

Hirayama Disease: Clinical, Radiological, and Electrophysiological Spectrum in a Case Series of Six Patients, Including a Radiologically Occult Variant

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ABSTRACT

Hirayama disease (HD) is a rare, benign, focal cervical myelopathy affecting predominantly young individuals, characterized by insidious onset of distal upper limb weakness and wasting involving C7-T1 myotomes. The objective of this case series was to describe the clinical, radiological, and electrophysiological spectrum of Hirayama disease, including bilateral and radiologically occult presentations, and their short-term clinical outcomes. This case series of six patients (5 males, 1 female) evaluated at a tertiary neurology centre, highlighting the clinical spectrum, imaging features, and electrophysiological findings of HD. Patients were aged 16-29 years and presented with painless, progressive distal upper limb weakness with variable unilateral or asymmetric bilateral involvement. The characteristic “oblique atrophy” pattern with sparing of brachioradialis was observed in all cases. Fasciculations and tremors were noted in a subset, and a history of cold-induced exacerbation of symptoms was frequently reported. One patient demonstrated a radiologically occult variant with normal dynamic MRI despite typical clinical and EMG features. Dynamic cervical spine MRI in flexion revealed anterior displacement of the posterior dura with lower cervical cord flattening in most patients. Electromyography consistently demonstrated chronic neurogenic changes in C7-T1 myotomes, occasionally with contralateral involvement. All patients were managed conservatively with cervical collar immobilization, restriction of neck flexion, and cold avoidance, with clinical stabilization during follow-up. This series highlights the heterogeneous presentation of HD, including bilateral and radiologically occult variants, and emphasizes the complementary role of dynamic MRI and EMG in diagnosis. Early recognition is essential to avoid misdiagnosis and ensure appropriate conservative management.

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INTRODUCTION

Hirayama disease (HD) is a rare, benign, focal, cervical myelopathy first described by Keizo Hirayama and colleagues in 1959 in young Japanese males presenting with unilateral hand and forearm wasting.[1] Over subsequent decades, the condition has been reported across multiple Asian populations as well as in Western countries, although the prevalence remains highest in South and East Asia.[2,3] The eponymous condition is also referred to by a variety of descriptive terms including brachial monomelic amyotrophy (BMA), juvenile segmental muscular atrophy, juvenile muscular atrophy of distal upper extremity (JMADUE), and benign focal amyotrophy.[4]

The disorder predominantly affects young males between the second and third decades of life, with an estimated male-to-female ratio of approximately 10:1 to 20:1.[5] The cardinal clinical features are insidious onset, slowly progressive, painless muscle weakness and wasting confined principally to the forearm and intrinsic hand muscles, characteristically sparing the brachioradialis a feature termed 'oblique atrophy'. [6] Unilateral involvement is typical, though asymmetric bilateral upper limb changes may be observed in up to one-third of cases.[7] Though the upper limb is most commonly affected, lower limb involvement (benign calf amyotrophy) has been reported in a few cases. It is usually painless, with rarely any sensory symptoms noted. Patients often complain of tremors or twitching in the affected muscles (fasciculations). There is steady progression of weakness and atrophy noted for few months to years which is followed by stabilisation, mostly within 5 years of symptom onset.

Clinical examination shows LMN signs in the involved region. No UMN signs present. Sensory involvement is characteristically absent, although mild sensory symptoms are occasionally reported. Cranial nerves, pyramidal system are normal. No extra-pyramidal symptoms, no bowel/ bladder involvement, no autonomic symptoms noted.

The pathophysiological basis is understood to involve a mismatch between the growth of the vertebral column and the spinal cord/dural sac during adolescent growth spurts, leading to flexion-induced anterior cord compression and ischaemia of the lower cervical anterior horn cells; this mechanism is detailed further in the Discussion.[8] Dynamic MRI of the cervical spine in flexion and extension has become the principal diagnostic imaging modality, demonstrating posterior dural detachment and displacement, cord flattening, and segmental cord atrophy.[9]

Electromyography (EMG) corroborates clinical findings by demonstrating chronic neurogenic changes, including large amplitude, long duration motor unit potentials and reduced recruitment, in the distribution of the affected myotomes, with fibrillations and positive sharp waves in

acute phases.[10] Notably, subclinical contralateral involvement on EMG is frequently identified.

