International Journal of Science and Research (IJSR)
ISSN: 2319-7064
Impact Factor 2025: 7.089

Bilateral Hirayama Disease Presenting as Slowly Progressive Distal Upper Limb Amyotrophy: A Rare Case Report

Dr. Bollipalli Pavan Teja Chowdhary[1], Dr. Aman Niranjan Agrawal[2], Dr. Abhijit Ramdas Rane[3]

[1]Junior Resident 3rd Year, General Medicine, Government Medical College and Maharashtra, Post Graduate Institute of Medical Education and Research, Nashik
Email: *pavan1517151[at]gmail.com*

[2]Junior Resident 3rd year, General Medicine, Government Medical College and Maharashtra, Post Graduate Institute of Medical Education and Research, Nashik
Email: *agrawalaman61998[at]gmail.com*

[3]Assistant Professor, General Medicine, Government Medical College and Maharashtra, Post Graduate Institute of Medical Education and Research, Nashik
Email: *abhijitrane73[at]gmail.com*

Abstract: ***Hirayama disease (HD), also known as juvenile muscular atrophy of the distal upper extremity, is a rare benign lower motor neuron disorder caused by dynamic compression of the lower cervical spinal cord during neck flexion. It predominantly affects adolescent and young adult males and typically follows a self-limiting course after 3–5 years. We report a 45-year-old male who presented with a 25-year history of gradually progressive bilateral distal upper limb weakness and marked hand muscle wasting. The patient experienced increasing difficulty in performing fine motor activities, including writing, buttoning clothes, and lifting objects, while proximal muscle strength, lower limb function, sensory examination, cranial nerves, and cerebellar function remained preserved. Electrophysiological studies demonstrated chronic anterior horn cell involvement at the bilateral C7–T1 segments, with fasciculations and chronic neurogenic motor unit changes. Magnetic resonance imaging of the cervical spine demonstrated findings consistent with Hirayama disease. This case is unusual because of its bilateral involvement and prolonged progressive course extending over two decades, unlike the classical self-limiting pattern. It highlights the importance of considering Hirayama disease in the differential diagnosis of distal upper limb amyotrophy and emphasizes the role of electrophysiological studies and dynamic cervical magnetic resonance imaging in establishing an accurate diagnosis.***

Keywords: Hirayama disease, Juvenile muscular atrophy of the distal upper extremity, Monomelic amyotrophy, Anterior horn cell disease, Dynamic cervical magnetic resonance imaging (MRI), Distal upper limb amyotrophy, Electromyography (EMG), Cervical flexion myelopathy, Bilateral Hirayama disease, Lower motor neuron syndrome

1. Introduction

Hirayama disease is a rare neurological disorder involving selective degeneration of the lower cervical anterior horn cells. It primarily affects adolescents and young adults, with a marked male predominance. The disorder is believed to result from repeated compression of the lower cervical spinal cord during neck flexion, producing chronic ischemic injury to the anterior horn cells. As a consequence, patients develop slowly progressive weakness and wasting of the distal muscles of the upper limbs, while sensory function, pyramidal tract signs, and lower limb involvement are generally absent.

Because of its rarity and clinical resemblance to other motor neuron disorders, Hirayama disease is frequently overlooked during the early stages of evaluation. Electrophysiological studies typically demonstrate chronic denervation confined to the C7–T1 myotomes, and dynamic flexion magnetic resonance imaging is considered the most useful investigation for confirming the diagnosis. Early recognition is clinically relevant because timely conservative management, including avoidance of excessive neck flexion and cervical immobilization in selected patients, may reduce further neurological deterioration.

The present report describes an unusual case of bilateral Hirayama disease with a remarkably prolonged 25-year clinical course. The atypical duration of progression and bilateral distribution expand the recognized clinical spectrum of this disorder and emphasize the need to consider Hirayama disease in patients presenting with chronic distal upper limb amyotrophy despite an extended duration of symptoms.

