Hiramaya Disease - A Case Report

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PRESENTATION OF CASE

A 20-year-old male was evaluated by us five months post development of weakness in right upper arm followed by intrinsic muscle atrophy in the right hand when his age was 18-years. Weakness was observed first during performing daily activities like mixing food in the plate, holding utensils, lifting books / schoolbag, opening bottles, and holding small objects. The weakness got worse over several months and got stabilized after around two years. Occasional cramps could be noted during the progression of the disease. Pain was pricking, sharp (not referred to other limb) associated with numbness and occasional burning along with dysesthesia in the right upper arm. His past medical history suggests wasting in the left lower limb below knee, noted when the patient was of the age of five years. He had to walk with the support of the stick. There is no history of trauma. There was no family history of any neuromuscular disease.

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Sr. No.	Muscles	Power Right Side	Power Left Side	Sr. No.	Muscles	Power Right Side	Power Left Side
1	Biceps	5/5	5/5	8	Abductor Pollicis Brevis	3/5	4/5
2	Triceps	4/5	5/5	9	First Dorsal Interossei	1/5	4/5
3	Deltoid	5/5	5/5	10	Flexor Pollicis	0/5	5/5
4	Brachioradialis	5/5	5/5	11	Lumbricals	2/5	5/5
5	Wrist Flexors	4/5	5/5				
6	Wrist Extensors	4/5	5/5	12	Finger Extensors	3/5	5/5
7	Digital Muscles	3/5	5/5	13	Thumb Extensors	2/5	5/5

Table 1. Clinical Assessment of Power in Various Muscles

On physical examination, vital signs were normal. On neurologic examination, there was significant atrophy of the right hand intrinsic muscles. Also, there was atrophy, to a lesser degree, in the right forearm flexor muscles (Fig. 1). Fasciculation's were absent. Tendon flexor reflex was found to be reduced (1/4) in the right distal upper limb whereas other reflexes were present throughout symmetrically. Fine resting Tremors were observed in both forearms.

Blood investigations like complete blood count (CBC), vitamin B12 and vitamin D3 level, erythrocyte sedimentation rate (ESR), Kidney, liver and thyroid function tests, were within normal range. The results for rheumatoid factor, antinuclear antibody, and anti-phospholipid antibody, human immunodeficiency virus (HIV), hepatitis B and hepatitis C were negative.

Routine examination and F-wave latencies of both the affected limb and the unaffected limbs were normal. Needle examination showed increased insertion activity with fewer fibrillation potentials in right C7, C8-T1 muscles. Longer duration MUAPs with severely reduced recruitment pattern (MUAP dropout) was seen in right 1st dorsal interosseous (FDI), extensor digitorum communis (EDC), triceps, and flexor carpi radialis muscles. Normal MUAPs was seen in right deltoid, biceps brachii muscles. However left extensor digitorum communis (EDC) and flexor carpi radialis also showed slightly increased amplitude duration and reduced recruitment of MUAPs. Sampled paraspinals showed normal insertion activity and no spontaneous activity was seen.

MRI scan of cervical and thoracic region suggests subtle altered signal intensity area, appearing hyperintense on T2 weighted images. Flattening of the cord more on the right side is seen from lower border of C4 up to lower border of C7. There is significant shift of posterior dura anteriorly seen extending from lower border of C3 up to lower border of D3 in flexion imaging.

DIFFERENTIAL DIAGNOSIS

Various differential diagnoses like, MND (Motor Neuron Disease), C8-T1 radiculopathy, ulnar neuropathy, multifocal motor neuropathy, toxic neuropathy, syringomyelia, Hansen's disease, amyotrophic lateral sclerosis, spinal cord tumours, degenerative motor neuron disease, traumatic cervical spinal cord injury, post-polio syndrome.⁽¹⁾ The motor neuron disease (MND) can be a very close differential diagnosis but Hirayama Disease typically progresses initially followed by spontaneous arrest several years after its onset unlike MND.

Figure 1. Atrophy of Right Thenar and Hypothenar Eminences Compared to Left Hand

Figure 2. Comparison of Right and Left Forearm

Figure 3. MRI of Neck in Flexion

Figure 4. MRI of Neck in Neutral Position

CLINICAL DIAGNOSIS

In the present case the clinical findings of insidious onset, juvenile male preponderance, the unilateral muscular weakness along with atrophy in the unilateral hand and forearm (Fig. 1, 2) which progresses for about more than 1 year to 3 years and then stabilizes over a sufficient period,^(2,3,4,5,6,7) male preponderance and the age of onset being 18 yrs., rare familial and usually sporadic in occurrence,^(4,5) associated atrophy and weakness of the intrinsic muscle of hand as well as the flexors of forearm along with relative sparing of the brachioradialis muscle (oblique atrophy),^(2,8) distal irregular jerky tremor in the upper limb, non-involvement of the muscles innervated by cranial nerves,^(6,7) no sensory disturbance and tendon reflex abnormalities, etc are all consistent with the Hirayama disease.

