Hirayama Disease: A Rare Case Report and Review

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Abstract
Hirayama disease, or brachial monomelic amyotrophy, is not a common neurological disease characterized by unilateral or asymmetric bilateral lower motor weakness of distal upper limbs. The basic pathophysiology is compression of the dural sac and spinal cord during flexion of the neck. A case of a 21-year-old male presented with chief complaints of tremors in both hands (right more than left) with gradually progressive weakness of the right hand and forearm. Electromyography (EMG), nerve conduction velocity (NCV), and magnetic resonance imaging (MRI) showed focal atrophy of lower cervical myotomes and confirmed the diagnosis of monomelic amyotrophy.

Introduction
Hirayama disease is an uncommon sporadic, self-limiting lower motor neurons disease; clinically restricted to the distal part of upper limbs, characterized by unilateral or bilateral (asymmetrical) weakness, and wasting of muscles (focal involvement) supplied by lower cervical cord predominantly C7, C8, and T1. It was first described in 1959 by Hirayama. The disease usually begins in the late teens and early 20s (15 and 25 years) with male preponderance (male: female ratio of 20:1).1

Diagnosis is based on clinical history, examination, neuroimaging (flexion MRI), and electrophysiological studies. Management mainly includes the administration of a cervical collar, muscle-strengthening exercises, and assistive devices like splinting and braces.1 We report a case of a 21-years male patient who presented with tremors in his right hand and bilateral asymmetrical (right > left) weakness and wasting of the upper limb. Electrophysiological studies and radiological investigations confirmed the diagnosis.

Case Description
A 21-year-old, right-handed gentleman with no prior comorbidities presented with complaints of tremors in both hands (in the right hand for 3 years and in the left hand for 2 years), right more than left, and gradually progressive distal weakness in the right hand for 2 years. The tremors were coarse, aggravated during finger extension, and were associated with transient worsening during winters. The hand weakness resulted in difficulty in carrying out day-to-day activities like fine movements at hand, including holding a pen, buttoning, unbuttoning, and tying shoelaces. Consequently, he had to discontinue his studies because of difficulty in writing. There was no history of any weakness in the lower limbs. Also, he had no history of pain, sensory loss, dysphagia, dysarthria, ptosis, diplopia, headache, cramps, fasciculations, or bowel or bladder dysfunction. History of any fever, trauma, poliovirus infection history and any occupational exposure to any heavy metals or toxins was ruled out. He has no history of similar neurological disorders in any other family member.

On examination, vitals were stable, and Mini-mental State Examination of 28/30. Central nervous system examination shows the bulk of the flexor aspect of the right forearm was reduced along with wasting of the thenar and hypothenar eminences. The tone was normal in all the limbs. The power was decreased (3/5) in the thenar, hypothenar, flexors, and extensors of the right forearm. Right brachioradialis was spared. Power in the left hand, forearm, shoulder, and right arm and shoulder, as well as in the bilateral lower limb, was normal. Tremors (minipolymyoclonus-like movement) were present in both hands (right > left). Fasciculations were seen around the right elbow. Deep tendon reflexes (DTR) were normal in all four limbs, and coordination was normal. No evidence suggestive of the sensory, autonomic system, cranial nerve, posterior column, and cerebellum involvement was present. The gait and stance were normal.

On investigations—complete blood count, kidney function tests, and liverfunction tests were normal. Thyroid-stimulating hormone was 1.55 µIU/mL, vitamin B12; 770 pg/mL, vitamin D; 30 ng/mL, and folate; 10.8 nmol/L. The extended autoimmune profile (antinuclear antibody, anti-double-stranded deoxyribonucleic acid, anti-histone, anti-Ro, anti-La, and anti-scleroderma) was negative. Erythrocyte sedimentation rate, C-reactive protein, rheumatoid factor, creatine kinase, and creatine kinase-myoglobin binding were normal. Tridot tests, including hepatitis B, hepatitis C, and human immunodeficiency virus, were nonreactive.

Nerve conduction studies showed normal distal latency with decreased amplitude, and conduction velocities were normal in the right ulnar nerve. The right median left ulnar, left radial, and left median nerve showed normal amplitude, normal conduction velocities, and normal latency. F-wave was normal in all the nerves tested. Sensory nerve conduction study was normal.

Electromyography (EMG) revealed preganglionic neurogenic involvement of the C5-T1 segments on the right side with evidence of denervation and chronic reinnervation changes. A plain MRI brain and cervical spine revealed no abnormality. During flexion of the neck, MRI cervical spine revealed anterior displacement of the posterior theca such that it compresses the cord between it and the spinal column anteriorly from C4-C6 with a prominence of epidual venous dural space dorsal to dura with mild cord thinning from C4-C6. The posterior dural sac crescent appeared in T2/short-tau inversion recovery (T2/STIR) as a hyperintense entity and enhanced uniformly postcontrast (Figs 1A and B).

On the basis of clinical examination, electrophysiological, and radiological studies, the patient was diagnosed with monomelic amyotrophy, and he was prescribed a cervical collar so that further spinal cord injury due to repeated flexion of the neck could be prevented. Muscle-strengthening exercises were also advised. The patient’s complaints have been stable for the past 6 months with no new symptoms.

