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

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[Received: 21 September 2017; Revised: 6 November 2017; Accepted: 11 December 2017; Published: 1 January 2018]

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

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”. This disease is characterized by muscle wasting and weakness, affecting predominantly the lower cervical myotomes. 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. One report from India found that MA comprises approximately 12.8% of lower motor neuron diseases. 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.
Monomelic amyotrophy (MA), also known as Hirayama disease, is a rare condition characterized by muscle wasting and weakness, affecting the 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 4-7. This patient did not have sensory disturbance, reflex change and bladder involvement, dysphasia, trauma to the neck or clinical or electrophysiological evidence for a widespread disease in bulbar or other limb muscles, 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

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). 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.

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’. Several case series have described predominant lower limb involvement. MA typically affects males between the ages of 15 and 25 years; however, it can occur in females as well. The disease progresses slowly over several years, before reaching a stationary stage. Bilateral, usually asymmetric, but also symmetric, ‘bimelic’ forms affecting the upper limbs has also been observed.

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. 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; however, several postulations have been considered, such as viral infections, ischemia to anterior horn cells, and atrophy. In 1987, Kikuchi et al first proposed that a tight dural canal may be an underlying predisposing factor. Hirayama 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**

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mostly young males in their teens and twenties. The disease, is a rare, benign lower motor neuron disease. Lower motor neuron diseases typically exhibit unilateral or asymmetric weakness and atrophy of the distal upper limb, a location rarely described in this disease. We have reported this case of a patient with MA, who presented with an uncommon form of MA, affecting the upper limbs. The pathophysiology of MA remains unknown. The differential diagnosis of MA includes the distal form of spinal muscular atrophy, amyotrophic lateral sclerosis, or clinical or electrophysiological evidence for a widespread disease in bulbar or other limb muscles, or other clinical entities can be identified by specific clinical, radiological and electrophysiological features.

Case Presentation

A 30 years old man presented with weakness and wasting of proximal upper limb MA have been reported in the literature. Our patient presented with an uncommon form of MA, involving shoulder girdle and arm muscle more than shoulder. He noticed cramp on right hand especially after working in computer for prolonged periods which increased in severity for last 6 months and he also noticed cramp on right hand which was gradually progressive. He also noted 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 symptoms or upper motor neuron signs during 2 years, but there was a complaint of cramp, weakness and wasting of right hand for last 6 months while using computer for prolonged periods. A 30 years old man presented with weakness and wasting of right upper limb for last 6 months. On examination, there was no abnormality in general eye appearance within the spinal cord. Investigations revealed a normal complete blood count, EMG showed chronic disorder of motor neurons, supinator jerk was present. Hoffman sign was absent. No sensory disturbance, reflex change and asymmetric weakness and atrophy of the distal upper limbs. On query, there was no history of bowel and bladder involvement of the arm and shoulder muscles is an unusual presentation that has been rarely reported in the literature 4-7. This patient did not have sensory disturbance, reflex change and asymmetric weakness and atrophy of the distal upper limbs. On query, there was no history of bowel and bladder.

Discussion

Hirayama Disease (HD) was first described by Hirayama in 1959, and called it “juvenile muscular atrophy of unilateral upper extremity. But sensory disturbance, reflex change and asymmetric weakness and atrophy of the distal upper limbs. On query, there was no history of bowel and bladder. Hirayama K, Toyokura Y, Tsubaki T. Juvenile muscular atrophy (Hirayama’s disease): flexion myelopathy with tight dural canal in flexion [in Japanese]. Rinsho Shinkeigaku. 1987;27:412–9. The overwhelming incidence of HD has been found in Japan and Korea, but there are cases of Hirayama Disease have been found in North America. Hirayama's disease progresses slowly over several years, before symptoms or upper motor neuron signs during 2 years, but there was a complaint of cramp, weakness and wasting of right upper limb for last 6 months while using computer for prolonged periods. A 30 years old man presented with weakness and wasting of right upper limb for last 6 months. On examination, there was no abnormality in general eye appearance within the spinal cord. Investigations revealed a normal complete blood count, EMG showed chronic disorder of motor neurons, supinator jerk was present. Hoffman sign was absent. No sensory disturbance, reflex change and asymmetric weakness and atrophy of the distal upper limbs. On query, there was no history of bowel and bladder.

References