Nerve conduction study in this case show positive sharp waves, fibrillations,^(3,6,7) and also the presence of large MUPs in upper or lower limbs suggesting denervation and re-innervation,^(3,5,9) suggesting proximal lesion at spinal level affecting predominantly right C7, C8 and T1 roots/ anterior horn cell with chronic re-innervation in muscles which are consistent with the Hirayama disease.

MR imaging studies of the cervical as well as thoracic spine in a neutral position (Fig. 4) reveals subtle altered signal intensity area, non-compressed intra-medullary high T2 signal intensity, localized atrophy of lower cervical cord, flattening of the cord more on the right side extending from lower border of C4 up to lower border of C7. On the other hand, MR imaging studies of the cervical and thoracic spine in flexion imaging (Fig. 3) reveals significant shift of posterior dura anteriorly

seen extending from lower border of C3 up to lower border of D3. All these MRI findings are consistent with the Hiramaya disease.⁽¹⁰⁾ Results of routine blood tests and CSF analysis are usually normal.⁽³⁾

Pathophysiology

The pathogenesis of this disease is not known. The widely accepted hypothesis on pathophysiology of Hiramaya Disease remains neck flexion associated cervical myelopathy affecting the anterior horn cells localized in lower cervical cord.⁽¹¹⁾ As the dura is, normally, most strongly attached at the foramen magnum, C2, C3 levels and at the coccyx, it increase in the length during the growth of the spinal canal and also it can slacks itself normally during cervical flexion. But during the flexion of neck, relatively short dura get stretched and tightened with the increased length of the vertebral canal in patients with Hiramaya disease. An anterior shifting of the posterior dural wall occurs thereby, compressing the spinal cord against the vertebral body.⁽¹²⁾ This frequent episodes of trauma and ischemia to the spinal cord results in cord atrophy and myelopathy, especially the anterior horn cells, at the level of most kyphosis. Persons who lack normal cervical lordosis (straight necks) are already having stretched nerve roots and thus are more prone. The incidence also found to be higher in individuals with slow initial growth and then sudden spurt of growth later. MRI scan, can reveal asymmetric lower cervical cord thinning.⁽¹³⁾ Presence of anterior displacement of the posterior wall along with a prominent crescent-shaped lesion in the posterior epidural space of the lower cervical canal in flexion, but disappearance of the same when the neck returns to a neutral position suggests ischemic aetiology due to congested posterior internal vertebral venous plexus.⁽¹⁰⁾

Also muscles of the distal areas are innervated by the neurons with long axons and require a more blood supply. Thus muscular atrophy results in especially in the hand and the ulnar side of the forearm in C5-T2 myotomes due to the chronic functional damage to these neurons for duration of two years or more.⁽¹⁴⁾

MANAGEMENT

Hirayama disease as it is a self-limiting disorder with no definitive line of treatment. But if diagnosed early, we can arrest the progression of the disease. Unless the skeletal growth is complete, one should avoid forward flexion of the neck. Physical exercises involving frequent neck flexion should be avoided to prevent progression of disease. This can also be prevented by regular use of a cervical collar. Also immobility like joint stiffness and muscle wasting can be prevented by appropriate prescription of Physiotherapy.⁽¹⁵⁾ Surgical interventions like duroplasty, anterior spinal decompression, posterior spinal fusion and even use of the cervical collar can be employed in the patients showing compression of the cervical cord against the adjacent body of vertebra.^(16,17)

FINAL DIAGNOSIS

In conclusion, we present a case of Hirayama disease where the patient is a juvenile male, with a non-progressive,

asymmetric muscular atrophy involving mostly cervical C4 - C7 myotomes leading to weakness and atrophy of intrinsic muscles of right hand and right forearm but the brachioradialis muscle is spared. This rare, benign, focal cervical poliomyelopathy appears to be caused by forward displacement of the posterior cervical dura, highlighted by the dynamic MRI study of cervical spine which is a hallmark diagnostic technique of the above condition. Though it is a self-limiting disease, early diagnosis of Hiramaya disease is important to prevent further flexion injury by the use of hard cervical collar.

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