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IMAGES IN CLINICAL NEUROLOGY

Unilateral polyminimyoclonus in Hirayama disease: A frequent misdiagnosis as a functional movement disorder

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Hirayama disease (HD), historically recognized as monomelic amyotrophy or juvenile asymmetric segmental spinal muscular atrophy, is a rare neurological condition first described by Hirayama et al.[1] in 1959. The disorder predominantly affects young males, with a peak onset typically occurring between adolescence and the early third decade of life.[2,3] Pathophysiologically, HD is considered a focal cervical myelopathy precipitated by the anterior displacement of the posterior cervical dural sac during neck flexion. This displacement results in transient spinal cord compression and chronic venous congestion within the lower cervical segments.[4,5] The clinical hallmark of HD is the insidious and progressive onset of symmetric or asymmetric muscular weakness and atrophy, primarily involving the distal upper extremities.[3-5] Associated clinical manifestations, reported in over 80% of cases, include cold paresis, sensory disturbances such as numbness or paresthesia, and irregular tremors. Notably, this irregular tremor characterized as polyminimyoclonus during finger extension is frequently exacerbated by cervical flexion.[2,3-5]

Functional movement disorders (FMDs) are frequently encountered in clinical neurology and represent a significant source of long-term disability.[6] These disorders typically exhibit a higher prevalence in females.[7] Patients often present with a diverse range of symptoms, including paroxysmal hyperkinesias such as tremor, dystonia, and myoclonic jerks, as well as sensory disturbances, limb weakness, nonepileptic seizures, gait and speech abnormalities, and vertigo. Notably, multiple movement phenomenologies frequently coexist in a single patient.[8] Among FMDs, tremor is the most common manifestation, primarily involving the upper and lower extremities. Diagnosis remains clinical, relying heavily on a meticulous history and a detailed neurological examination.[7,8] Herein, we reported the case of a 23-year-old female patient with HD who was initially misdiagnosed with a functional tremor.

A 23-year-old right-handed female who regularly participated in upper-limb-dominant fitness activities was admitted to our clinic with a six-year history of progressive weakness and tremor in the right hand. The initial symptoms, which emerged at age 17 years, consisted of irregular finger tremors. At that time, the patient was misdiagnosed with a FMD and prescribed a regimen of antidepressants and anxiolytics. Despite long-term adherence to these treatments, no clinical improvement was observed. Eight months following the initial onset, the patient developed progressive weakness in the right hand. Neurological examination at our center revealed unilateral wrist extension weakness (Medical Research Council grade 3/5) and polyminimyoclonus of the right-hand fingers at rest (Video 1). During the neurological examination, it was noted that the polyminimyoclonus affecting the patient's right-hand fingers remained persistent

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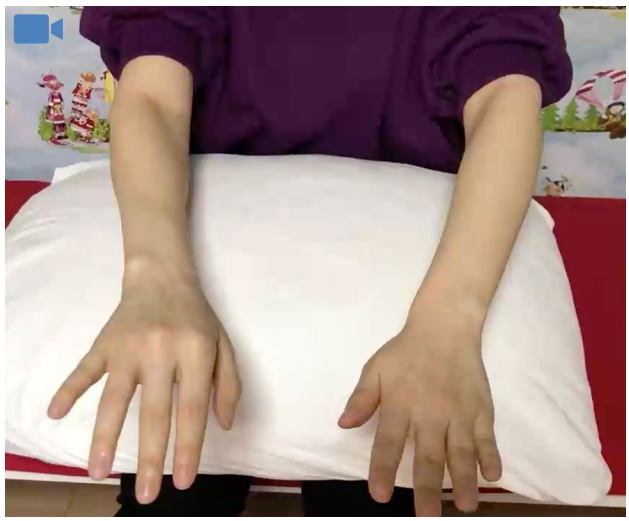

Video 1. Polyminimyoclonus observed in the resting right-hand fingers.