Symptoms often worsen in winter due to prolonged cold exposure, a phenomenon known as "cold paresis". They typically improve with warmer temperatures, possibly due to slowed nerve conduction and reduced blood flow, although the mechanisms remain unclear. The disease has no definitive cure, only attempts to minimize further damage to the cord are advised. Despite the importance of early recognition to institute cervical immobilisation the primary disease-modifying intervention HD remains under-recognised and frequently misdiagnosed as amyotrophic lateral sclerosis (ALS), hereditary spastic paraplegia, or other motor neuron disorders.[11] This six-case series from a tertiary neurology centre describes the clinical, radiological, and electrophysiological features of patients with confirmed HD, highlighting diagnostic challenges and the spectrum of disease presentation.

MATERIALS AND METHODS

This case series included six patients with a clinical diagnosis of Hirayama disease evaluated at a tertiary neurology centre. Clinical history and neurological examination were undertaken to document the pattern and distribution of weakness and wasting, lower motor neuron signs, brachioradialis sparing, fasciculations, and cold-induced symptom exacerbation. All patients underwent dynamic cervical spine magnetic resonance imaging using flexion-extension sequences to assess cord atrophy, flattening, and posterior dural displacement. Electromyography was performed to identify chronic neurogenic changes and determine the involved myotomes, including possible subclinical contralateral involvement. Patients were managed conservatively with cervical collar immobilisation or neck-flexion restriction and avoidance of cold exposure, and clinical status was assessed during follow-up. This study was approved by the Institutional Ethics Committee, Bombay Hospital-Indore, approval no. [BHI/DDMS/EC]2026-06:004, dated 20/6/2026]. Written informed consent was obtained from all patients included in this case series for the use of their clinical data and the clinical photographs reproduced in this manuscript. No patient-identifying information has been included.

CASE PRESENTATIONS

Case 1: Adolescent-Onset Right Upper Limb Dominant Presentation

A 16-year-old male with no prior medical history presented with an 8-9-month history of gradual-onset, progressive painless weakness of the right hand with difficulty performing activities of daily living, accompanied by progressive thinning of the right hand and forearm. There was no left-hand involvement by subjective report,

no sensory symptoms, and no lower limb symptoms. There was no history of trauma or neck pain.

Neurological examination revealed significant atrophy of the right hand and forearm with mild atrophic changes over the palmar aspect of the left hand. Power was 4/5 across the right elbow and wrist, and 2/5 in intrinsic hand muscles. The brachioradialis was preserved bilaterally. Deep tendon reflexes were reduced on the right with relative preservation on the left. No fasciculations, sensory deficits, bulbar weakness, cerebellar signs, or upper motor neuron (UMN) signs were observed.

Investigations: Dynamic MRI cervical spine (flexion-extension protocol) demonstrated changes consistent with Hirayama disease. EMG revealed chronic neurogenic changes on the left (contralateral) side.

Management and Follow-up: The patient was commenced on cervical collar immobilisation with instructions to restrict cervical flexion and avoid cold exposure. At follow-up, he reported mild subjective worsening during winter months; however, no objective neurological deterioration was identified on examination. Conservative management was continued.

Case 2: Bilateral Upper Limb Involvement with Fasciculatory Tremors

A 20-year-old male presented with a 7-month history of weakness of the right hand, progressive thinning of the ulnar aspect of the hand, and abnormal movements (fasciculatory tremors) of the right hand. Weakness had begun in the ring and little fingers and subsequently progressed to involve the entire hand and forearm. No left-hand symptoms were reported subjectively, and there were no sensory symptoms, lower limb involvement, neck pain, or history of trauma.

