HIRAYAMA DISEASE:A CASE SERIES HIGHLIGHTING PHENOTYPIC SPECTRUM AND DIAGNOSTIC CHALLENGES

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INTRODUCTION

Hirayama disease is a rare, self-limiting cervical myelopathy predominantly affecting adolescent and young adult males usually progressing for varying period followed by spontaneous arrest within several years. typically presents as insidious-onset, asymmetric distal upper limb weakness with lower motor neuron features. However, the clinical spectrum can vary, posing diagnostic challenges

OBJECTIVE

We report two cases of young males aged 33 and 20 years who presented with progressive upper limb weakness. Both underwent Clinical, Electrophysiological and Dynamic Cervical MRI Evaluvation.

RESULTS

A 33 year old male apparently asymptomatic 6 years ago, developed pain in the right little finger radiating to the wrist, followed by numbness and reduced temperature perception which gradually progressed to involve the right arm. Within 6 months, he developed difficulty holding objects and fine motor impairment later associated with difficulty raising the arm and combing hair. After 3 years, wasting of right hand and arm muscles was noted. 4 years back he developed similar symptoms in the left upper limb beginning with pain and numbness in the hand progressing to the entire limb followed by weakness (distal > proximal) and difficulty with daily activities. Six months back wasting of left hand and forearm was observed. Currently, he has flail upper limbs bilaterally, with preserved lower limb and neck strength, and no gait or bulbar involvement.

On Examination,Vitals stable,Neck length -1.5 cm,Height neck ratio of 13.5 cm.Higher mental function and cranial nerve examinations were normal with no features of Horner syndrome.Bilateral upper limb weakness with wasting in C5-T1 Myotome with oblique amyotrophy wasting of thenar more than hypothenar(split hand sign).Lower limb motor examination normal.superficial reflexes were present with Bilateral plantar flexor .Deep tendon reflexes were 2+ throughout.Sensory examination revealed Suspended sensory loss in C4-T1 dermatomes in upperlimb whereas lower limb sensations were intact.He also had hesitancy and urge incontinence.Cerebellum,posterior column and gait were normal.

Routine investigations were normal.Cardiac Evaluvation was normal.Nerve conduction study done bilateral ulnar axonopathy.CT brain done and was normal.MRI Cervical spine with dynamic flexion and extension and contrast were done.Routine investigations were normal.Cardiac Evaluvation was normal.Nerve conduction study done bilateral ulnar axonopathy.CT brain done and was normal.MRI Cervical spine with dynamic flexion and extension and contrast were done.Routine investigations were normal.Cardiac Evaluvation was normal.Nerve conduction study done bilateral ulnar axonopathy.CT brain done and was normal.MRI Cervical spine with dynamic flexion and extension and contrast were done.

FIGURE ;1

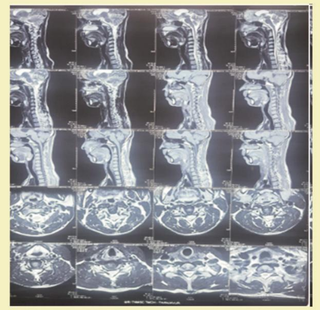

Figure 2-showing Cord atrophy from C4-C7 levels .Anterior-posterior flattening of spinal cord

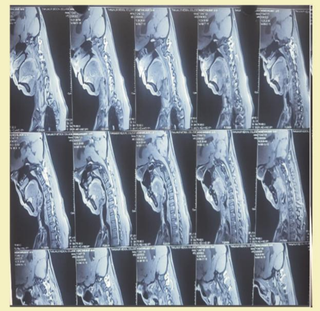

Figure 3-on contrast administration ,Enhancing prominent epidural veneous spaces with flow voids

MRI Cervical spine with Dynamic flexion and extension with contrast revealed T2 cord hyperintensity from C4 to C7 levels.On Flexion ,anterior displacement of dura from C2-C3 level to C7-D1 level .On Contrast administration,enhancing prominent Epidural venous spaces with flow voids features suggestive of Hirayama disease.