and nondistractible despite the introduction of concurrent cognitive and motors tasks. Although the patient reported paresthesia along the extensor surface of the forearm, objective sensory testing was normal. Deep tendon reflexes were hypoactive in the upper limbs, while those in the lower limbs were normoactive. Plantar responses were flexor bilaterally. Laboratory evaluation, including routine hematological and biochemical profiles and serum anti-GM1 (ganglioside M1) immunoglobulin M antibodies, was normal. Furthermore, brain magnetic resonance imaging (MRI) showed no structural abnormalities. However, nerve conduction studies (NCS) and needle electromyography (EMG) supported the clinical suspicion of chronic focal anterior horn cell involvement within the C6-C7 spinal segments, with neurophysiological abnormalities being more pronounced on the right side. Notably, despite the absence of clinically evident muscle atrophy, NCS and EMG revealed chronic denervation potentials in the contralateral homologous muscles, indicating subclinical involvement. The combined results of the NCS and needle EMG are presented in Table 1. Cervical spine MRI in neutral-position sagittal T2 images revealed a loss of normal cervical lordosis and minimal cord atrophy at the C6-7 level (Figures 1a, b and c). Cervical spine MRI in the flexion position showed a narrowed anterior subarachnoid space, and axial T2-weighted images displayed intramedullary bilateral anterior horn cell hyperintensity at the C6-7 level, also known as the "snake eyes sign" (Figure 2).

Given the characteristic clinical presentation, neurophysiological evidence, and supportive neuroimaging findings, a definitive diagnosis of HD was established. The patient was managed conservatively and discharged with a soft cervical collar to restrict cervical flexion. Furthermore, the patient was advised to discontinue upper-limb-dominant fitness activities to mitigate repetitive mechanical stress. During follow-up, the patient remained clinically stable, with no reported progression of neurological dysfunction.

The majority of reported HD cases originate from Asian populations, particularly from Japan, China, and India. In contrast, the disease remains relatively underrecognized in North America, Europe, and Australia,[2-5] where the literature is primarily limited to small case series. Herein, we presented a rare and atypical manifestation of HD that deviated from the classical clinical phenotype in several key aspects. First, while HD exhibits a profound male predominance with a reported male-to-female ratio of approximately 20:1,[2,3] our patient is female. Second, the temporal progression of symptoms in our case was atypical; whereas HD typically presents with muscle weakness followed by atrophy and irregular tremor, our patient's primary complaint was a localized hand tremor that preceded any detectable weakness. Notably, this tremor was not exacerbated by cervical flexion, a feature frequently observed in classical postural tremors of HD.[2,4,5] This atypical presentation led to a prolonged misdiagnosis of functional tremor, for which the patient received unsuccessful psychiatric intervention. It is worth noting that while tremor is a hallmark of FMDs, such tremors often spare the individual fingers, unlike the polyminimyoclonus observed in our patient. Eight months following the onset of the tremor, the patient developed a gradual weakness in right-hand wrist extension. Furthermore, although "oblique amyotrophy," characterized by a distinct oblique border of muscular atrophy involving the volar and dorsal forearms, is considered a pathognomonic feature of HD, clinically evident muscular atrophy was notably absent in our patient. The clinical course remained self-limiting, with spontaneous stabilization following an initial six-year period of symptom progression. Fourth, while the median and ulnar nerve distributions are the primary sites of clinical and electrophysiological involvement in the majority of reported HD cases,[2-5] the radial motor nerve was the primary site of involvement in our case. Although HD typically presents as a unilateral monomelic amyotrophy, it often manifests

TABLE 1
Combined NCS and needle EMG in the patient with HD

Nerve / Recording	Stimulation site	Onset latency (ms)	Amplitude sensory-μV motor-mV	Distance (mm)	Conduction velocity (m/s)
Right median sensory / index finger	Wrist	2.55	58.5	150	58.8
Right ulnar sensory / digiti minimi	Wrist	2.29	46.5	150	65.5
Right radial sensory / polex finger	Distal dorsal forearm	2.32	14.5	150	64.7
Right median motor / APB muscle	Wrist - elbow	3.56 / 6.65	6.2 / 6.1	200	63.4
Right ulnar motor / ADM muscle	Wrist - above elbow	3.13 / 6.38	6.7 / 6.5	230	70.7
Right radial motor / EIP muscle	Forearm - spiral groove	3.27 / 6.00	1.83 / 1.26	140	51.2
Left median sensory / index finger	Wrist	2.50	57.6	150	60.0
Left ulnar sensory / digiti minimi	Wrist	2.30	49.4	150	64.6
Left radial sensory / polex finger	Distal dorsal forearm	2.34	16.4	150	64.1
Left median motor / APB muscle	Wrist - elbow	3.47 / 6.51	7.0 / 6.3	200	65.7
Left ulnar motor / ADM muscle	Wrist - above elbow	3.12 / 6.03	7.6 / 6.9	230	72.3
Left radial motor / EIP muscle	Forearm - spiral groove	3.17 / 5.74	3.13 / 2.75	140	54.3
Right sural sensory / lateral malleolus	Ankle	1.77	23.5	90	50.8
Right peroneal motor / EDB muscle	Ankle-fibula head	3.75 / 9.2	5.0 / 4.6	320	54.7
Right tibial motor / AHL muscle	Ankle - poplitea	5.26 / 12.40	7.1 / 5.9	395	55,3
Left sural sensory / lateral malleolus	Ankle	1.86	22.4	90	48.3
Left peroneal motor / EDB muscle	Ankle - fibula head	3.96 / 9.64	4.8 / 4.4	320	56.3
Left tibial motor / AHL muscle	Ankle - poplitea	5.0 / 11.95	6.9 / 6.2	395	56.8