Examination demonstrated lower motor neuron (LMN) signs bilaterally (right greater than left), with marked wasting of the dorsal ulnar surface of the right hand and forearm. Motor power was 3/5 at the right wrist and fingers, and 4/5 at the right elbow. Brachioradialis power was relatively preserved. Deep tendon reflexes were reduced (1+) on the right. Fasciculatory tremors were evident in the right hand. No sensory deficits, bulbar, or UMN signs were elicited.

Investigations: MRI cervical spine with dynamic study showed focal cord atrophy in the lower cervical and cervico-dorsal junction with asymmetric cord flattening consistent with Hirayama disease. EMG demonstrated chronic neurogenic changes bilaterally (right > left).

Management and Follow-up: Conservative management with neck flexion restriction was instituted. Follow-up review showed no significant worsening of motor deficit, though a mild increase in tremor amplitude was noted. No new neurological findings were elicited.

Case 3: Left Upper Limb Presentation with Normal Neuroimaging

A 17-year-old male presented with a one-year history of gradually progressive, painless, purely motor weakness and wasting of the left upper limb, initially involving the hand and subsequently progressing to the forearm. There was no history of neck pain, trauma, lower limb involvement, or sensory symptoms.

Clinical examination revealed wasting and weakness specifically within the C7-T1 distribution of the left upper limb with LMN signs. Brachioradialis power was preserved. Adson's test was negative. No sensory involvement, bulbar involvement, UMN signs, or nerve thickening were detected. The right upper limb was neurologically intact.

Investigations: Dynamic MRI of the cervical spine with flexion and extension sequences was within normal limits, representing a recognised radiologically occult variant of Hirayama disease. EMG demonstrated LMN changes in the left C7-T1 anterior myotome distribution, consistent with monomelic amyotrophy.

Management and Follow-up: Supportive care was prescribed including cold exposure avoidance and cervical movement restriction. Follow-up assessment revealed worsening of weakness, particularly accentuated during winter months, without development of UMN signs or sensory features. Conservative treatment was maintained.

Case 4: Classic Cord Compression with Documented Dural Displacement

A 25-year-old male presented with a 3-month history of insidious onset, minimally progressive weakness of the right upper limb, causing difficulty with routine manual tasks. Subjectively, no symptoms were reported on the left side. The patient denied pain, sensory disturbance, or trauma. He noted subjective worsening during the winter season, followed by subsequent stabilisation.

Examination disclosed bilateral upper limb wasting and weakness (right > left), with distally predominant weakness (power 4/5 at the right wrist and fingers, 4+/5 at the right elbow). Fasciculatory tremors were observed in the right hand in the outstretched position. Brachioradialis power was preserved bilaterally. No sensory, bulbar, or UMN signs were identified.

Investigations: MRI cervical spine with dynamic protocol demonstrated anterior displacement of the posterior dura from C3 to C6 on the flexion sequence, causing anterior displacement of the underlying spinal cord with cord atrophy at the anterior aspect at C3-C5 levels. EMG confirmed chronic neurogenic changes affecting both upper limbs without lower limb involvement.

Management and Follow-up: A diagnosis of Hirayama disease was established and supportive treatment commenced with emphasis on cervical collar use, neck flexion restriction, and cold avoidance. Subsequent review demonstrated clinical stability.

Case 5: Bilateral Insidious Onset with Delayed Contralateral Progression

A 27-year-old male presented with a one-year history of insidious progressive thinning of the left hand followed by a 3-month history of right-hand thinning. He denied subjective weakness, lower limb involvement, dysphagia, voice change, sensory symptoms, neck pain, or trauma.

Examination demonstrated wasting involving the right hand more than the left. Motor power was 4/5 in the right hand and 4+/5 in the left hand, with normal power in the forearms and arms bilaterally. Deep tendon reflexes were normal. No fasciculations, UMN signs, sensory involvement, lower limb, or bulbar signs were elicited.

Investigations: Dynamic MRI of the cervical spine revealed focal cord atrophy in the lower cervical region with asymmetric cord flattening, more pronounced on the left consistent with Hirayama disease. EMG demonstrated asymmetric chronic neurogenic changes in both upper limbs.