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CASE 2

20 year old male presented with insidious onset Painless progressive asymmetric distal onset weakness of left upper limb associated with thinning of left forearm muscles for 2 years.Patient then developed weakness of distal weakness of right upper limb for 6 months associated with wasting over hand and forearm with flexion deformity of medial two fingers.Patient had no Neckpain or weakness of lower limbs or Autonomic disturbances.On Examination,Vitals were stable.Height neck ratio was normal.Higher mental functions and cranial nerves were normal.Motor Examination revealed hypotonia in both upper limbs with distal power of 3/5 and proximal near normal .Asymmetric distal more than proximal weakness with weakness more in left arm than right. Wasting was seen in hand and forearm muscles especially medial compartment with clawing of medial two fingers. Reflexes were diminished in both upper limb,whereas lower limb reflexes were normal .No sensory or Cerebellar signs.

Routine investigations were Normal. Nerve conduction studies Bilateral Ulnar and left median motor axonopathy.EMG in Right and left Deltoid,Right biceps ,Right brachioradialis,Left FDI and Left Extensor indices were suggestive of Neurogenic pattern.MRI Cervical spine with dynamic study done wherein on flexion anterior displacement of Dura from C5 to D1 level with widened posterior epidural space with dilated and tortuous dural venous spaces.

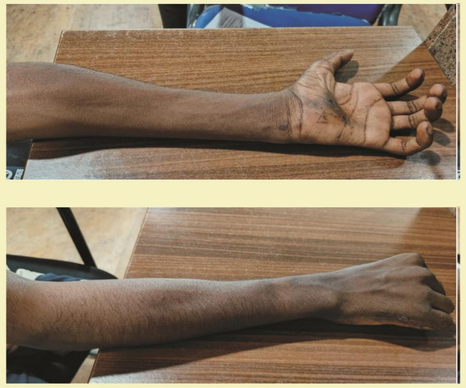

Figure 4A&B-wasting of muscles of forearm and hand with preserved Brachioradialis.

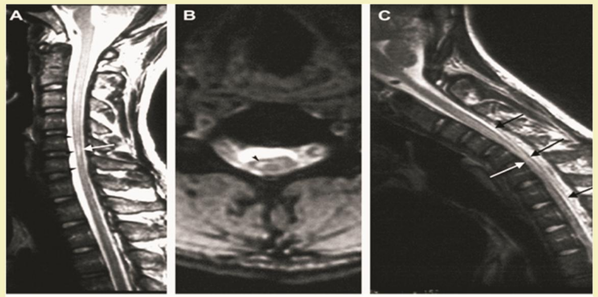

Figure 5 A,B&C-shows cord Atrophy,T2 signal changes,Asymmetric cord flattening and Anterior displacement of dural sac in dynamic flexion study.

DISCUSSION:

The two cases presented highlight the clinical and radiological spectrum of Hirayama disease (HD), a flexion-induced cervical myelopathy predominantly affecting young males. Though classically described as an asymmetric distal upper limb weakness with a self-limiting course, both cases demonstrate atypical and advanced features that expand the traditional phenotype.Pathophysiologically, Hirayama results from a developmental disproportion between the cervical vertebral canal and the dural sac during adolescence. On neck flexion, anterior displacement of the posterior dura compresses the lower cervical cord against the vertebral bodies, producing ischemic injury to the anterior horn cells. The resultant cord flattening and venous congestion are best demonstrated on dynamic flexion MRI, which remains the diagnostic hallmark.

In Case 1, a 33-year-old male showed unusually late presentation with progressive bilateral upper-limb weakness, sensory dissociation, and autonomic symptoms. MRI revealed classical findings of cord atrophy and T2 hyperintensity from C4–C7 along with anterior dural shift and enhancing posterior epidural venous plexus. Such advanced and symmetrical involvement indicates chronic and widespread ischemic injury extending beyond the anterior horns explaining the sensory and autonomic features.

In contrast, Case 2- A 20-year-old male, exhibited the typical presentation of painless, asymmetric distal upper-limb weakness and wasting without sensory or autonomic findings. Electrophysiological studies in both cases demonstrated motor axonopathy in the ulnar and median distributions with neurogenic EMG changes, supporting anterior horn cell involvement.This case demonstrates bilateral asymmetric distal upper limb involvement with flexion deformity which usually deviates from monoamelic amyotrophy pattern with early involvement of other limb.

CONCLUSION

These observations underscore that while Hirayama most often presents as benign Monomelic Bmyotrophy, bilateral and atypical variants do occur sometimes mimicking amyotrophic lateral sclerosis, spinal muscular atrophy or syringomyelia. The distinguishing feature remains the characteristic flexion-induced dynamic changes on MRI which will be absent in those mimics.

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