**EAS Journal of Radiology and Imaging Technology**
*Abbreviated Key Title: EAS J Radiol Imaging Technol*
ISSN: 2663-1008 (Print) & ISSN: 2663-7340 (Online)
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

Volume-7 | Issue-4 | Jul-Aug-2025 | DOI: https://doi.org/10.36349/easjrit.2025.v07i04.002

**Case Report**

# Atypical Presentation of Hirayama Disease with Unusual Proximal Muscle Involvement and Rapid Progression to Distal Muscle: A Case Report

Dr. Harini V[1], Dr. Venkatraman Indiran[2*], Dr. Balambighai V[3]

[1]Post Graduate, Department of Radiodiagnosis, Sree Balaji Medical College and Hospital.
[2]Professor, Department of Radiodiagnosis, Sree Balaji Medical College and Hospital, 7, CLC Works Road, Chromepet, Chennai – 600044, India
[3]Assistant Professor, Department of Neurology, Sree Balaji Medical College and Hospital

**Article History**
Received: 15.05.2025
Accepted: 03.07.2025
Published: 05.07.2025

**Journal homepage:**
https://www.easpublisher.com

**Quick Response Code**

**Abstract:** Monomelic amyotrophy, known as Hirayama disease, is a rare neurological condition that mostly affects young men. It is characterized by distal muscle atrophy and asymmetric weakness in the upper limbs, which are typically brought on by dynamic cervical spinal cord compression. We report a rare case of Hirayama disease in 2024 who is a 27-year-old male patient presented with weakness of right upper limb for the past 3 months and wrist drop involving right hand for the past three weeks. Clinical examination, lab investigation, nerve conduction study and flexion MRI of cervical spine was done. MRI findings, particularly focal atrophy and asymmetrical spinal cord flattening at the C4-C6 level, were key in confirming the diagnosis and guiding treatment decision. Imaging findings played a pivotal role in the diagnosis and management of Hirayama disease in this patient. His condition improved due to early intervention with immunosuppressive therapy, physiotherapy and regular follow up.

**Keywords:** Hirayama; monomelic; amyotrophy; cord; atrophy; weakness.



## INTRODUCTION

Hirayama disease, or monomelic amyotrophy, is a rare neurological condition predominantly affecting young males between the ages of 15 and 25 years [1].This illness, initially identified by Dr. Hirayama in 1959, is characterized by asymmetric weakening and atrophy of the upper limb muscles that gradually proceed, usually affecting one side of the body [2].The condition is believed to be caused by dynamic mechanical compression of the cervical spinal cord during neck flexion, with primary focus on the lower cervical spinal cord. This compression is considered to produce ischemia of the anterior horn cells, leading to muscular weakening and atrophy [3].

Hirayama disease, common in Asian populations (especially in Japan and India), is underdiagnosed due to gradual onset. While self-limiting, delayed diagnosis can lead to significant functional disability despite its generally benign progression [4,5]. Hirayama disease typically presents with gradual onset of muscle weakness and atrophy in the distal upper limb, sparing biceps, triceps, and deltoid muscles. It is usually unilateral, with fasciculations, tremors, and cold paresis, and lacks sensory symptoms [2,6]. The primary diagnosis of Hirayama disease is clinical, supported by imaging. Cervical spine flexion MRI is the gold standard, showing localized atrophy, asymmetric cord flattening, and hyperintensity at C4–C6 levels. Nerve conduction tests and EMG help differentiate it from other motor neuron diseases [7]. Hirayama disease, though benign and self-limiting, can cause functional impairment if diagnosed late. Early intervention with physical therapy and cervical collars can prevent progression. Surgical intervention, like anterior cervical decompression, is rare but may be necessary in severe cases to preserve function [8].

This case highlights atypical onset of proximal muscle weakness with rapid progression to distal muscle weakness in Hirayama disease, complicating diagnosis and potentially delaying intervention. It emphasizes the need to consider this rare disorder in young patients with upper limb weakness, advocating for early diagnosis and better management to improve outcomes in such cases.

***Corresponding Author:** Dr. Venkatraman Indiran
Professor, Department of Radiodiagnosis, Sree Balaji Medical College and Hospital, 7, CLC Works Road, Chromepet, Chennai – 600044, India

## CASE PRESENTATION

A 27-year-old young male working as a physical labourer presented to the Neurology department in 2024 with weakness of right upper limb for the past 3 months and wrist drop involving right hand for the past three weeks. On detailed history taking, the patient revealed he had developed subacute onset of proximal right upper limb weakness in the form of difficulty in doing his professional activities three months ago, prompting a CT brain to rule out a stroke which was normal. Later, gradually he developed difficulty in doing daily tasks such as holding objects, mixing food, buttoning his shirt, and combing his hair along with intermittent numbness in the right upper limb. He denied any fever, visual disturbances, or unsteadiness. There was no bladder dysfunction, altered sensorium, or recent surgeries. His medical history included no chronic conditions like hypertension or diabetes.

