Hirayama Disease: Diagnosis to be Missed Without Flexion MRI

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Case Report

ABSTRACT

Hirayama disease, also known as monomelicamyotrophy (MMA), is a rare cervical myelopathy that manifests itself as a self-limited, asymmetrical, slowly progressive atrophic weakness of the forearms and hands predominantly in young males. The forward displacement of the posterior dura of the lower cervical dural canal during neck flexion has been postulated to lead to lower cervical cord atrophy with asymmetric flattening. We report a case of Hirayama disease in a 18-year-old male presenting with gradually progressive asymmetrical weakness and wasting of right hand and forearms.

KEY WORDS: hirayama disease (HD), monomelicamyotrophy (MMA), motor neuron disease (MND)

INTRODUCTION:

Hirayama disease (HD), is a sporadic juvenile muscular atrophy of the distal upper extremities, which predominantly affects the lower cervical cord. It mainly develops in the late teens and early 20's with a male preponderance. The typical clinical features include insidious onset and slow progression of unilateral or bilateral muscular atrophy with weakness of the forearms and hands. Sensory disturbance, autonomic involvement, and upper motor neuron signs like hyperreflexia are rare[1]. The motor neuron disease (MND) is a very close differential diagnosis of HD, but, unlike MND, the disease progresses initially and is followed by spontaneous arrest several years after the onset.

CASE REPORT:

A 18-year-old male presented with a 4 years history of slowly progressive weakness and thenar muscle atrophy that started in the right hand and forearm. The hand weakness limited several activities of his daily living. There was no history of neck pain, sensory involvement, difficulty in walking, bowel or bladder involvement. His past medical history was insignificant; there was no history of trauma, toxins exposure or allergies. None of his family members had a similar complaint.

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Blood investigations were within normal range. Cervical spine MRI was performed on 1.5-tesla siemensmagnetom MR scanner. T1 & T2 weighted spin echo sequences along with diffusion weighted sequence were taken. The MRI in neutral position reveal minimal cord thinning in the region of C4-C5(predominantly involving right half)without obvious cord compression. MRI in flexion position (maximum flexion by patient upto the extent till he don't experience any discomfort), showing loss of dural attachment and anterior dural shift of approximately 4-5mm(extending from C3 to C7 level) with prominent posterior epidural space. The cord is displaced anteriorly. MRI imaging features were consistent with HD.

DISCUSSION:

This disease was initially recognized in Japan in 1959 by Hirayama et al and was reported under the name of "juvenile muscular atrophy of unilateral upper extremity"[2]HD is characterized by insidious onset asymmetrical weakness and wasting of muscles of upper limb with male predominance between 15 & 25 years of age. The disease usually progresses for few years and then is followed by arrest of progression. The clinical features may include irregular coarse tremors in the fingers of the affected hands. The sensory, reflexs, and cranial nerve examinations are generally normal.

Tashiro et al. [3]outlined the criteria for diagnosis of HD: (1) Distal predominant muscle weakness and atrophy in forearm and hand; (2) Involvement of the unilateral upper extremity almost always all the time; (3) Onset between the ages of 10 to early 20s; (4) Insidious onset with gradual progression