Needle Electromyography	Spontaneous activity					Motor Unit Action Potentials (MUAPs)						Voluntary activity
	Fibs	PSW	Fasc	CRD	Other	Nml	LongD	Hamp	Poly	ShortD	Lamp	
Right deltoid						+++						Normal
Right biceps	None	None	None				++	++	+			Reduced recruitment
Right triceps	None	None	+				+++	+++				Reduced recruitment
Right EIP	None	None	+				+++	+++				Reduced recruitment
Right EDC	None	None	+				+++	+++				Reduced recruitment
Right APB						+++						Normal
Right ADM						+++						Normal
Left deltoid						+++						Normal
Left biceps	None	None	None				++	++	+			Reduced recruitment
Left triceps	None	None	None				++	++				Reduced recruitment
Left EIP	None	None	None				++	++				Reduced recruitment
Left EDC	None	None	None				++	++				Reduced recruitment
Left APB						+++						Normal
Left ADM						+++						Normal
Right thoracal Psp						+++						Normal
Left thoracal Psp						+++						Normal
Right tibialis Ant						+++						Normal
Left tibialis Ant						+++						Normal

APB, abductor pollicis brevis; ADM, abductor digiti minimi; EIP, extensor indicis proprius, EDB, extensor digitorum brevis; AHL, abductor hallucis longus tibialis; Ant: Tibialis anterior, thoracal; Fibs: Fibrillation potentials, PSW: Positive spike waveform; Fasc, fasciculation potentials; CRD, complex repetitive discharges; LongD, long duration; Hamp, high amplitude; Poly, polyphasi; ShortD, short duration; Lamp, low amplitude; EDC, extensor digitorum communis; Psp, thoracal paraspinal. Electrophysiologic studies were performed with a Dantec Keypoint EMG machine (Natus Medical, Inc., San Carlos, CA, USA). Sensory and motor NCS were performed with antidromic methods. Sensory and motor conduction velocity (upper extremity normal: > 50 m/s, lower extremity normal: > 40 m/s), motor nerve compound muscle action potential amplitudes (normal: > 4 mV, for peroneal motor, normal: > 2 mV), and sensory nerve compound muscle action potential amplitudes (normal: > 10 μV, for sural sensory, normal: > 7 μV) were assessed.

