



## Case Report

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# A Case Presentation on a Patient with the Familial Form of Monomelic Amyotrophy



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## Abstract

Monomelic Amyotrophy, also known as Hirayama disease, is a rare motor neuron disease that causes a painless, asymmetric weakness and atrophy in the distal upper extremities. The cause is unknown and it typically affects Asian males in their second or third decade. We report a case of a 21-year-old Bhutanese refugee with a three-year history of weakness and atrophy in his left upper extremity associated with a hand tremor and significant family history of similar findings in a sibling. Usually the disease is sporadic in nature, but the case we report is a familial presentation. This presentation was consistent with the clinical diagnosis and was accompanied by diagnostic imaging. Magnetic resonance imaging showed asymmetric atrophy of the spinal cord. With flexion positioning on both axial and sagittal planes, tightening of the dura along dorsal aspect of thecal sac from C3 to thoracic segments was observed.

**Keywords:** Monomelic amyotrophy; Hirayama disease; HirD

**Abbreviations:** MMA: Monomelic Amyotrophy; HirD: Hirayama Disease; UE: Upper Extremities; LUE: Left Upper Extremities; MRI: Magnetic Resonance Imaging; EMG: Electromyography

## Introduction

Monomelic Amyotrophy (MMA), also known as Hirayama disease (HirD), is a rare motor neuron disease typically affecting Asian males in the second or third decade [1-3]. Typical presentation is a painless, asymmetric weakness and atrophy in the distal upper extremities (UE)-usually the hand and forearm [3]. Current etiology is unknown, but low-grade vascular ischemia has been postulated [1,2]. We report a case of MMA with unilateral UE weakness and atrophy with findings on EMG and flexion/non-flexion MRI.

## Case Presentation: Monomelic Amyotrophy

A 21-year-old Bhutanese refugee presented with a three-year history of weakness and atrophy in his left upper extremity (LUE) along with a hand tremor worsening with movement. He denied paresthesia, pain, bulbar symptoms or prior history of neck trauma or infection. His brother had similar unilateral UE weakness progressing to paralysis.

Neurological examination revealed atrophy in left intrinsic hand and forearm muscles, measuring 13mm proximally and 15mm distally. Severe left-sided weakness was seen in the finger abductors, extensors, and flexors, moderate symptoms in wrist flexors and extensors, and subtle weakness on elbow

extension and flexion. Mild, asymmetric LUE hyperreflexia was accompanied by an absent Hoffman's sign in both hands with 2+, symmetric lower extremity (LE) reflexes with neither ankle clonus nor Babinski reflex. The remainder of the examination detected no apparent abnormalities. LUE Nerve conduction studies were within normal limits while electromyography (EMG) revealed chronic reinnervation in multiple cervical myotomes of the LUE with reduced recruitment, increased amplitude and prolonged duration in all but the deltoid muscles unilaterally. Increased insertional activity with mild spontaneous activity in the left biceps and triceps muscles was discerned. The findings were consistent with a chronic disorder of motor neurons, axons, or both-specifically affecting the LUE C7-C8-T1 myotomes. Needle examination in the left leg, right arm and bilateral cervical, thoracic, and lumbosacral paraspinal regions revealed normal findings.

