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Case Report

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Monomelic Amyotrophy (Hirayama Disease) with Proximal Upper Limb Involvement: A Case Report

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Abstract

Monomelic amyotrophy is an uncommon, benign, unilateral disorder of the lower motor neurons, affecting predominantly the hand and forearm muscles. A 28-year-old man presented with insidious-onset, slowly progressive, unilateral weakness and atrophy of his shoulder girdle and arm muscles on right side. A neurological examination revealed weakness and atrophy in his right deltoid, infraspinatus, supraspinatus and triceps muscles. Electromyography demonstrated an active and chronic neurogenic pattern affecting his right C5 and C6 myotomes; magnetic resonance imaging of his cervical spine showed snake eye appearance within the spinal cord. Upper limb proximal form of monomelic amyotrophy is a rare clinical entity with a wide differential diagnosis. Physicians, especially neurologists, should be familiar with this benign condition to avoid inappropriately labeling patients as having amyotrophic lateral sclerosis and other disorders with less favorable outcomes. *[Journal of National Institute of Neurosciences Bangladesh, 2018;4(1): 63-66]*

Keywords: Amyotrophic lateral sclerosis; electromyography; hirayama disease; magnetic resonance imaging; proximal monomelic amyotrophy

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Introduction

Monomelic amyotrophy (MA), also known as Hirayama disease, is a rare, benign lower motor neuron disease. Hirayama et al. originally reported this clinical entity in 1959, and called it "juvenile muscular atrophy of unilateral upper extremity"[1]. This disease is characterized by muscle wasting and weakness, affecting predominantly the lower cervical myotomes[2]. It affects mostly young males in their teens and twenties. The disease is more prevalent in India, Japan, and other Asian countries, but many cases have been reported from other parts of the world as well[3-5]. One report from India found that MA comprises approximately 12.8% of lower motor neuron diseases[5]. MMA typically exhibits unilateral or asymmetric weakness and atrophy of the distal upper extremity. But sensory disturbance, reflex change and upper motor neuron (UMN) signs are rare. Proximal involvement of the arm and shoulder muscles is an unusual presentation that has been rarely reported in the literature.

We have reported this case of a patient with MA, who presented with symptoms and signs in his proximal upper limb, a location rarely described in this disease[5-7].