2. Case Report

Patient Name: Ramesh
Age/Sex: 50-year-old Male
Address: Nashik

The patient presented on 2nd June 2026 with complaints of progressive weakness and wasting of both hands for approximately 25 years. The illness began insidiously around 25 years back with difficulty performing activities requiring fine hand movements. Initially, he experienced reduced grip strength and difficulty lifting heavy objects. Over the following years, he developed increasing difficulty in writing, buttoning clothes, opening bottle caps, holding small objects, and performing other activities requiring manual dexterity. The weakness progressed slowly and was associated with

gradual thinning of both hands and distal forearms, leading to significant functional impairment.

There was no history of sensory symptoms such as numbness, tingling, or pain. He denied neck pain, radicular symptoms, bowel or bladder disturbances, cranial nerve symptoms, respiratory complaints, trauma, fever, or constitutional symptoms. There was no weakness involving the proximal muscles of the upper limbs or the lower limbs. There was no similar illness among family members.

On examination, the patient was conscious, alert, and oriented. Higher mental functions were normal. Cranial nerve examination was unremarkable. Motor examination revealed marked bilateral wasting of the thenar, hypothenar, first dorsal interosseous, and distal forearm muscles with weakness of hand grip, finger abduction, thumb opposition, and fine finger movements. Proximal muscle power in both upper limbs was preserved (Medical Research Council grade 5/5), and lower limb strength was normal. Muscle tone was normal. Deep tendon reflexes were preserved and symmetrical, with bilateral flexor plantar responses. Sensory examination, cerebellar examination, and gait were normal.

Initial Investigations (Day of Admission)

- Hemoglobin: **14.2 g/dL**
- Total Leukocyte Count: **7,600 cells/mm³**
- Platelet Count: **2.45 × 10⁵/mm³**
- Blood Urea: **28 mg/dL**
- Serum Creatinine: **0.9 mg/dL**
- Serum Sodium: **139 mmol/L**
- Serum Potassium: **4.2 mmol/L**
- Serum Chloride: **102 mmol/L**
- Aspartate Aminotransferase (AST): **26 U/L**
- Alanine Aminotransferase (ALT): **24 U/L**
- Alkaline Phosphatase: **86 U/L**
- Total Bilirubin: **0.8 mg/dL**
- Thyroid-Stimulating Hormone (TSH): **2.1 µIU/mL**
- Free T4: **1.2 ng/dL**
- Vitamin B12: **520 pg/mL**
- Fasting Blood Glucose: **92 mg/dL**
- Serum Calcium: **9.3 mg/dL**

Electrophysiological Studies (EMG/NCS)

Nerve conduction studies of both upper limbs demonstrated:

- Sensory nerve action potentials (SNAPs) of both median and ulnar nerves showed delayed onset latencies with preserved amplitudes and conduction velocities.
- Compound muscle action potentials (CMAPs) of both median nerves and the left ulnar nerve showed delayed distal motor latencies with preserved conduction velocities.
- Right median CMAP was not recordable.
- F-wave latencies from both median nerves were prolonged and persistent.

Needle Electromyography (EMG)

Needle EMG was performed in the **bilateral Abductor Pollicis Brevis (APB)** and **Abductor Digiti Minimi (ADM)** muscles and revealed:

- Fasciculation potentials
- High-amplitude motor unit action potentials (MUAPs)
- Polyphasic MUAPs
- Reduced interference pattern
- Findings consistent with **chronic neurogenic denervation and reinnervation**

Impression

The electrophysiological study was **suggestive of a chronic proximal neurogenic lesion involving the bilateral C7, C8, and T1 anterior horn cells**, consistent with the **Hirayama Disease.**

...EMG/NCV study conducted for both upper limbs...

SNAPs recorded from both median and ulnar nerves were of delayed onset latencies, normal amplitude and conduction velocity.