On clinical examination, he was alert, well-nourished, and his vital signs were stable. Neurological assessment revealed right upper limb muscle atrophy, especially in the shoulder girdle muscles [Figure 1A]. There was no wasting in the hand muscles [Figure 1B]. There was mild distal hypotonia in the right upper limb. Muscle strength was 4/5 in proximal and 3/5 in distal muscles of the right upper limb, with normal strength (5/5) in the left upper limb and in bilateral lower limbs. Sensory examination showed reduced pain and touch sensation throughout the right upper limb, but there were no signs of incoordination and fasciculations.

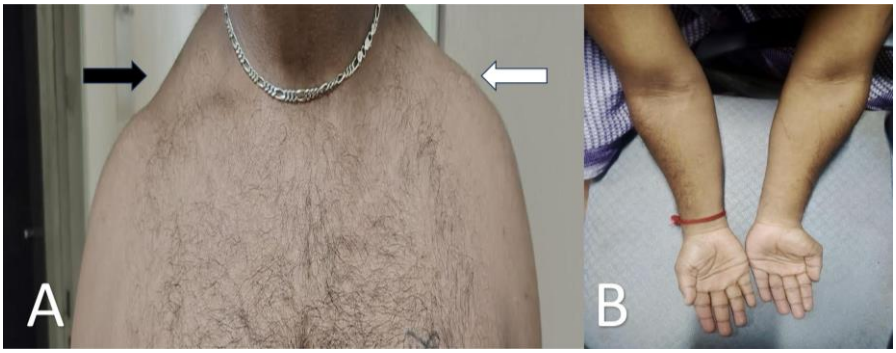


**Figure 1: Clinical photograph (A) showing right shoulder girdle muscle atrophy (black arrow) and normal left shoulder girdle muscles (white arrow). Clinical photograph (B) showing no obvious forearm and thenar muscle wasting**

Nerve conduction study revealed right axillary, musculocutaneous, and radial nerve sensorimotor neuropathy suggesting brachial plexus involvement. Cervical spine MRI showed focal T2 hyperintensity and asymmetrical right hemi-cord flattening at C4–C6 levels, consistent with Hirayama disease, but could not demonstrate forward falling of dura on flexion MRI [Figure 2A, 2B]. He was not willing for EMG/CSF analysis. Other investigations, including biochemical parameters, echocardiogram, ECG, ANA, anti-dsDNA and other autoimmune workup were within normal limits. The patient was diagnosed with right upper limb monoparesis, right wrist drop, and hemicord atrophy due to Hirayama disease. Due to rapid progression, he was treated with trial of pulse dose intravenous methylprednisolone and physiotherapy, resulting in significant improvement and he is under close follow up. The patient provided informed consent to the publication of this case.

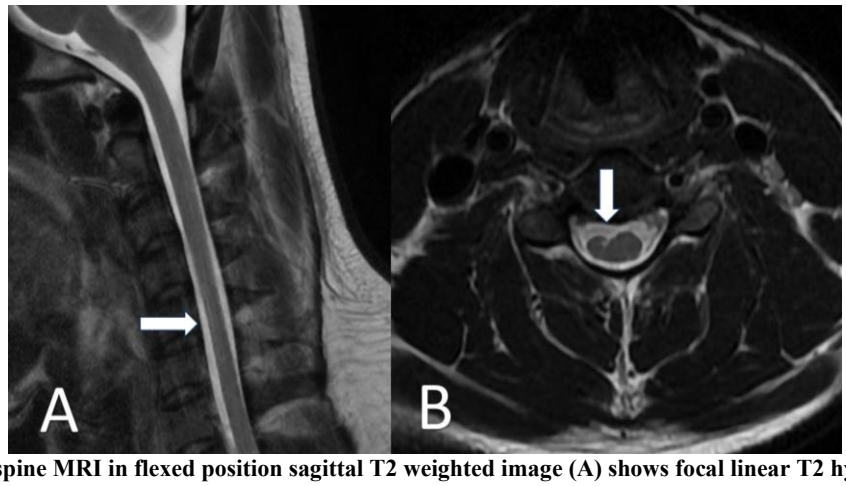


**Figure 2: Cervical spine MRI in flexed position sagittal T2 weighted image (A) shows focal linear T2 hyperintensity (white arrow). Axial T2 weighted image (B) shows asymmetrical right hemi-cord flattening at C5 level. No forward falling of posterior dura on the sagittal image**