Management and Follow-up: Conservative treatment was initiated. On follow-up, the patient reported mild increase in weakness now involving the left forearm, with preserved brachioradialis power bilaterally and no new right forearm involvement, consistent with expected natural history.

Case 6: Right UL presentation

A 29-year-old female presented with 4-year history of insidious onset, progressive weakness of right UL. She denied any symptoms on the left side. No sensory involvement, no bowel or bladder involvement. Occasional neck pain present. No history of any trauma.

Examination disclosed significant right UL atrophy, distal weakness 4/5 across wrist and finger joints. Fasciculatory tremors were seen in the right hand in the outstretched position. Brachioradialis power was relatively preserved. No sensory, bulbar or UMN signs were noted.

Investigations: MRI cervical spine with dynamic protocol showed subtle prominence of central canal in lower cervical and upper dorsal spine, but no epidural flow voids or any enhancement. EMG showed LMN changes in right C8-T1 anterior myotomal muscles- consistent with the diagnosis.

Management: A diagnosis of Hirayama disease was established and supportive treatment commenced with emphasis on cervical collar use, neck flexion restriction, and cold avoidance, with advice for close follow up to note for any development of new symptom.

Table 1 summarises the clinical, radiological, and electrophysiological characteristics of all six patients together with their management and follow-up outcomes.



Figure 1a



Figure 1b



Figure 1c

Figure 1a. Case 1: Palmar view of both hands demonstrating asymmetric intrinsic muscle wasting. Marked thenar and hypothenar atrophy is evident on the right hand compared to the relatively preserved left hand, consistent with C7-T1 myotome involvement.

Figure 1b Case 1: Resting posture of both hands showing characteristic clawing tendency of the right hand with loss of normal digital extension, reflecting weakness of the intrinsic hand muscles. The left hand maintains a comparatively normal posture.

Figure 1c Case 1: Clinical demonstration of brachioradialis power testing. The brachioradialis muscle bulk and power are preserved, producing the characteristic "oblique atrophy" pattern a hallmark clinical sign of Hirayama disease reflecting sparing of the C5-C6 myotomes.



Figure 2a



Figure 2b

Figure 2a Case 2: Bilateral upper limb examination showing asymmetric wasting of the forearms and hands, right greater than left. Wasting of the dorsal and ulnar musculature of the right forearm is visible, with comparatively milder involvement of the left side.

Figure 2b Case 2: Close-up dorsal view of both hands demonstrating prominent wasting of the right first dorsal interosseous and hypothenar eminence, consistent with ulnar-predominant intrinsic muscle denervation in the C8-T1 distribution.



Figure 3a



Figure 3b



Figure 3c

Figure 3a Case 3: Palmar view of the left hand demonstrating intrinsic muscle wasting with loss of normal thenar and hypothenar bulk. Mild flexion posturing of the fingers is noted, reflecting weakness of the intrinsic hand muscles in the C7-T1 distribution.

Figure 3b Case 3: Grip strength assessment of the left hand. Reduced grip strength with inability to fully flex the fingers against resistance is demonstrated, corresponding to the C8-T1 distribution weakness. The examiner's hand provides resistance to illustrate the degree of motor deficit.

Figure 3c Case 3: Bilateral upper limb examination in resting position showing asymmetric wasting of both forearms and hands, left greater than right. Marked slimming of the left distal forearm and hand is evident compared to the right, with loss of normal muscle bulk in the C7-T1 distribution. This image also illustrates the clinical pattern of predominantly unilateral involvement with subclinical contralateral changes.

**Figure 4a****Figure 4b****Figure 4c**

Figure 4a Case 4: Palmar view of both hands in standing position demonstrating bilateral intrinsic muscle wasting, right greater than left. Loss of normal palmar fullness with flattening of the thenar and hypothenar eminences bilaterally is evident, reflecting the asymmetric bilateral involvement characteristic of Hirayama disease.

Figure 4b Case 4: Dorsal view of both hands showing wasting of the first dorsal interosseous and other intrinsic muscles bilaterally, more pronounced on the right. Prominence of the extensor tendons due to loss of overlying muscle bulk is visible.