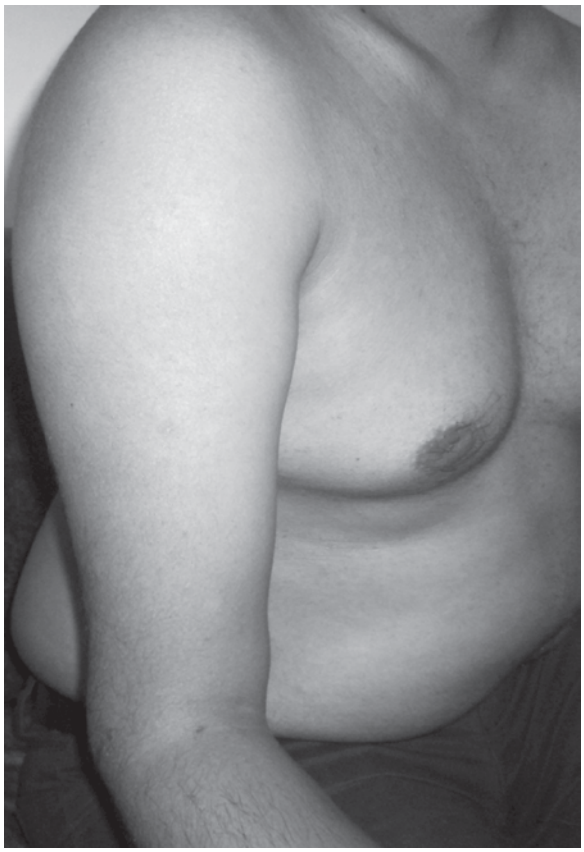

Figure I: wasting of the triceps and mild wasting of deltoid muscle

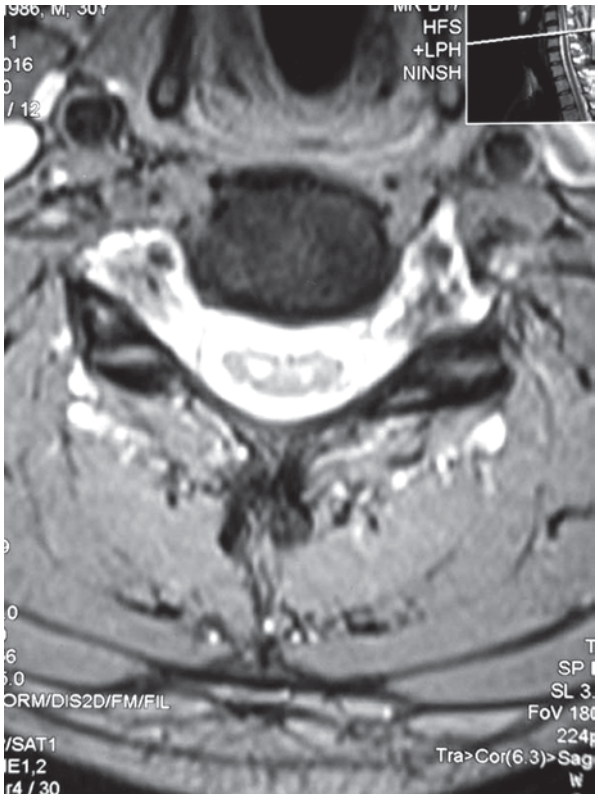

Figure II: Axial View of Cervical Spine Showing Snake Eye Appearance within the Spinal Cord

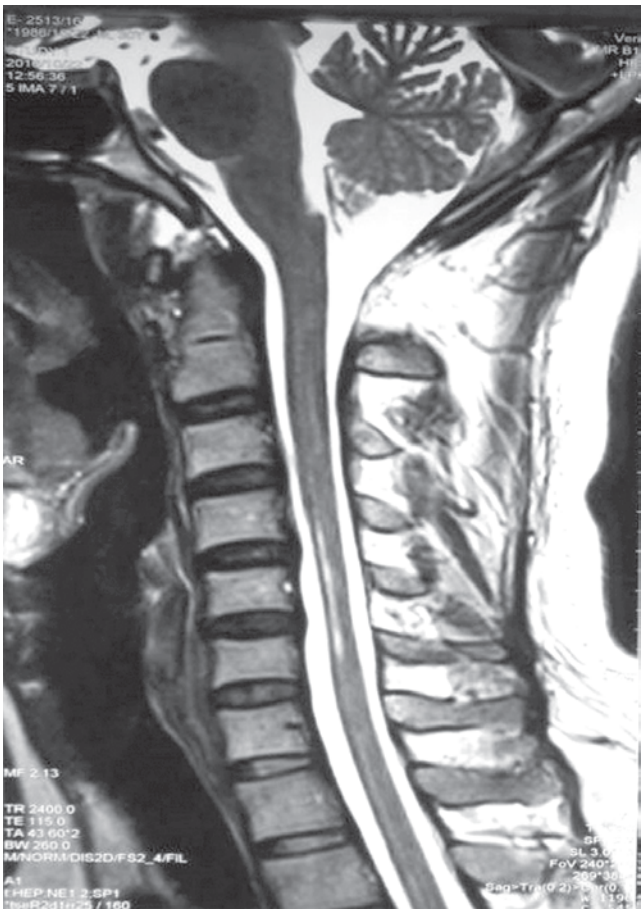

Figure III: Sagital view of cervical spine showing linear T2-hyperintensity noted in anterior part of spinal cord at C4-C6 level with mild atrophy of the cord at this Level

Case Presentation

A 30 years old man presented with weakness and wasting of right upper limb for 2 years. He also experienced weakness of right upper limb with difficulties in overhead activities which was gradually progressive. He noticed wasting of right upper limb muscles which involved shoulder girdle and arm muscle more than forearm and hand. He also noticed cramp on right hand especially after working in computer for prolonged period which increased in severity for last 6 months and occasional twitching of muscle over deltoid region. Apart from right upper limb he denied any complaint in other limbs. On query, there was no history of bowel and bladder involvement, dysphasia, trauma to the neck or fever. He had no family history of similar illness. On examination, there was no abnormality in general examination. Nervous system examination revealed a normal higher psychic function, cranial nerve including fundus revealed no abnormality. Motor examination of right upper limb showed wasting of deltoid and triceps muscle with a reduced tone. Power was 4/5 distally and 3/5 proximally. Biceps and triceps jerks were absent but

supinator jerk was present. Hoffman sign was absent. No sensory impairment or abnormal cerebellar sign was noted. Investigations revealed a normal complete blood count, EMG showed chronic disorder of motor neurons, their axons or both affecting right upper limb. Magnetic resonance imaging of his cervical spine showed snake eye appearance within the spinal cord.

Discussion

Monomelic amyotrophy, also known as Hirayama disease, is a lower motor neuron disease diagnosed overwhelmingly in adolescent males (>10:1 M:F; aged 15-25 years)[8]. The first cases were reported in 1959 in the Japanese population, and more recently, pediatric cases of Hirayama Disease have been found in North America[9].

It is characterized by insidious-onset, asymmetric, unilateral weakness and atrophy of the hand and forearm muscles, with sparing of the brachioradialis, giving rise to an appearance called 'oblique amyotrophy'[1,9-10]. Several case series have described predominant lower limb involvement[3,11]. MA typically affects males between the ages of 15 and 25 years[12]; however, it can occur in females as well[13-14]. The disease progresses slowly over several years, before reaching a stationary stage[3]. Bilateral, usually asymmetric, but also symmetric, 'bimelic' forms affecting the upper limbs has also been observed[3].