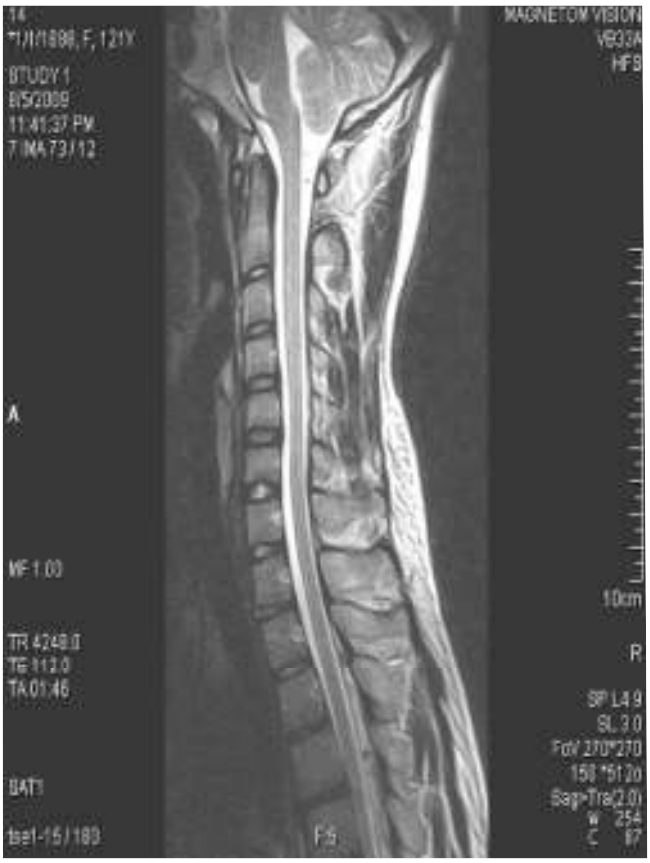

Figure 1: T2 saggital MRI cervical spine in neutral position shows cord thinning at C4-C5 level.

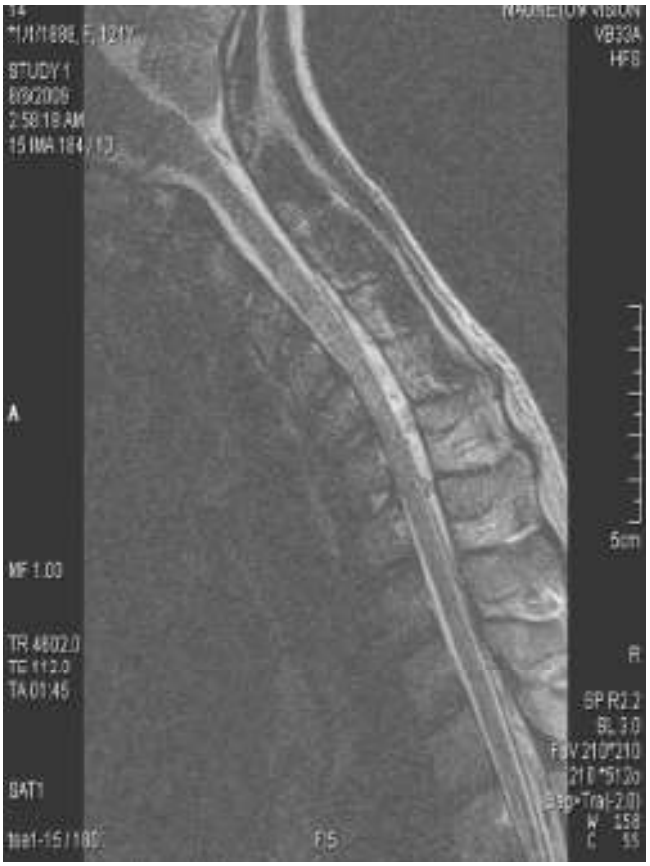

Figure 2: T2 saggital MRI cervical spine in flexion shows anterior dural shift and prominent posterior epidural space.

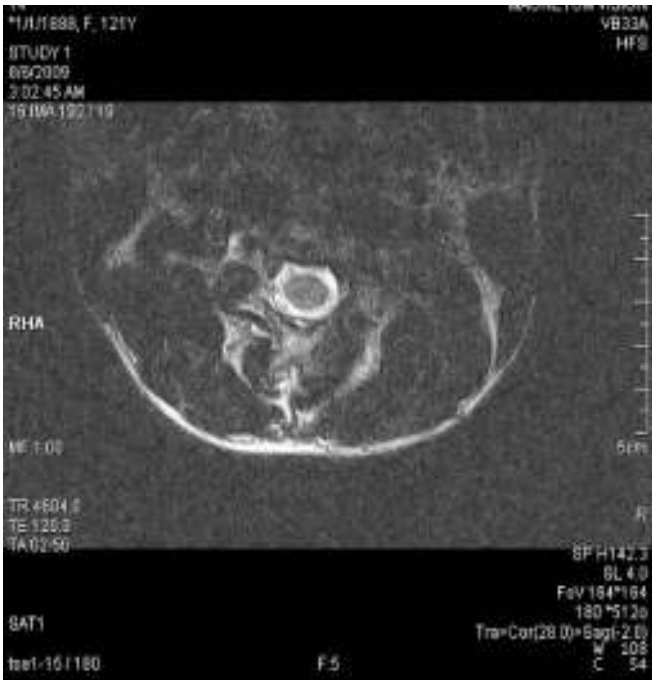

Figure 3: T2 axial MRI shows cord thinning in neutral position.