Figure 4c Case 4: Demonstration of brachioradialis power- preserved in this patient with Hirayama disease.

**Figure 5a****Figure 5b**

Figure 5a Case 5: Dorsal view of both hands showing intrinsic muscle wasting, left greater than right. Flattening of the first dorsal interosseous space and loss of normal metacarpal eminence fullness are evident bilaterally, consistent with the patient's bilateral presentation with left-dominant wasting.

Figure 5b Case 5: Palmar view of both hands demonstrating bilateral thenar and hypothenar muscle wasting. Loss of normal palmar fullness is more pronounced on the left, while the right shows milder but visible intrinsic muscle atrophy, illustrating the asymmetric bilateral pattern of Hirayama disease in this patient.

**Figure 6a****Figure 6b****Figure 6c**

Figure 6a Case 6: Palmar view of both hands demonstrating intrinsic muscle wasting on the right hand. Note the significant atrophy of the thenar muscles.

Figure 6b Case 6: Dorsal aspect of the right hand showing significant wasting in the first dorsal interosseous space

Figure 6c Case 6: Demonstration of the brachioradialis power in the affected limb- preserved.

Table 1: Summary of clinical features, investigations, management, and follow-up outcomes in six patients with confirmed Hirayama disease

Case	Age/ Sex	Duration	Affected Limb(s)	Brachioradialis	Clinical Features	MRI Findings*	EMG Findings*	Treatment	Follow-up
1	16M	8-9 mo	Right UL (bilateral wasting)	Preserved (right)	Wasting R hand/forearm; bilat- eral palmar atrophy; power 2/5 R hand; reduced DTR R; no fasciculations	Flexion-extension changes con- sistent with Hirayama disease	Chronic neu- rogenic changes (left side)	Cervical col- lar; neck re- striction; cold avoidance	Mild winter exacerbation; stable on con- servative Rx
2	20M	7 mo	Right UL > Left UL	Preserved	Wasting dorsal ulnar R hand/forearm; power 3/5 wrist/fingers; fas- ciculatory tremors R hand; LMN signs bilateral (R>L)	Focal cord atro- phy lower cervi- cal/cervico-dorsal junction; asym- metric cord flat- tening	Chronic neu- rogenic changes bi- lateral (R>L)	Conservative; neck re- striction	Mild tremor increase; no new deficits
3	17M	12 mo	Left UL	Normal	Wasting/weakness C7-T1 distribution; LMN signs; no sen- sory involvement; Adson negative	Normal flexion- extension MRI	LMN changes left C7-T1 anterior myo- tome; monomelic amyotrophy pattern	Supportive; cold avoid- ance; neck restriction	Increased weakness in winter; no UMN signs
4	25M	3 mo	Right UL > Left UL (bilateral wasting)	Preserved	Distal weakness power 4/5 wrist/fingers; fas- ciculatory tremors R hand; B/L UL wasting (R>L)	Anterior dis- placement poste- rior dura C3-C6 on flexion; cord atrophy C3-C5 levels	Chronic neu- rogenic changes both UL; no LL changes	Conservative; neck re- striction; cold avoidance	Stable after initial pro- gression
5	27M	12 mo (L); 3 mo (R)	Left UL > Right UL (bilateral)	Normal	Wasting R hand > L hand; power 4/5 L hand, 4+/5 R hand; forearm/arm power normal; no fascicula- tions	Focal cord atro- phy lower cervi- cal; asymmetric flattening more on left	Asymmetric chronic neu- rogenic changes both UL	Conservative; neck re- striction; cold avoidance	Mild L fore- arm weakness on follow-up; R forearm normal
6	29 F	4 yr	Right UL	Preserved	Wasting/weakness C8- T1 distribution; LMN signs; no sensory involvement; Adson negative	Subtle promi- nence of central canal in lower cervical and upper dorsal spine	LMN changes in right C8-T1 anterior myo- tomal muscles	Conservative; neck re- striction; cold avoidance	Stable

DTR = deep tendon reflexes; EMG = electromyography; LMN = lower motor neuron; MRI = magnetic resonance imaging; Rx = treatment; UL = upper limb; UMN = upper motor neuron

*All MRI/EMG were read by the same radiologist/electrophysiologist across cases.