CMAPs recorded from both median and left ulnar nerves were of delayed distal latencies, amplitude and conduction velocity.

CMAPs could not be recorded from right median nerve.

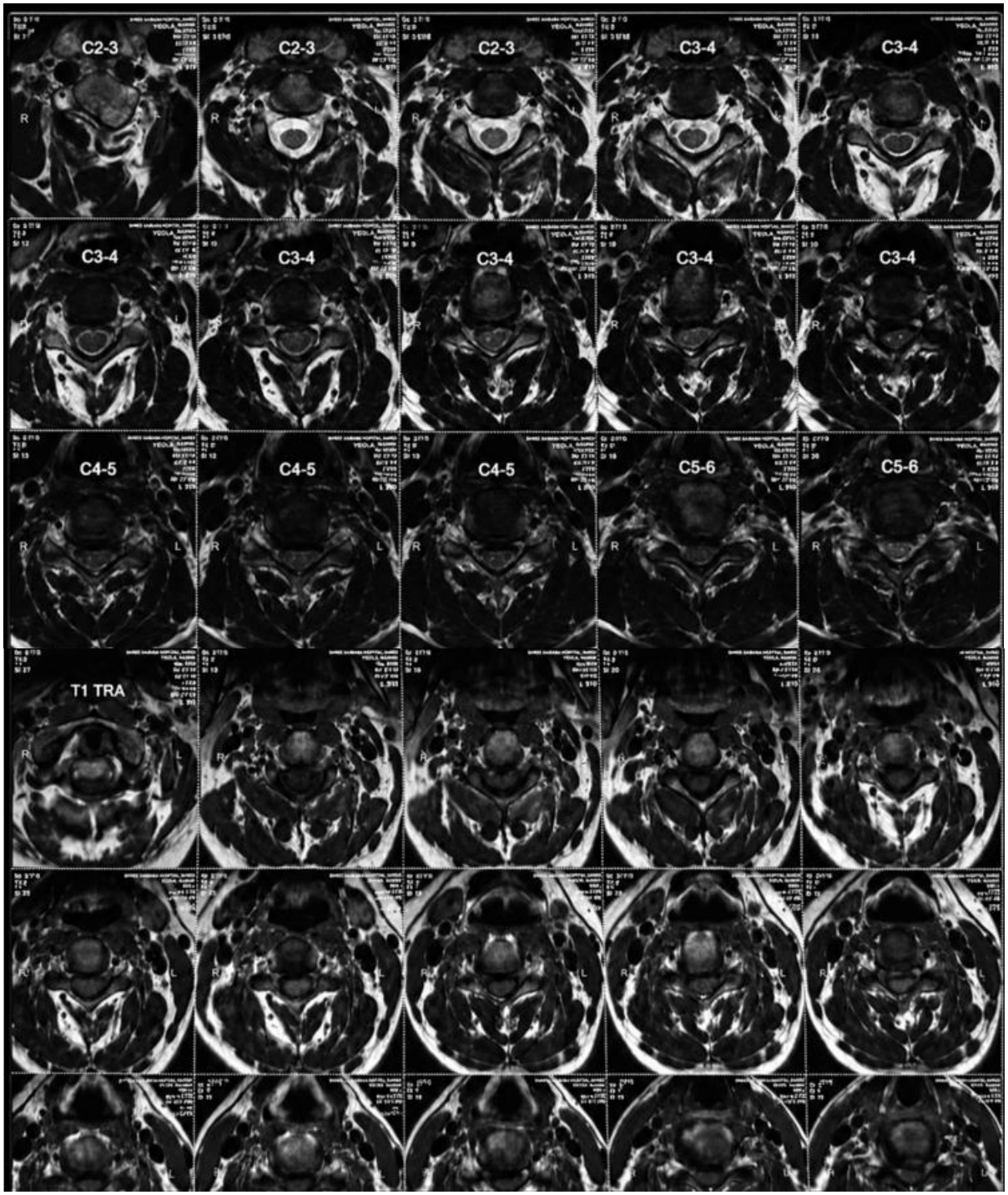
F waves recorded from both median nerves were of delayed latencies and were persistent.