Our patient presented with an uncommon form of MA, affecting the shoulder and arm muscles instead of the commonly seen hand and forearm disease. Rare cases of proximal upper limb MA have been reported in the literature[4-7]. This patient did not have sensory symptoms or upper motor neuron signs during 2 years, or clinical or electrophysiological evidence for a widespread disease in bulbar or other limb muscles, indicating the benign nature of this condition.

The differential diagnosis of MA includes the distal form of spinal muscular atrophy, amyotrophic lateral sclerosis (ALS), post-polio syndrome, multifocal motor neuropathy with conduction block (MMNCB), as well as structural lesions of the cervical cord. These clinical entities can be identified by specific clinical, radiological and electrophysiological features.

The pathophysiology of MA remains unknown[4]; however, several postulations have been considered, such as viral infections, ischemia to anterior horn cells, and atrophy[6,9]. In 1987, Kikuchi et al[15] first proposed that a tight dural canal may be an underlying predisposing factor. Hirayama[9] suggested a model of focal venous ischemia due to compression and flattening of the lower cervical cord arising from forward displacement of the cervical dural sac and spinal cord, caused by recurrent neck flexion.

Conclusion

Monomelic amyotrophy should be suspected in patients presenting with slowly progressive weakness and atrophy restricted to one limb, followed by a static phase. While most reported cases involve the lower cervical myotomes, affecting the hand and forearm muscles, proximal upper limb involvement can be seen rarely. EMG and MRI studies are helpful in confirming the diagnosis and ruling out other clinical entities presenting in a similar fashion. Treatment is conservative in most patients, with the use of a cervical collar in appropriate cases, and physiotherapy.

References

1. Hirayama K, Toyokura Y, Tsubaki T. Juvenile muscular atrophy of unilateral upper extremity: a new clinical entity. Psychiatry Neurol Jpn. 1959; 61:2190–7
2. Peiris JB, Seneviratne KN, Wickremasinghe HR, Gunatilake SB, Gamage R. Non familial juvenile distal spinal muscular atrophy of upper extremity. J Neurol Neurosurg Psychiatry. 1989;52:314–9
3. Pradhan S. Bilaterally symmetric form of Hirayama disease. Neurology. 2009;72:2083–9
4. De Freitas MR, Nascimento OJ. Benign monomelic amyotrophy: a study of twenty-one cases. Arq Neuropsiquiatr. 2000;58:808–13
5. Neves MA, Freitas MR, Mello MP, Dumard CH, Freitas GR, Nascimento OJ. Benign monomelic amyotrophy with proximal upper limb involvement. Arq Neuropsiquiatr. 2007;65:524–7
6. Orsini M, Freitas MR, Catharino A, Mello MP, Nascimento OJ. Upper limb proximal form of monomelic amyotrophy: on purpose of 2 cases. Rev Bras Neurol. 2008;44:13–7
7. Yilmaz O, Alemdaroğlu I, Karaduman A, Haliloğlu G, Topaloğlu H. Benign monomelic amyotrophy in a 7-year-old girl with proximal upper limb involvement: case report. Turk J Pediatr. 2011;53:471–6
8. Nalini A, Gourie-Devi M, Thennarasu K, et al. Monomelic amyotrophy: Clinical profile and natural history of 279 cases seen over 35 years (1976-2010). Amyotrophic lateral sclerosis & frontotemporal degeneration 2014:1-9
9. Hirayama K. Juvenile muscular atrophy of unilateral upper extremity (Hirayama disease)--half-century progress and establishment since its discovery. Brain and nerve Shinkei kenkyu no shinpo 2008;60:17-29
10. Talbot K. Monomelic amyotrophy or Hirayama's disease. Pract Neurol. 2004;4:362–5
11. Hirayama K, Tomonaga M, Kitano K, Yamada T, Kojima S, Arai K. Focal cervical poliopathy causing juvenile muscular atrophy of distal upper extremity: a pathological study. J Neurol Neurosurg Psychiatry. 1987;50:285–90
12. Gourie-Devie M, Suresh TG, Shankar SK. Monomelic amyotrophy. Arch Neurol. 1984;41:388–94
13. Hirayama K. Juvenile muscular atrophy of unilateral upper extremity (Hirayama disease): half-century progress and establishment since its discovery. Brain Nerve. 2008;60:17–29
14. De Freitas MR, Nascimento OJ. Benign monomelic

amyotrophy: a study of twenty-one cases. Arq Neuropsiquiatr. 2000;58:808–13
15. Gourie-Devie M, Suresh TG, Shankar SK. Monomelic amyotrophy. Arch Neurol. 1984;41:388–94
16. Kikuchi S, Tashiro K, Kitagawa M, Iwasaki Y, Abe H. A mechanism of juvenile muscular atrophy localized in the hand and forearm (Hirayama's disease): flexion myelopathy with tight dural canal in flexion [in Japanese]. Rinsho Shinkeigaku. 1987;27:412–9