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**DISCUSSION**

Hirayama disease, also called monomelic amyotrophy, is not a common neurological entity affecting young males more than females in their late teens and early 20s. The condition is usually self-limiting, and characteristically involvement is limited to a few myotomes in the extremity, which is affected. This results in unilateral involvement and atrophy of muscles innervated by C7, C8, and T1, which travels obliquely along the volar and dorsal aspect of the forearm to the hands and forearms muscles with brachioradialis muscle being spared ("oblique amyotrophy" pattern). Disorder has also been termed benign focal amyotrophy, juvenile asymmetric segmental spinal muscular atrophy, and juvenile muscular atrophy of the distal upper extremity in the literature. It usually manifests as gradual onset muscle weakness and atrophy that can be unilateral or bilateral. Bilateral muscle weakness and atrophy (symmetrically or asymmetrically) are seen only in 10% of cases and were present in this case also.1,2

In neurological examination, there are absent or hypoactive muscle stretch reflexes or DTR in the muscles supplied by the spinal cord segment, which is involved with no upper motor neuron signs. The cranial nerves, pyramidal tracts, and autonomic nervous system are normal. Autonomic involvement, for example, cold skin, cold paresis (i.e., increase in weakness on exposure to cold environment), excessive sweating, bilateral minipolymyoclonus, and hair loss over the dorsum of the hands has been reported in 36% of cases. The progression of disease in terms of trophic changes is steady for the first 2–3 years; after that, most of the patients within 5 years stabilized. In about 20% of cases, the spread may be observed in the contralateral limb also.1

Proposed diagnostic criteria by Tashiro et al. for the Hirayama disease include (1) distal weakness and muscular atrophy in the forearm and hand; (2) unilateral upper extremity; (3) onset at the age between 10 and 20; (4) insidious onset with gradual progression for the first few years, followed by stabilization; (5) no involvement of the lower limbs; (6) No sensory disturbance and tendon reflex abnormalities; (7) exclusion of other diseases (motor neuron disease, multifocal motor neuropathy, spinal cord tumors, syringomyelia, abnormalities of cervical vertebral, anterior interosseous, or deep ulnar neuropathy).2 The above criteria were fulfilled in this reported case.

The neurological examination revealed lower motor neuron type of weakness of the hand and forearm muscles of the right hand with sparing of brachioradialis muscle without any sensory dysfunction. Neuroimaging and EMG studies confirmed the clinical diagnosis of Hirayama disease (diagnosis of amyotrophic lateral sclerosis (ALS) was ruled out due to segmental cervical atrophy instead of diffuse atrophy as seen in ALS).1

Electromyography (EMG) shows evidence of chronic denervation of affected muscles, with or without acute denervation potentials (in the form of positive sharp waves, fasciculations, and fibrillation potentials). EMG findings may be abnormal in seemingly healthy muscles.2 EMG, in this case, also showed almost the same findings.

Findings of MRI cervical spine (during neck flexion) are one of the essential pieces of evidence for the diagnosis of Hirayama disease according to expert lead guidelines using a modified Delphi technique.3 MRI cervical spine (during neck flexion) revealed anterior displacement of the posterior theca such that it compresses the cord between it and the spinal column anteriorly from C4-C6 with the prominence of epidural venous dural space dorsal to dura with mild cord thinning from C4-C6. The posterior dural sac crescent appeared in T2/STIR as a hypointense entity and enhanced uniformly postcontrast.

A study by Jasem et al. proposed a rare form of mechanically induced chronic ischemic focal cervical myelopathy, in which local anterior compression of the dura and spinal cord against the back of the vertebral bodies occurs. It is caused by repeated neck flexion resulting in ischemia of anterior horn cells because of compression of the anterior spinal artery. The difference in balance between the growth of the vertebrae and the dura mater leads to a "tight dural canal" and "overstretched cord," which is unable to compensate during neck flexion for the increased length of the posterior wall. Negative pressure is also generated in the posterior venous plexus of the marrow.2 The affected part of the spinal cord is flattened anteroposteriorly, the anterior horn is largely gliotic and atrophied, and both large and small motor neurons are reduced in number, as shown in studies.4

Differential diagnosis of “Hirayama disease” includes—ALS (most common), spinal muscle atrophy, multifocal motor neuropathy with conduction block, syringomyelia, compressive myelopathy, C8-T1 radiculopathy, cervical spondylotic myelopathy, multifocal motor neuropathy, toxic neuropathy, and postpolio syndrome.2,5

The mainstay of treatment includes prevention of flexion of the neck by using a cervical collar (for a period of about 3–4 years) that may arrest the progression of the disease. Muscle-strengthening exercises and assistive devices like splinting and braces can also be used along with a cervical collar.2,3

**CONCLUSION**

“Hirayama disease” is not a commonly encountered condition in clinical practice. A
high index of suspicion should be kept if any young male patient presents with lower motor weakness of hands and forearms, unilateral or bilateral (asymmetrical). Flexion MRI, NCV, and EMG should be performed to establish the diagnosis at the earliest. Early diagnosis and timely administration of cervical collars can prevent further atrophy and may be helpful in halting the disease process.

REFERENCES