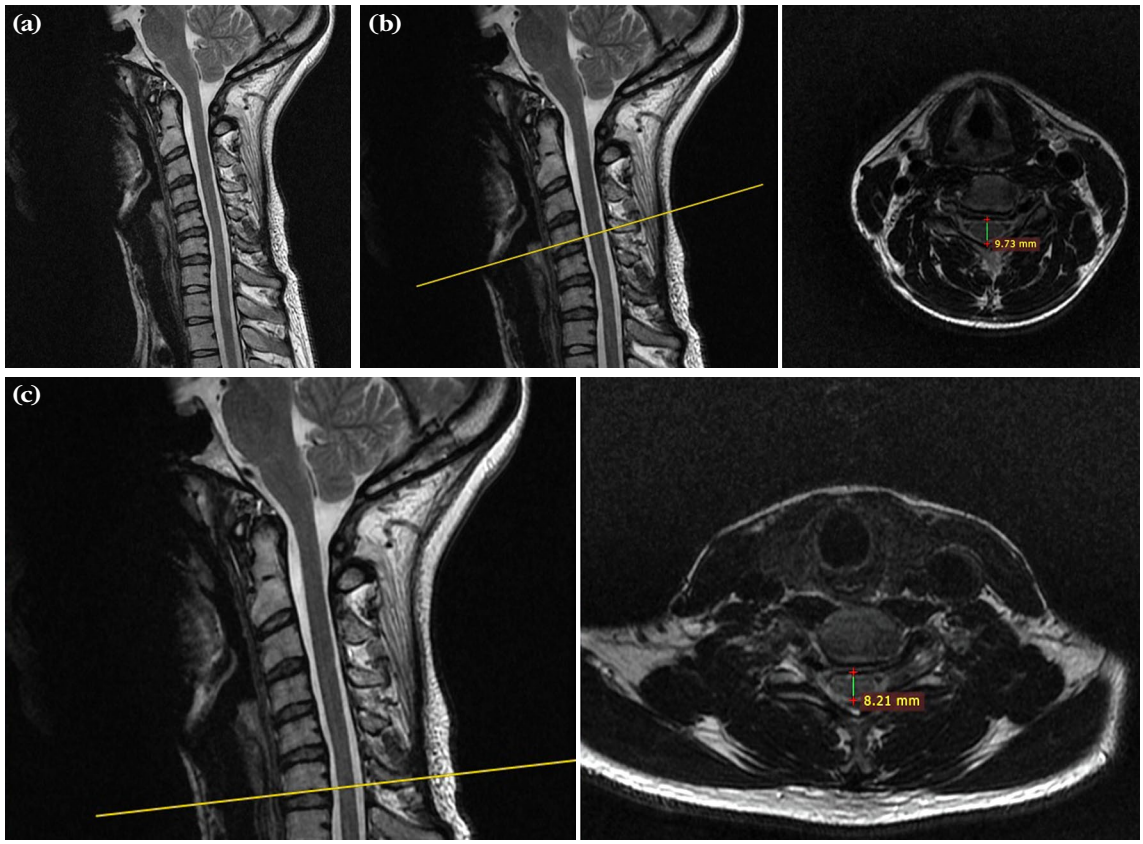

Figure 1. **(a)** Cervical spine MRI, neutral-position sagittal T2 images, showing minimal cord atrophy at the C6–7 level. **(b)** Cervical spine MRI, neutral-position sagittal and axial T2 images at the C4 level, demonstrating no abnormality of the spinal cord. The spinal cord diameter measures 9.73 mm. **(c)** Cervical spine MRI, neutral-position sagittal and axial T2 images, demonstrating minimal cord atrophy at the C6–7 level. The spinal cord diameter measures 8.21 mm.
MRI, magnetic resonance imaging.

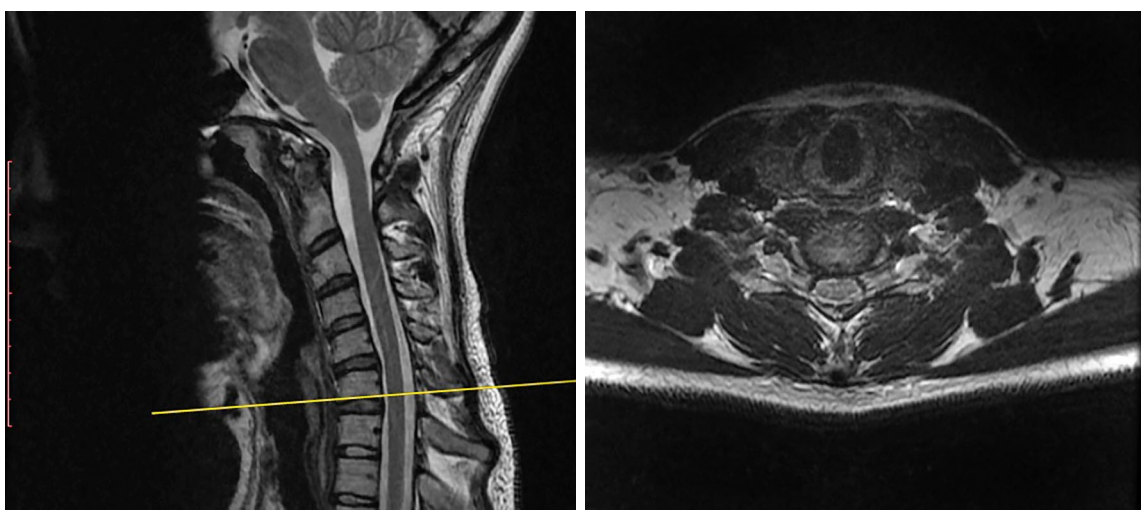

Figure 2. Cervical spine MRI in flexion showing a narrowed anterior subarachnoid space. Axial T2-weighted image in flexion demonstrating slight hyperintensity of the bilateral anterior horn cells ("snake eyes sign") at the C6–7 level.
MRI, magnetic resonance imaging.

