



A case of HIRAYAMA disease

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KEYWORDS :

1. Introduction : Hirayama disease, also termed non-progressive juvenile spinal muscular atrophy of the distal upper limbs, is a type of cervical myelopathy related to flexion movements of the neck. It is considered a benign motor neuron disorder with a stationary stage after a progressive course.

2. Case Report : Mr MAYURPURI GOSWAMI , 20 YEAR old male patient was admitted on 6/9/16 in ER of VSGH , AHMEDABAD. Patient presented with chief complain of left upper limb weakness since 1 year and thinning of left upper limb since 6 months. Patient was relatively asymptomatic before 1 year then he developed insidious onset , gradually progressive distal weakness in the form of difficulty in breaking chapatis , buttoning-unbuttoning and taking heavy objects from the over shelf. This is associated with thinning of left upper limb and twitching under the skin of left upper limb. There is no h/o weakness in any other limbs .No h/o headache , loss of consciousness , seizure, abnormal behavior. No h/o diplopia , blurring of vision, decreased sensation over face , facial asymmetry, dysphagia , nasal regurgitation of food. No h/o sensory symptoms. No h/o bowel-bladder incontinence/retention .No h/o neck pain /back pain. No h/o joint pain , rash , oral-genital ulcers , Reynaud's phenomenon, dry eyes , dry mouth. No h/o flexor spasm or tightness of limbs at night.

3. CNSEXAMINATION :

Higher function – normal

Cranial nerves – normal

Pupil – B/L equally reacting to light

Power- right U/L , Bilateral L/L – all three limbs power is 5/5 but in left U/L power is proximally 4/5 and distally 3/5.

Tone increased in all limbs except in the right U/L which is of normal tone.

DTR- ++ In right U/L, Bilateral L/L but in Left U/L DTR IS ABSENT.

Planters – B/L Extensor.

Sensory – normal

Bowel-bladder – normal

Cerebellum – normal

4. Lab values :

Hb 13.1 , tc 5310 , plt 2.42 lack

RFT with electrolytes and LFT with enzymes within normal limits

TSH – 1.02 microIU/ml [NORMAL]

B12 – 365 pg/ml [NORMAL]

CRP – Negative

CSF R/M – Normal

SPTH – Normal

EMG NCV- neurogenic changes in left U/L



5. TREATMENT : Patient was treated with muscle relaxant like baclofen and other agent like riluzole.

6. DISCUSSION : Hirayama disease is characterised by focal amyotrophy with unilateral or asymmetric bilateral weakness and

wasting of muscles innervated by C7, C8, and T1. It's an insidious onset, chronic, often self-limiting disorder with male preponderance, seen between the ages of 15 and 25 years. Hirayama et. al first reported this disease in the year 1959, but pathologic study was not done till 1982, because of its benign course. At pathologic examination, these authors found the lesions only in the anterior horns of the spinal cord from C-5 to T-1, particularly marked at C-7 and C-8. Most commonly seen in Japan and other Asian countries like India and Malaysia. The pathogenesis of the disorder is unknown – probable causes suggest that an imbalanced growth between the patient's vertebral column and spinal canal contents. This imbalanced growth will cause disproportional length between the patient's vertebral column and the spinal canal contents, which will cause a "tight dural sac" or "overstretch of the cord" in the neutral position and an anteriorly displaced posterior dural wall when the neck is flexed. Current neuroradiologic techniques are able to show forward displacement of the posterior wall of the lower cervical dural canal in neck flexion, which is presumed to be a primary pathogenetic mechanism of Hirayama disease. 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. Normally, the slack of the dura can compensate for the increased length in flexion; therefore, the dura can still be in close contact with the walls of the spinal canal without anterior displacement. 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. In patients with Hirayama disease, conventional X ray of the cervical spine usually show no specific abnormalities except straight alignment or scoliosis. Myelography may show the forward movement of the posterior dural wall when the neck is flexed; however, myelography is difficult to perform, as it is difficult to retain the contrast medium in the cervical subarachnoid space when the neck is flexed, regardless of the patient's position. MRI studies in neck flexion, which are easy to obtain, will show the forward displacement of the posterior wall and a well-enhanced crescent-shaped mass in the posterior epidural space of the lower cervical canal. 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. MRI shows atrophy of the lower cervical cord in a neutral position and there will be abnormal cervical curvature (straight or kyphotic) and loss of attachment between the posterior dural sac and subjacent lamina, which is a most valuable in Hirayama disease.

6. Conclusion : Even though Hirayama disease is a rare self-limiting disease, early diagnosis is necessary. Use of a simple cervical collar to prevent neck flexion, has been shown to halt the progression of the disease. Diagnosis of Hirayama disease is mainly based on flexion MRI of cervical spine. Asymmetry is one of the most characteristic findings of this disease, both clinically and radiologically. Thus, in cases of adolescent onset slowly progressive distal upper limb weakness followed by stabilization, with neurogenic changes in the EMG and the findings of asymmetric cord atrophy on regular nonflexion cervical spine MRI studies, especially at the lower cervical

cord, one should keep in mind the diagnosis of Hirayama disease. When this sign is seen, a flexion MRI study should be performed to confirm the diagnosis.

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