## DISCUSSION

This case of Hirayama disease is proximal involvement and rapid progression to distal muscle and treatment was guided by both clinical presentation and investigative findings. The progressive weakness of the right upper limb, especially in the proximal muscles, combined with MRI evidence of focal cord atrophy at the C5-C6 level, pointed towards a diagnosis of Hirayama disease. The diagnosis of Hirayama disease in this patient was significantly aided by the crucial role of radiological imaging, particularly MRI of the cervical spine, which proved essential in differentiating it from other causes of upper limb weakness. MRI was pivotal in demonstrating focal volume loss at the C4-C6 level, with asymmetrical flattening of the right half of the spinal cord and minimal focal T2 hyperintensity. These characteristic radiological features strongly indicated Hirayama disease, which is a type of cervical myelopathy caused by dynamic cord compression during neck flexion.

Radiological findings play a central role in diagnosing this condition by detecting subtle structural abnormalities of the spinal cord that might be missed in routine clinical evaluations. Dynamic MRI, especially during flexion of the neck, is considered the gold standard for diagnosing this condition, as it reveals the forward displacement of the posterior dural sac and subsequent compression of the spinal cord though it was not present in our case.

## CONCLUSION

Imaging findings play a pivotal role in the diagnosis and management of Hirayama disease in this patient. MRI findings, particularly focal atrophy and asymmetrical spinal cord flattening at the C4-C6 level, were key in confirming the diagnosis and guiding treatment decisions. The collaboration between the radiology and neurology teams ensured timely and effective intervention, highlighting the importance of advanced imaging techniques in diagnosing rare neurological disorders with atypical presentation. Early radiological detection is essential for preventing disease progression and improving patient outcomes in cases of Hirayama disease.

**Conflict of Interest:** The authors declare no conflicts of interest related to the publication of this case report.

## REFERENCES


1. 1.Iacono S, Di Stefano V, Gagliardo A, Cannella R, Virzì V, Pagano S *et al.,* Hirayama disease: Nosological classification and neuroimaging clues for diagnosis. J Neuroimaging. 2022 Jul;32(4):596-603.
2. Hirayama K, Tsubaki T, Toyokura Y, Okinaka S. Juvenile muscular atrophy of unilateral upper extremity. Neurology 1963; 13:373-80.
3. Wang H, Tian Y, Wu J, Luo S, Zheng C, Sun C *et al.,* Update on the Pathogenesis, Clinical Diagnosis, and Treatment of Hirayama Disease. Frontiers in neurology. 2022 Feb 1;12: 811943.
4. Narayana Gowda B. S, Mohan Kumar J, Basim P. K. Hirayama's Disease - A Rare Case Report with Review of Literature. Journal of orthopaedic case reports, 2013 Jul-Sep;3(3):11–14.
5. Gomathy, Saranya B, Agarwal A, Garg A, Vishnu Venugopalan Y. Hirayama Disease: Review on Pathophysiology, Clinical Features, Diagnosis and Treatment. Touch REVIEWS in Neurology. 2022;18(2):109–16
6. Kira J, Ochi H. Juvenile muscular atrophy of the distal upper limb (Hirayama disease) associated with atopy. Journal of neurology, neurosurgery, and psychiatry. 2001,70(6), 798–801.
7. Kalekar T, Prabhu A. S, M Suhas. (2023). Cervical Spine Magnetic Resonance Imaging Findings in Hirayama Disease. Cureus. 2023 Jun 5;15(6): e40015.
8. Singh R, Hudson M, Meyer J. H, Neal M. T, Patel, N. P. Surgical treatment of spinal cord compression due to Hirayama disease: illustrative case. Journal of neurosurgery. 2022 Mar 7;3(10): CASE21697.9. Kayastha S. R, Pandey A, Pandey A, Keshari S, Pandey A. Hirayama disease successfully treated by posterior fixation: a case report. Annals of medicine and surgery 2024 Apr 23;86(6):3708–3712.


**Cite This Article:** Harini V, Venkatraman Indiran, Balambighai V (2025). Atypical Presentation of Hirayama Disease with Unusual Proximal Muscle Involvement and Rapid Progression to Distal Muscle: A Case Report. *EAS J Radiol Imaging Technol, 7*(4), 74-76.