DISCUSSION

Epidemiology and Clinical Spectrum: All six patients in this series were young adults aged 16-29 years, comprising five males and one female, consistent with the established demographic profile of Hirayama disease. Population-based studies suggest a peak incidence in the second and third decades, and the condition is significantly more prevalent in Asian males, consistent with prior reports from India, Japan, China, and Sri Lanka.[2,3,7] While the classic presentation is unilateral dominant upper limb wasting, this series demonstrates the full phenotypic spectrum ranging from a purely unilateral clinical picture (Cases 1, 3, 6) to subclinical contralateral EMG involvement (Cases 1, 2, 4, 5) and overt bilateral clinical wasting (Cases 2, 4, 5). The bilateral subclinical involvement detected electrophysiologically underlines that the condition should not be considered exclusively unilateral, and bilateral EMG sampling is

strongly recommended even in clinically unilateral cases.[7]

A notable feature in four of our six cases was cold paresis the subjective worsening of motor symptoms during cold weather. This phenomenon, attributed to temperature-dependent slowing of nerve conduction velocity, cold-induced vasoconstriction reducing already compromised anterior horn cell perfusion, and increased spinal cord compression through cold-related muscle stiffness, has been described as a highly characteristic, though subjective, historical feature of HD.[11,12] Recognition of cold paresis by clinicians can meaningfully accelerate the diagnostic pathway and should be specifically enquired about during history-taking in all young males presenting with distal upper limb weakness.

Pathophysiology: The prevailing hypothesis for HD pathogenesis implicates a dynamic, flexion-induced cervical myelopathy arising from disproportionate growth of

the osseoligamentous spinal column relative to the neural structures during adolescent growth spurts.[8] During neck flexion, the dural sac unable to accommodate this length discrepancy is displaced anteriorly, compressing the posterior cervical spinal cord. The consequent reduction in anterior horn cell perfusion produces focal ischaemia, primarily affecting the lower cervical segments (C7-T1) supplied by the sulcal arteries, which are recognised as watershed territories particularly vulnerable to microvascular compromise.[9]

Chronic repeated flexion-induced ischaemia leads to selective anterior horn cell loss, explaining the LMN phenotype confined to the C7-T1 myotomes with preservation of posterior column and corticospinal tract function. The characteristic brachioradialis sparing giving rise to the 'oblique atrophy' sign reflects the relative preservation of the C5-C6 root levels and their anterior horn populations.[6] Pathological studies in confirmed HD cases have demonstrated asymmetric cord flattening with marked anterior horn cell atrophy and secondary axonal degeneration.[1]

Diagnostic Considerations: The diagnosis of HD is primarily clinical, supported by neuroimaging and electrophysiological investigations. Dynamic MRI in the flexion position remains the single most informative investigation, demonstrating posterior dural detachment, an engorged posterior epidural venous plexus (crescent-sign), and spinal cord compression and atrophy.[9,13] In a systematic review, the sensitivity of dynamic MRI for HD diagnosis was reported to be 88-95% when flexion sequences were specifically obtained; neutral-position MRI alone significantly underestimates the degree of dural displacement.[5] Case 3 in the present series represents the radiologically occult variant wherein dynamic MRI is non-contributory which has been reported in approximately 10-15% of HD cases. The diagnosis in such patients depends on the clinical syndrome and EMG findings.[14]

EMG characteristically shows chronic neurogenic motor unit changes in the C7-T1 distribution, often with subtle contralateral involvement. In acute phases, fibrillation potentials and positive sharp waves may be superimposed. The distribution of changes helps differentiate HD from ALS (which may affect the lower limbs and show fasciculation potentials more widely), multifocal motor neuropathy (which typically shows conduction block), and hereditary spastic paraplegia.[10] Serum creatine kinase may be mildly elevated, reflecting ongoing denervation-atrophy, but is non-specific.