as an asymmetric bilateral involvement. Subclinical denervation findings can be detected via EMG in the asymptomatic contralateral limb showing no clinical weakness or atrophy in 30 to 50% of patients. This involvement of the contralateral side reflects a more widespread affection of the spinal cord (specifically the anterior horn cells) rather than a focal lesion limited to a single nerve or root, although it was significantly more severe on one side. Notably, the presence of EMG abnormalities in the asymptomatic limb does not inevitably imply future clinical progression or functional decline in that extremity.

The precise pathophysiology of HD remains elusive; however, it is widely attributed to multifactorial mechanisms, specifically recurrent microtrauma and ischemic processes. These factors are hypothesized to induce microcirculatory disturbances within the anterior horn cells of the lower cervical spinal cord, leading to the characteristic focal amyotrophy.[3,5] Various activities, including upper-limb-dominant sports, basketball, and prolonged desk work, have been reported in association with this condition.[4,5] We hypothesize that long-term fitness activities in our HD patient may have contributed to the development of the condition. Optimized MRI, including dynamic or neck flexion positioning, is crucial for diagnosing suspected HD, as many characteristic features, such as anterior dural displacement, loss of dural attachment, obliteration or significant reduction of the posterior cervical subarachnoid space, epidural space enlargement, flow voids, and enhancement, are best visualized with these specialized imaging techniques.[4,9,10] Consistent with our patient's findings, previously reported HD features on routine cervical spine MRI include lower cervical cord atrophy, loss or reversal of the physiological cervical lordosis, and T2-weighted hyperintensities within the anterior horn cells.[2,3,10] Consequently, the prevailing pathogenic hypothesis for HD posits that excessive anterior displacement of the posterior dura during neck flexion induces ischemic injury to the cervical anterior horn or nerve roots.[9] Loss of dorsal dural attachment to the lamina/pedicle, potentially secondary to immunological abnormalities of the dura and posterior ligaments, has also been implicated in the pathogenesis.[11] Hirayama disease must be rigorously differentiated from other conditions manifesting as upper limb muscular atrophy, including amyotrophic lateral sclerosis, cervical spondylotic myelopathy, syringomyelia, intramedullary spinal cord tumors, and peripheral nerve compression syndromes. A definitive diagnosis can be achieved by integrating the characteristic age of onset and clinical presentation with dynamic cervical MRI findings and electrophysiological evaluations. Although HD is typically a self-limiting disorder and no definitive treatment currently exists to reverse established muscle weakness, early intervention is crucial. Timely management has been shown to halt disease progression and significantly minimize long-term functional disability. Conservative management remains the cornerstone of treatment for patients with HD.[12] The application of cervical collars, combined with physiotherapy, has been demonstrated to significantly decelerate disease progression and stabilize clinical symptoms. Recently, several studies have highlighted the potential benefits of surgical interventions, such as spinal decompression and cervical spinal fusion, in selected cases.[11,13] However, as surgical outcomes vary, a cautious approach to patient selection is imperative. In the present case, we recommended the consistent use of a soft cervical collar and advised the patient to refrain from upper-limb-dominant fitness activities. To date, the patient has exhibited modest functional improvement and achieved clinical stabilization.

In conclusion, it is essential to recognize that HD may initially present with asymmetric, irregular, and polyminimyoclonic finger tremors. Furthermore, characteristic cervical spine MRI findings may be absent during the early stages of the disease. Therefore, clinicians should maintain a high index of suspicion when evaluating young patients with atypical myoclonic tremors. Such presentations, which may initially be misidentified as functional (psychogenic) movement disorders, should be carefully evaluated as potential early clinical manifestations of HD.

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

Author Contributions: S.K., E.A.: Idea consept, control/supervision, design, data collection and/or interpretation, literature review, critical review, references and fundings, materials; S.K.: Analysis and/or interpretation, writing the article.

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