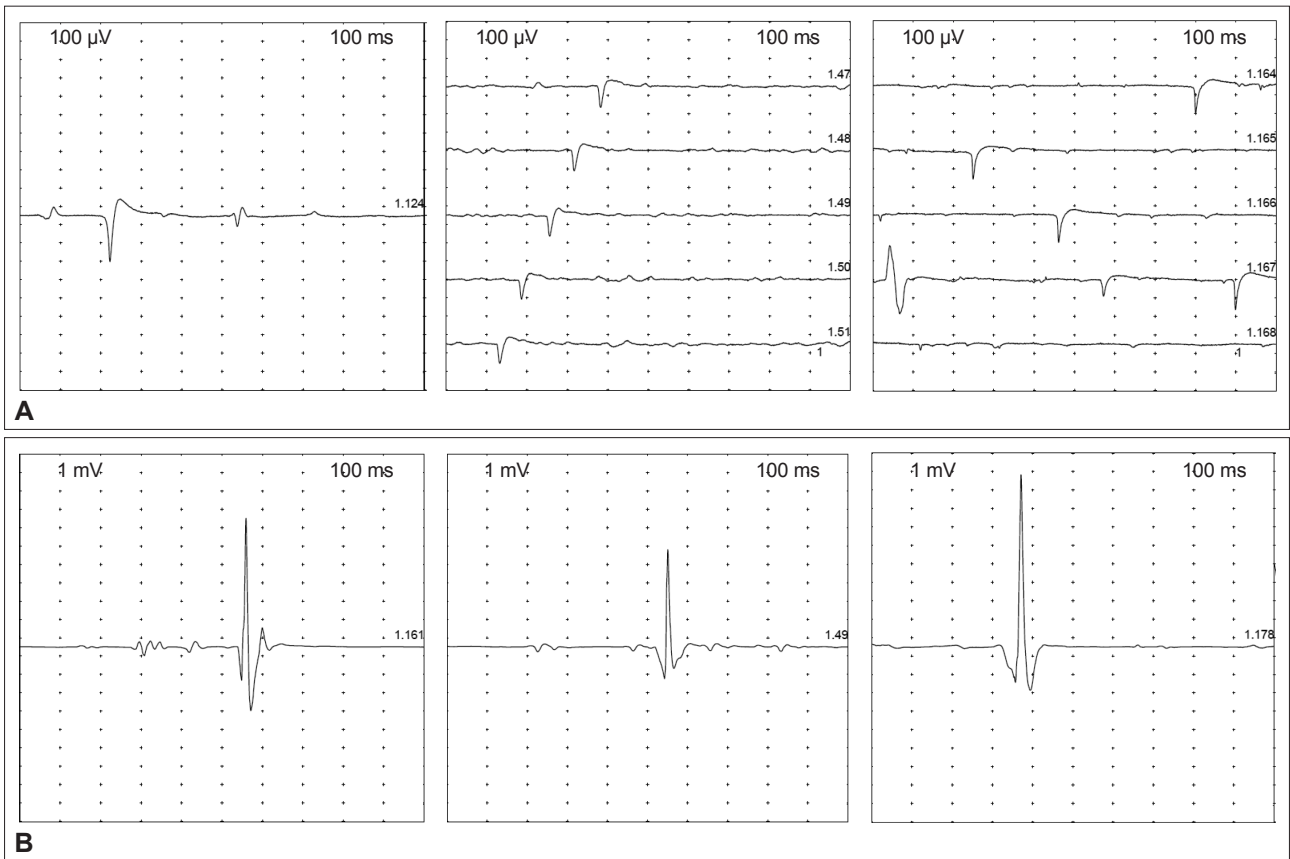
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Supplementary Figure 1. Electromyography (EMG) findings. A: Positive sharp waves in the left first dorsal interosseous, flexor digitorum profundus and abductor digiti minimi muscles. B: High-amplitude and long-duration MUAPs in the left first dorsal interosseous, flexor digitorum profundus and abductor digiti minimi muscles. MUAPs, motor unit action potentials.