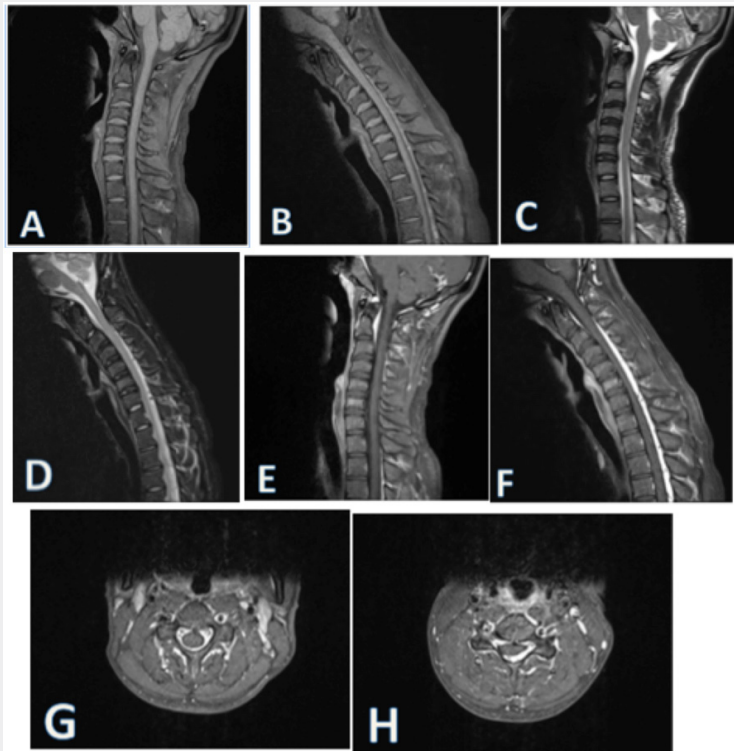
Complete blood count, comprehensive metabolic panel, and creatinine kinase were within normal limits while low-normal vitamin B12, copper, and vitamin E values were revealed at 211, 85, and 5.1, respectively. The patient was also found to have high and mildly elevated methylmalonic acid and homocysteine levels at 1180 and 14.2, respectively. Ceruloplasmin and angiotensin-

1-converting enzyme levels were within normal limits while human immunodeficiency virus and rapid plasma reagin antibody testing were negative.

### Cervical spine MRI non-flexion

Axial T1 and T2-weighted demonstrated asymmetrical atrophy of the left half of the cord. Flexion: Sagittal T1 pre- and post-contrast, and T2-weighted imaging showed prominent and

diffuse contrast enhancement of the enlarged and engorged dural venous plexus extending from C3 to upper thoracic segments. Tightening of the dura along the dorsal aspect of the thecal sac was accentuated on flexion positioning (both axial and sagittal) by diffuse narrowing of the thecal sac. MRI of brain and lumbar spine were unremarkable and all neuroaxes did not show any intramedullary lesion (Figure 1).



**Figure 1:** Nonflexion sagittal T1-weighted MRI, B. Flexion sagittal T1-weighted MRI, C. Nonflexion sagittal T2-weighted MRI, D. Flexion sagittal T2-weighted MRI, E. Contrast-enhanced nonflexion sagittal T1-weighted MRI, F. Contrast-enhanced flexion sagittal T1-weighted MRI, G. Axial MRI above the prominent dural venous plexus, H. Axial MRI at the level of the dural venous plexus.

EMG and comparison of non-flexion and flexion MRI confirmed MMA. Parenteral vitamin B12 and oral copper supplementation was started. Regular physical and occupational therapy with home exercise along with counseling against repetitive neck flexion activity were advised. Despite these measures, neither improvement nor progression of the disease was discerned during follow-ups at 3, 6 and 9 months.

### Discussion

MMA is a rare disorder of anterior horn cells affecting distal UE, predominantly in the C7-T1 myotomes, sparing any sensory deficits affecting young Asian males [1,4]. Unilateral asymmetric insidious progressive atrophy and weakness in the affected extremity are seen for 1-3 years. Symptoms then plateau and a coarse tremor may develop. Typically unilateral, bilateral involvements have also been observed [5]. Most presentations of this disease are thought to be sporadic with familial cases coming to light [5].

While the etiology of MMA is undetermined, a low-grade venous ischemia of the spinal cord, which lies in the watershed

area, possibly precipitated by trauma to the arm or neck, or by recurrent flexion and extension of his neck has been considered the causative factor [1]. MRI helps early on with detection of anteriorly displaced dura mater during neck flexion and changes in the curvature of the spinal cord. With disease progression, atrophy and asymmetric flattening of the lower cervical cord have been detected [4,6,7]. EMG studies have shown active as well as chronic denervation in the affected myotomes.

Our patient's presentation was consistent with both the clinical diagnosis as well as diagnostic imaging of MMA. However, unlike the typical sporadic nature, our patient's brother was also reported to suffer from the same illness (Familial Form).

### Conclusion

The patient's clinical presentation along with EMG, and MRI confirmed the diagnosis of MMA.

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