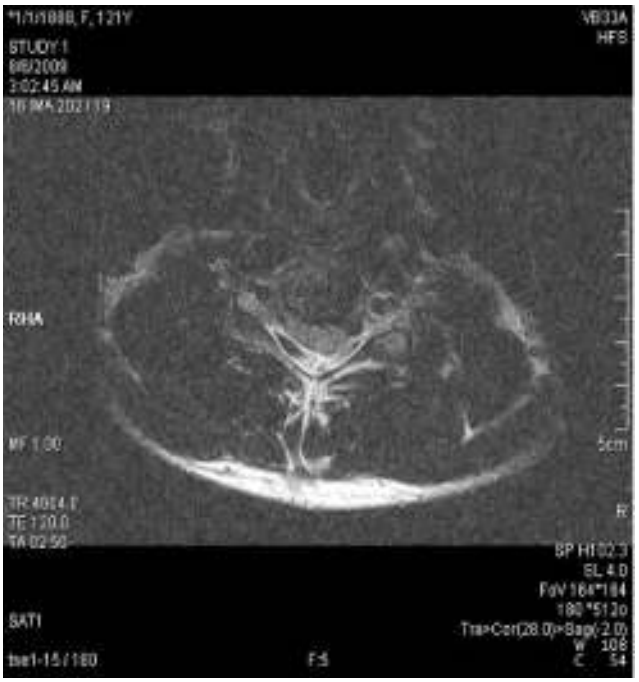

Figure 4: T2 axial MRI shows anterior dural shift in flexion position.

for years, followed by stabilization; (5) No lower extremity involvement; (6) No sensory disturbance and tendon reflex abnormalities; (7) Exclusion of other diseases (e.g., motor neuron disease, multifocal motor neuropathy, brachialplexopathy, syringom-yelia).
The present patient met most of the criteria laid down by Tashiro et al.

The exact pathogenesis of HD is still unknown. A pathological study by Hirayama et al[4] demonstrated cell shrinkage and necrosis, mild gliosis, and some circulatory insufficiency in the anterior horns of the spinal cord from the lower cervical to upper thoracic levels, particularly at the C7 and C8 levels. The most widely accepted hypothesis is a cervical myelopathy associated with neck flexion, proposed by Kikuchi et al[5]. Normally, the spinal dura mater is loosely attached to the vertebral canal by the nerve roots and the periosteum at the foramen magnum and the dorsal surfaces of C2 and C3 and the other at the coccyx. The relatively short and tight dura mater seen in patients with HD is unable to compensate for the increased length of the vertebral canal during neck flexion. This results in tightening of the dural canal during neck flexion, which leads to an anterior shift of the posterior dural wall, causing spinal cord compression against the vertebral body. This repeated neck flexion results in multiple episodes of ischaemia and chronic trauma to the spinal cord, which eventually leads to myelopathy, as evidenced by asymmetric lower cervical cord thinning in the MRI.

The differential diagnosis of HD includes the distal form of spinal muscular atrophy, amyotrophic lateral sclerosis (ALS), multifocal motor neuropathy with conduction block, and toxic neuropathy as well as structural lesions of the cervical cord. The key to diagnose this disease is based on the typical clinical features and dynamic MRI study when the neck is flexed. MR studies in flexion show not only the anterior displacement of the posterior wall but also a well-enhanced crescent-shaped lesion in the posterior epidural space of the lower cervical canal. This lesion typically disappears when the neck returns to a neutral position, confirming it to be a congested posterior internal vertebral venous plexus rather than a vascular malformation.

MR imaging studies of the cervical spine in a neutral position can reveal several features such as localised lower cervical cord atrophy, asymmetrical cord flattening, and loss of attachment between the posterior dural sac and subjacent lamina, as well as noncompressed intramedullary high T2 signal intensity.

CONCLUSION:

Hirayama disease is a self-limiting disorder and there is no consensus on the definitive treatment. However, early diagnosis is necessary because a cervical collar and physiotherapy may arrest the progression of the disorder by limiting the neck flexion.

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