



Case Report:

Hirayama Disease - A Variant of Motor Neuron Disease and Role of Flexion MRI in Diagnosis

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Abstract: Hirayama disease is a monomyelic variant of motor neuron disease (MND) and has distinctive features of male predominance, asymmetric involvement of upper extremities with a self limiting course. Flexion MRI (magnetic resonance imaging) forms the main stay for diagnosis of this condition. Here we report such an unusual case of Hirayama disease in a male patient of 20 years who presented with weakness and atrophy in right upper limb. Careful clinical examination will help to use the flexion MRI studies for the diagnosis of this condition as done in our case.

Key Words: Hirayama disease; Motor neuron disease; Flexion MRI; Monomyelic

Introduction:

Hirayama disease, also known as juvenile non-progressive cervical amyotrophy or monomelic amyotrophy is a rare focal motor neuron disease that primarily affects young Asian males (15-25 years old). The first case was reported by Hirayama in 1959. The mechanism for this clinical condition is mainly an imbalance in growth of the vertebrae and the dura mater. Flexion MRI (magnetic resonance imaging) forms the main stay for diagnosis of this condition. As the disease is non progressive after a certain period, early diagnosis will be of benefit to the patient.

Case Report:

A 20 years old male patient was admitted with complaints of weakness right hand from 4 years which was insidious onset and progressive type, mainly restricting his fine movements of fingers later gradually involving his movements at wrist. There was no history of difficulty in lifting his right upper

limb above head or movements at elbow. There was no history of abnormal sensations or sensations in the involved limb. But patient had complaints of slight thinning of hand compared to left side. There were no complaints in rest of the limbs. The disease was non progressive from last three months. On detailed neurological examination, there was atrophy of forearm and hand muscles of right side (Fig. 1), had power of grade 3 at all joints of hand and wrist with flexors weaker than extensors. All reflexes were 2+ and the sensory system was normal. Intermittent fasciculations were appreciated in right forearm muscles. Rest of the nervous system examination was normal.

On investigations, biochemical and haematological parameters were normal. Patient was planned for imaging studies keeping possibility of monomyelic variant of motor neuron disease. On magnetic resonance imaging of cervical spine in non-flexed position of neck, there were hyperintensities in cervical spinal cord extending to upper thoracic cord (Fig. 2). On flexion imaging (neck flexion) there was appearance of longitudinal mass behind dura which was due to anterior shifting of posterior dura at the lower cervical spinal canal causing congestion of venous plexus (Fig. 3). These features were strongly suggestive of Hirayama disease. Electrophysiologic studies showed decreased amplitude in median nerves with normal latencies and low velocities; motor unit action potentials were seen in the muscles innervated by C7, C8 and T1 nerve roots, with fibrillations and positive waves. Patient was advised to use soft cervical collar. On follow up, there was no progression

of the disease. He was not advised surgery as the disease was in a non progressive stage.



Fig. 1: Right upper limb showing atrophy at wrist and small muscles of hand.



Fig. 2: Flexion MRI showing congested venous plexus appearing as a mass (due to anterior shift of posterior dura).



Fig. 3: Non flexion MRI of spine showing hyperintensities in cervical and upper thoracic spinal cord with no congestion of venous plexus.

Discussion:

Hirayama disease is a monomyelic variant of motor neuron disease and has distinctive features of male predominance, asymmetric involvement of upper extremities with a self limiting course.¹ The atrophy and weakness predominantly involves the intrinsic hand muscles (interossei, thenar, and hypothenar muscle groups) as well as the ulnar side of the forearm.^{2,3} Hirayama disease is a benign disorder with a stationary stage after a progressive course. Hirayama disease was initially recognized in Japan in 1959 and reported under the name of juvenile muscular atrophy of unilateral upper extremity.³ In the series of Kikuchi et al.³ there were 17 males and 1 female and the progression of symptoms arrested within 5 years.

The explanation for this disease mechanism is based on anatomical orientation. The spinal dura mater is a loose sheath that is anchored in the vertebral canal by the nerve roots and by attachment to the periosteum in two places: one at the foramen magnum and the dorsal surfaces of C-2 and C-3, and the other at the coccyx. The remainder of the dura mater is only suspended and cushioned in the spinal canal by the epidural fat, venous plexus, and loose connective tissues.⁴ In neck extension, the dura mater of the cervical spine is slack and thrown into transverse folds. In neck flexion, the dura becomes tighter, because the length of the cervical canal increases as the neck moves from extension to flexion. The difference in length between extension and flexion from T-1 to the top of the atlas is 1.5 cm at the anterior wall and 5 cm at the posterior wall.⁵ Normally, the slack of the dura can compensate for the increased length in flexion. In Hirayama disease, the dural canal is no longer slack in extension, because of an imbalance in growth of the vertebrae and the dura mater. Therefore, a tight dural canal is formed, which cannot compensate for the increased length of the posterior wall during flexion. This causes an anterior shifting of the posterior dural wall, with consequent compression of the cord. This compression may cause microcirculatory disturbances in the territory of the anterior spinal artery or in the anterior portion of the spinal cord. The

chronic circulatory disturbance resulting from repeated or sustained flexion of the neck may produce necrosis of the anterior horns, which are most vulnerable to ischemia.⁶

In patients with Hirayama disease, conventional radiographic studies of the cervical spine usually show no specific abnormalities. MR studies in neck flexion, can show not only the anterior displacement of the posterior wall but also a well-enhanced crescent-shaped mass in the posterior epidural space of the lower cervical canal.⁷ This mass is thought to represent congestion of the posterior internal vertebral venous plexus rather than vascular malformations or tumors, because it vanishes once the neck returns to a neutral position.⁸

Some pathophysiological factors are responsible for venous engorgement. First, the negative pressure in the posterior spinal canal resulting from anterior shifting of the dural canal increases the flow to the posterior internal vertebral venous plexus.⁷ Second, the compressed anterior internal vertebral venous plexus caused by anterior displacement of the dural canal increases the burden of the posterior internal vertebral venous plexus. And, third, the posture of neck flexion decreases the venous drainage of the jugular veins, impeding the venous return of internal vertebral venous plexus.⁷ In combination, these factors cause the formation of an engorged posterior internal vertebral venous plexus, which, in imaging, becomes a striking and specific characteristic of this disease.

So, in our case it was 20 year male patient who was diagnosed on clinical basis and confirmed by flexion magnetic resonance imaging studies.

Learning point: Careful detailed clinical history and examination helps in the diagnosis of this unusual condition and flexion MRI studies can be used accordingly for confirmation.

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