EMG study was performed in both APB and ADM. Fasciculations were recorded. MUAPs recorded were of high amplitude, polyphasic with reduced interference pattern.

IMP :- This EMG/NCV study is suggestive chronic proximal neurogenic lesion at bialter C7C8T1 anterior horn cell level , consistent with clinical diagnosis of Hirayama's disease . To correlate clinically .

Imaging Studies

Magnetic resonance imaging of the cervical spine demonstrated lower cervical spinal cord changes compatible with Hirayama disease. Dynamic flexion cervical MRI showed anterior displacement of the posterior dural sac with compression of the lower cervical spinal cord and widening of the posterior epidural space, supporting the diagnosis.



Management

The patient was advised to avoid excessive neck flexion and was initiated on conservative management with a cervical collar, physiotherapy, occupational therapy, and hand muscle strengthening exercises. He was counselled regarding the benign but slowly progressive nature of the disease and advised regular neurological follow-up to monitor clinical progression and functional status.

3. Discussion

Hirayama disease is a rare, non-progressive or slowly progressive lower motor neuron disorder that predominantly affects the distal muscles of the upper limbs. The condition is characterized by selective involvement of the anterior horn cells of the lower cervical spinal cord, most commonly affecting the C7, C8, and T1 myotomes. It predominantly occurs in young males during the second and third decades of life and usually manifests with unilateral or asymmetrical distal upper limb weakness and muscle wasting. Bilateral involvement, as observed in our patient, is considerably less common and may create diagnostic uncertainty.

The proposed mechanism of Hirayama disease involves repeated forward displacement of the posterior cervical dural sac during neck flexion, resulting in transient compression of the lower cervical spinal cord. Chronic repetitive compression produces ischemic injury to the anterior horn cells supplied by the anterior spinal artery, leading to denervation and subsequent muscle atrophy. This mechanism explains the

selective motor involvement with preservation of sensory function and the absence of upper motor neuron signs.

Our patient presented with a remarkably slow progression of bilateral hand weakness and muscle wasting over approximately 25 years. The disease initially affected fine motor activities such as writing, buttoning clothes, and gripping objects before gradually interfering with routine daily activities. Despite the prolonged duration of illness, there was no involvement of the lower limbs, cranial nerves, sensory pathways, or autonomic functions. This clinical pattern is highly suggestive of a focal anterior horn cell disorder rather than a generalized motor neuron disease.

Electrophysiological evaluation played a crucial role in establishing the diagnosis. Nerve conduction studies demonstrated preserved sensory nerve action potentials with predominantly motor abnormalities, while needle electromyography revealed fasciculations, large-amplitude polyphasic motor unit action potentials, and reduced recruitment, indicating chronic denervation and collateral reinnervation involving the bilateral C7–T1 myotomes. These findings are characteristic of chronic anterior horn cell involvement and support the diagnosis of Hirayama disease.

Magnetic resonance imaging of the cervical spine further strengthened the diagnosis by demonstrating lower cervical spinal cord abnormalities consistent with Hirayama disease. Dynamic flexion imaging is particularly valuable because it demonstrates anterior displacement of the posterior dural sac with resultant compression of the lower cervical spinal cord, which is considered the radiological hallmark of the disease.

Several conditions may mimic Hirayama disease, including amyotrophic lateral sclerosis, multifocal motor neuropathy, cervical spondylotic amyotrophy, spinal muscular atrophy, and syringomyelia. In the present case, the absence of sensory deficits, preserved lower limb function, lack of upper motor neuron signs, characteristic electrophysiological findings, and supportive cervical imaging favored the diagnosis of Hirayama disease while making these alternative diagnoses less likely.

