Classical Hirayama Disease Presenting as Progressive Spastic Quadriparesis

Dear Sir,

Hirayama disease (HD) is a rare cervical flexion-induced myelopathy predominantly seen in young Asian males. Typically, it is a pure lower motor neuron (LMN) disorder with unilateral or bilateral distal upper limb wasting and weakness with cold paresis and commonly subclinical pyramidal involvement.[1] As used by most studies, HD criteria include lack of sensory and tendon reflex abnormality and exclusion of other diseases.[2] Prominent pyramidal, sensory, and autonomic involvement are seldom observed.[3] Characteristic flexion-induced dynamic changes are seen on MRI. Earlier reports have documented HD progressing to spastic paraparesis.[4] Here, we document a 23-year-old man with distal bilateral HD associated with classical HD and progressive spastic quadriparesis as a manifestation of severe HD.

A man aged 23 years had progressive asymmetrical distal upper limb weakness and wasting for 1 year. He also had transient paresthesias and radiating lancinating pains in upper limbs on neck flexion. After 10 months, he developed spasticity of lower limbs. On examination, he had minipolymyoclonus, wasting of shoulder girdles, arms, and forearm muscles with preserved brachioradialis [Figure 1]. There was mild and moderate spasticity of upper and lower limbs, respectively. Muscle weakness was moderate in upper limbs and mild in lower limbs. Tendon reflexes were hyperactive with patellar and ankle clonus. There was spastic gait.

The cervical spine MRI showed reversal of cervical lordosis, marked cord atrophy from C4 to C7), and snake eye appearance [Figure 2a and b]. On flexion, prominent ventral displacement of the posterior dura and epidural venous plexus engorgement was observed from C3 to C7 levels in postcontrast T1W sequence [Figure 2c and d].

Classically, HD presents predominantly as slowly progressive unilateral or asymmetrical distal upper limb weakness and wasting in adolescent males. Although the etiopathogenesis remains unknown, it is proven that HD arises due to (i) compression of anterior spinal cord and anterior epidural venous plexus against posterior surface of vertebral bodies on neck flexion with resultant engorgement of posterior venous plexus and vascular insufficiency to anterior horn cells and[5] (ii) rapid lengthening of spinal column compared to spinal cord and dural sac during growth spurt in adolescence resulting in tightening and stretching of dura, and further getting displaced anteriorly on neck flexion.[6]

Clinical features were suggestive of pure motor involvement with both LMN and upper motor neuron (UMN) features and UMN pattern of weakness in lower limbs. In recent clinician-led guidelines (modified Delphi method), 61% of neurologists reported that pyramidal tract signs do not exist in patients with HD. Only 19% agree that pyramidal tract signs might occur in some HD patients.[7] Differential diagnosis of young onset amyotrophic lateral sclerosis (ALS), syringomyelia, and intrinsic cord lesions can be considered in HD with pyramidal involvement. Syringomyelia may not always present with dissociative sensory loss. Other diseases such as syringomyelia, spinal cord tumors, ossification of posterior longitudinal ligament, cervical spondylosis, other cervical vertebral abnormalities, motor neuron disease, and motor neuropathy were excluded. Lhermitte phenomenon or barber chair phenomenon is an electric shock-like sensation that occurs on flexion of the neck. This sensation radiates down the spine, often into the legs, arms, and sometimes to the trunk. It is not a feature described in HD but previously described in only one case report.[8] The presence of this phenomenon suggests involvement of posterior column of spinal cord apart from anterior horn cells and corticospinal tracts. Thus, this phenomenon indicates that HD is indeed a flexion-induced compressive myelopathy, denoting the significance of surgical intervention in HD.

MR imaging findings include localized lower cervical cord atrophy, asymmetric cord flattening, and loss of the attachment of the posterior dura. The loss of the attachment of the posterior dura in the neutral position imaging is reported with a sensitivity and specificity of 93% and 98%, respectively. The combination of neutral and flexion imaging features had a sensitivity and specificity of 71% and 100% for the diagnosis of HD, respectively.[9]

Electroneuromyography revealed neurogenic lesion only in cervical segments. Cervical spine flexion MRI contrast study is a critical investigation to confirm the diagnosis. It signifies the dynamic apposition of spinal cord against the vertebra by anterior displacement of posterior dura and enhanced posterior epidural venous plexus which disappears once the neck returns to a neutral position.