Differential Diagnosis: The differential diagnosis of HD includes amyotrophic lateral sclerosis (ALS), which must be specifically excluded as the clinical picture can overlap in the early stages. Key distinguishing features of HD are the self-limiting course, restriction to C7-T1 myotomes without bulbar or lower limb involvement, absence of UMN signs, and the young male demographic. ALS is characterised by progressive disease involving multiple levels, UMN and LMN sign coexistence, and a

universally fatal prognosis.[15] Other differential diagnoses include: cervical spondylotic myelopathy, multifocal motor neuropathy, Kennedy disease, hereditary motor neuropathy, and syringomyelia. [4]

As underscored in this series and in prior reports, rare cases initially diagnosed as HD have subsequently been reclassified as ALS on long-term follow-up.[15] For this reason, ongoing surveillance of all HD patients is recommended, with a low threshold for revisiting the diagnosis if unexpected UMN signs, bulbar features, or lower limb involvement emerge.

Management and Prognosis: Conservative management remains the cornerstone of HD treatment. The primary intervention is cervical immobilisation with a rigid or semi-rigid cervical collar to restrict neck flexion, thereby limiting repetitive cord compression. Studies have shown that sustained use of cervical orthosis for 3-4 years can stabilise or even modestly improve motor function, likely by halting progressive ischaemic injury.[16] Cold exposure avoidance is a simple but practically important adjunct, as evidenced by the winter exacerbation reported in four of six cases in this series.

Surgical options, including anterior cervical discectomy and fusion (ACDF) or posterior cervical decompression, have been reported but are generally reserved for patients with rapidly progressive disease refractory to conservative measures.[17] Evidence for surgical benefit is largely based on small case series, and neurosurgical intervention remains contentious given the self-limiting natural history of the disease. Importantly, decompressive surgery does not appear to restore lost motor function, though it may arrest further progression.[17]

The natural history of HD is broadly favourable. Disease activity typically plateaus within 3-5 years of onset, with the majority of patients achieving functional stability. However, residual muscle weakness and wasting persist to varying degrees, and patients should be counselled accordingly. Physical and occupational therapy, adaptive equipment, and patient education regarding cold avoidance and activity modification are important elements of long-term management.

LIMITATIONS

This case series is limited by its small sample size and retrospective single-centre design. MRI images from the original clinical report were unavailable for formal re-review; figure descriptions are based on reported findings. Formal disease severity scoring (e.g., Medical Research Council grading across all myotomes) and longitudinal quantitative muscle MRI data were not obtained. Genetic studies and serum biomarkers were not systematically performed. Prospective multicentre studies with standardised assessment protocols and long-term follow-up are needed to better characterise the phenotypic spectrum and outcomes of HD in South Asian populations.

CONCLUSION

This series of six patients with Hirayama disease illustrates the heterogeneous clinical presentations encountered in a tertiary neurology setting, ranging from classic unilateral distal upper limb wasting to asymmetric bilateral involvement and a radiologically occult variant with normal dynamic MRI. The cohort included five males and one female, consistent with the typical demographic profile, and cold paresis was a frequent and notable feature. Dynamic cervical MRI in flexion-extension sequences, along with electromyography, remains the diagnostic cornerstone and should be performed in all suspected cases, as neutral-position MRI may be non-diagnostic. Conservative management with cervical collar immobilisation, restriction of neck flexion, and avoidance of cold exposure provided effective stabilization in most patients and may slow disease progression. The generally favourable natural history should be clearly communicated to patients. Continued follow-up is essential due to the rare possibility of evolution into motor neuron disease. Increased clinical awareness across specialties will facilitate early diagnosis and reduce misdiagnosis and unnecessary investigations.

Individual Author's Contribution: AT contributed to the study conception, study design, and manuscript preparation. PPB contributed to the study conception, study design, data collection, data analysis and interpretation, and manuscript preparation. VJS contributed to the study design.

Declaration of Non-use of generative AI Tools: This article was prepared without the use of generative AI tools for content creation, analysis, or data generation. All findings and interpretations are based solely on the authors' independent work and expertise.

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