Although Hirayama disease is classically described as a self-limiting disorder that stabilizes within three to five years, our patient demonstrated an unusually prolonged clinical course extending over two decades. Such atypical presentations broaden the recognized clinical spectrum of the disease and emphasize the importance of maintaining a high index of suspicion in patients presenting with chronic distal upper limb amyotrophy. Early recognition is important because conservative measures, including avoidance of repeated neck flexion, cervical collar immobilization, physiotherapy, and rehabilitation, may help prevent further neurological deterioration and improve functional outcomes.

This case highlights the need to consider Hirayama disease in patients with isolated distal upper limb wasting, even when the duration of symptoms is unusually long or bilateral involvement is present. Careful clinical assessment combined with electrophysiological studies and dynamic cervical magnetic resonance imaging remains essential for establishing the diagnosis and differentiating this rare disorder from other anterior horn cell diseases.

4. Conclusion

Hirayama disease should be considered in patients presenting with insidious-onset distal upper limb weakness and muscle wasting, particularly when sensory function and upper motor neuron signs are preserved. Although the disorder classically affects young males and usually stabilizes after a limited period of progression, atypical presentations with bilateral involvement and prolonged disease duration may occur, creating a significant diagnostic challenge.

This case emphasizes the importance of careful clinical evaluation, detailed electrophysiological assessment, and cervical magnetic resonance imaging, including dynamic flexion sequences when available, in establishing the diagnosis and differentiating Hirayama disease from other anterior horn cell disorders. Early recognition is essential to avoid unnecessary investigations and inappropriate treatment. Increased awareness of atypical clinical presentations will facilitate timely diagnosis, appropriate conservative management, and improved long-term functional outcomes for affected patients.

References

[1] Hirayama K. Juvenile muscular atrophy of unilateral upper extremity: A new clinical entity. *Psychiatr Neurol Jpn.* 1959;61:2190–2197.
[2] Hirayama K. Non-progressive juvenile spinal muscular atrophy of the distal upper limb (Hirayama disease). In: de Jong JMBV, editor. *Handbook of Clinical Neurology.* Vol. 15. Amsterdam: Elsevier; 1991. p. 107–120.
[3] Tashiro K, Kikuchi S, Itoyama Y, Tokumaru Y, Sobue G, Mukai E, et al. Nationwide survey of juvenile muscular atrophy of distal upper extremity (Hirayama disease) in Japan. *Amyotroph Lateral Scler Other Motor Neuron Disord.* 2006;7(1):38–45.
[4] Hassan KM, Sahni H, Jha A. Clinical and radiological profile of Hirayama disease: A flexion myelopathy due to tight cervical dura. *Neurol India.* 2012;60(4):408–413.
[5] Pradhan S. Bilaterally symmetric Hirayama disease. *Neurology.* 2009;72(24):2083–2089.
[6] Raval M, Kumari R, Dung AA, Guglani B, Gupta N, Gupta R. MRI findings in Hirayama disease. *Indian J Radiol Imaging.* 2010;20(4):245–249.
[7] Chen CJ, Chen CM, Wu CL, Ro LS, Chen ST, Lee TH. Hirayama disease: MR diagnosis. *AJNR Am J Neuroradiol.* 1998;19(2):365–368.
[8] Sonwalkar HA, Shah RS, Khan FK, Gupta AK, Bodhey NK, Vottath S. Imaging features in Hirayama disease. *Neurol India.* 2008;56(1):22–26.
[9] Lehman VT, Luetmer PH, Sorenson EJ, Carter RE, Gupta V. Cervical spine MRI findings in Hirayama disease: Importance of dynamic flexion imaging. *AJNR Am J Neuroradiol.* 2013;34(4):974–980.
[10] Misra UK, Kalita J. *Clinical Neurophysiology.* 4th ed. New Delhi: Elsevier India; 2019. Chapter on anterior horn cell disorders and electrophysiological evaluation.