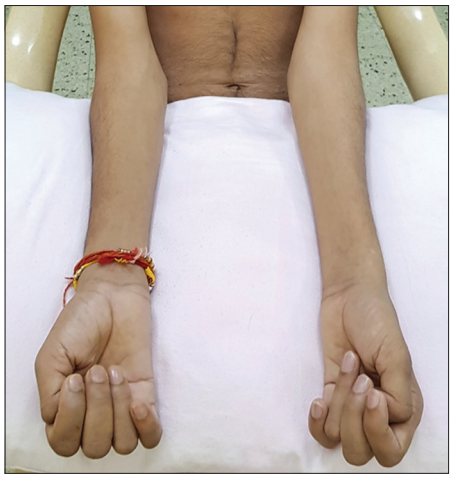

Figure 1: Note the severe wasting and weakness of the forearm and hand muscles. Relative sparing of the Brachioradialis is present

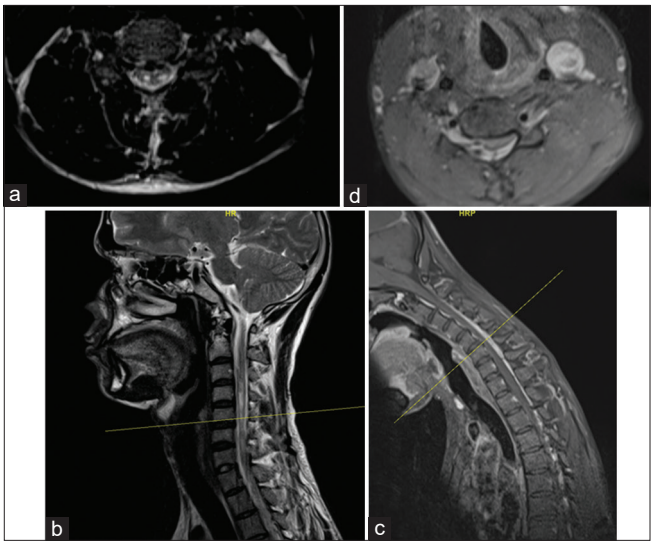

Figure 2: (a) MRI cervical spine (T2WI axial): snake eye appearance at C5 level. (b) (T2WI sagittal): loss of cervical lordosis and cervical cord atrophy and intramedullary T2 hyperintensity predominantly involving the anterior horn cells at C4–7 levels; and (c) postcontrast flexion T1W sagittal image of the cervical spine (sagittal) and (d) (axial) shows ventral displacement of the posterior dura with cord compression and enhancement of posterior epidural venous plexus from C3 to C7 levels

Autonomic dysfunction was also present, indicating an extended form of HD. Studies by Hassan *et al*.[10] have showed autonomic dysfunction in 36% and 46% of HD cases, respectively. Similarly, UMN features were seen in 18% and 12% of the cases reported by Hassan *et al*. and Sonwalkar *et al*.,[11] respectively.

Advanced HD can be misinterpreted as ALS and differentiating them is of utmost importance. The absence of LMN features in lower limbs and typical dynamic MRI cervical spine picture confirms the diagnosis of HD in this case. Since HD is secondary to cervical flexion-induced cord injury, neck immobilization with strengthening exercise for weak upper limb muscles forms the first line of treatment. However, early surgical intervention such as anterior cervical decompression and fusion, posterior instrumented fusion, and laminectomy should be considered, which will benefit patients with progressive weakness to prevent permanent neurological sequalae.[12]

HD may mimic ALS and should be kept in the differential diagnosis when a young male presents with spastic quadriparesis, as surgical intervention can potentially halt the progression and lead to significant improvement. HD may show autonomic dysfunction, unlikely to be observed in ALS. Dynamic contrast MRI cervical spine study is crucial to confirm HD as findings may be missed in the neutral position.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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