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Hirayama Disease - A Rare Case

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Abstract: ***21 years old male brought by relatives with complaints of weakness of left upper limb which progressed slowly over a period of 2-3 years. Based on clinical findings and detain neurological examination with other contributing factors like age, sex and progression of a disease, high suspicion of Hirayama's disease was made. On further investigation like MRI cervical spine in flexion and extension it was found that anterior shift of the cervical dural sac from the lamina at the C4-C7 region. On the basis of MRI findings; diagnosis of Hirayama's disease was confirmed and patient was started on physiotherapy and neck extension exercises. Patient was asked for regular follow up in OPD and there was improvement in power of left upper limb on further follow up. Thus following case study presents a rare case of Hirayama's disease.***

Keywords: Hirayama Disease

1. Introduction

Hirayama disease is a juvenile muscular atrophy [1] of distal upper extremity also known as oblique atrophy. It is a type of cervical myelopathy which is characterised by progressive distal upper limb muscle atrophy followed by spontaneous arrest after few years of onset.

The cause of cervical myelopathy is forward displacement of posterior cervical dural sac which is more prominent during neck flexion. Long term compression of cervical cord results in micro-ischemia and eventual atrophy of anterior horn cells. There is reduction in anterior-posterior diameter of cervical cord by greater than 50% along with central necrosis and diminished large and small nerve cells suggestive of chronic ischemic changes without the macrophage infiltration. There is no obvious vascular change to cause ischemic changes in cervical spinal cord.

2. Case Presentation

21 year old male presented with a complaint of weakness of left upper limb since 3 to 4 years. Patient was apparently asymptomatic 3 to 4 years back when he first noticed weakness in the left upper limb, which was insidious in onset and gradually progressive. Initially patient noticed weakness of fingers characterised by difficulty in holding spoon or glass of water; at first patient was able to lift but was not able to hold things for long time and objects were used to fall from his hand later on which progressed to such extent that patient currently is unable to lift and hold the objects. Later on after 2 yrs of onset patient noticed; he is unable to lift a bucket full of water above the ground but he was able to comb his hair with left hand and is also able to lift his left hand overhead for purposeful work. Since last 1 yr there is no progression of weakness in either fingers or shoulder muscles. There was no involvement of right upper limb and both lower limbs. It was not associated with tingling or numbness also he is able to differentiate between cold and hot water during bath and able to feel clothes he is wearing on both upper and lower limb.

On Examination-
Patient is conscious Oriented to time, place and person
Blood pressure-110/80 mmHg
Pulse Rate-82 beats per minute

Systemic examination-

CNS-
1) Higher mental functions-Normal
MMSE-28/30

2) Power
(During flexion and extension)

	Right	Left
At shoulder joint At elbow joint	5/5	5/5
During flexion	5/5	5/5
During Extension	5/5	3/5
At wrist joint	5/5	3/5

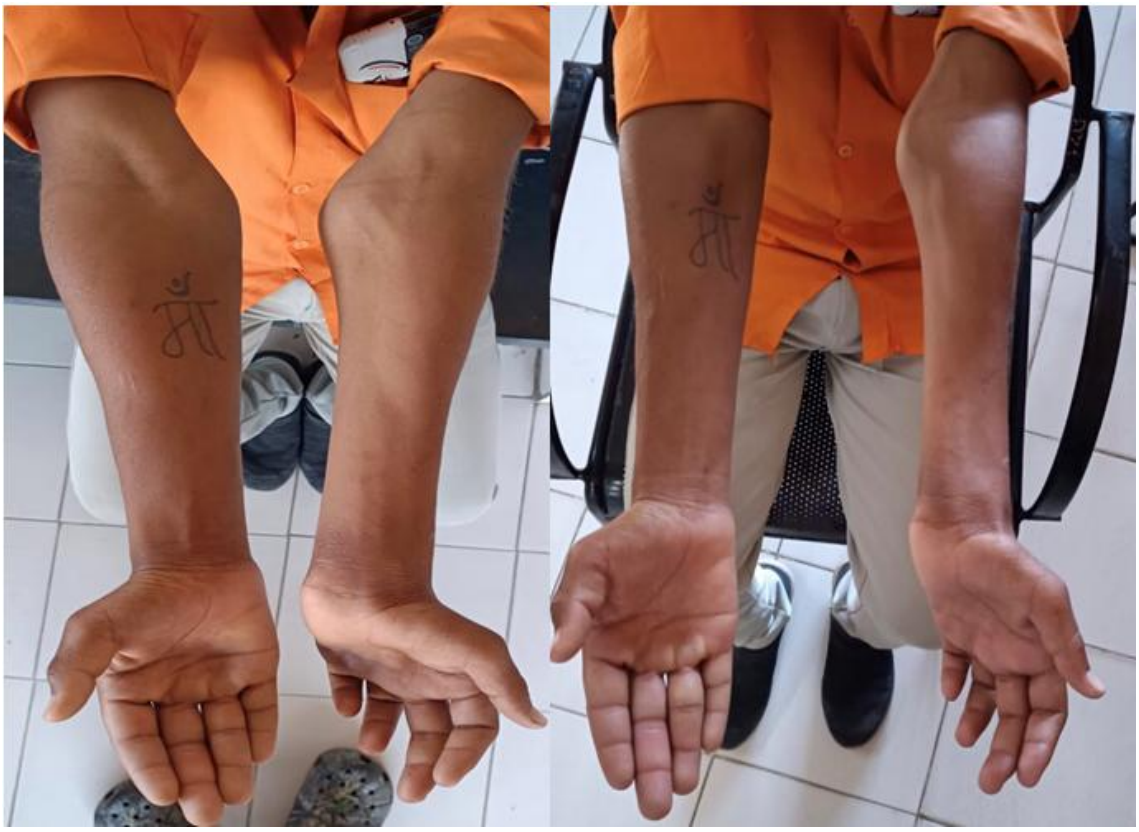
Intrinsic muscles of hands were completely normal on right side while on left side patient was unable to adduct /abduct / approx the thumb against other fingers. There was difficulty in abducting and adducting rest four fingers of left hand indicating weakness of interosseous muscles and lumbricals.

3) Bulk: There was a visible atrophy of intrinsic muscles of hands and forearm group of muscles and triceps on left side compared to that on right side.

With dimensions in upper limb as follows
Right UL-Above elbow 30 cm below elbow 24 cm
Left UL-Above elbow 26 cm below elbow 16 cm

4) Tone-Normal on right side, while there was hypotonia on left side (Triceps and forearm)
5) Reflexes-While all the Deep tendon reflexes and superficial reflexes on right side were normal, on left side triceps and supinator reflex were diminished, rest were normal
6) Sensory system-Normal on both sides
7) Cerebellum-No signs of cerebellar involvement and Gait was also normal
8) Cranial nerve-No signs of cranial nerve involvement
9) No signs of involvement of extrapyramidal system
10) No signs of involvement of Autonomic nervous system

CVS-Heart sounds normal, no murmur
RS-Breath sound bilaterally equal No adventitious sounds
P/A-Soft Non tender No organomegaly.



(Visible wasting and atrophy of left forearm as well as hand compared to the right forearm and hand)

Investigations

Lab values as follows

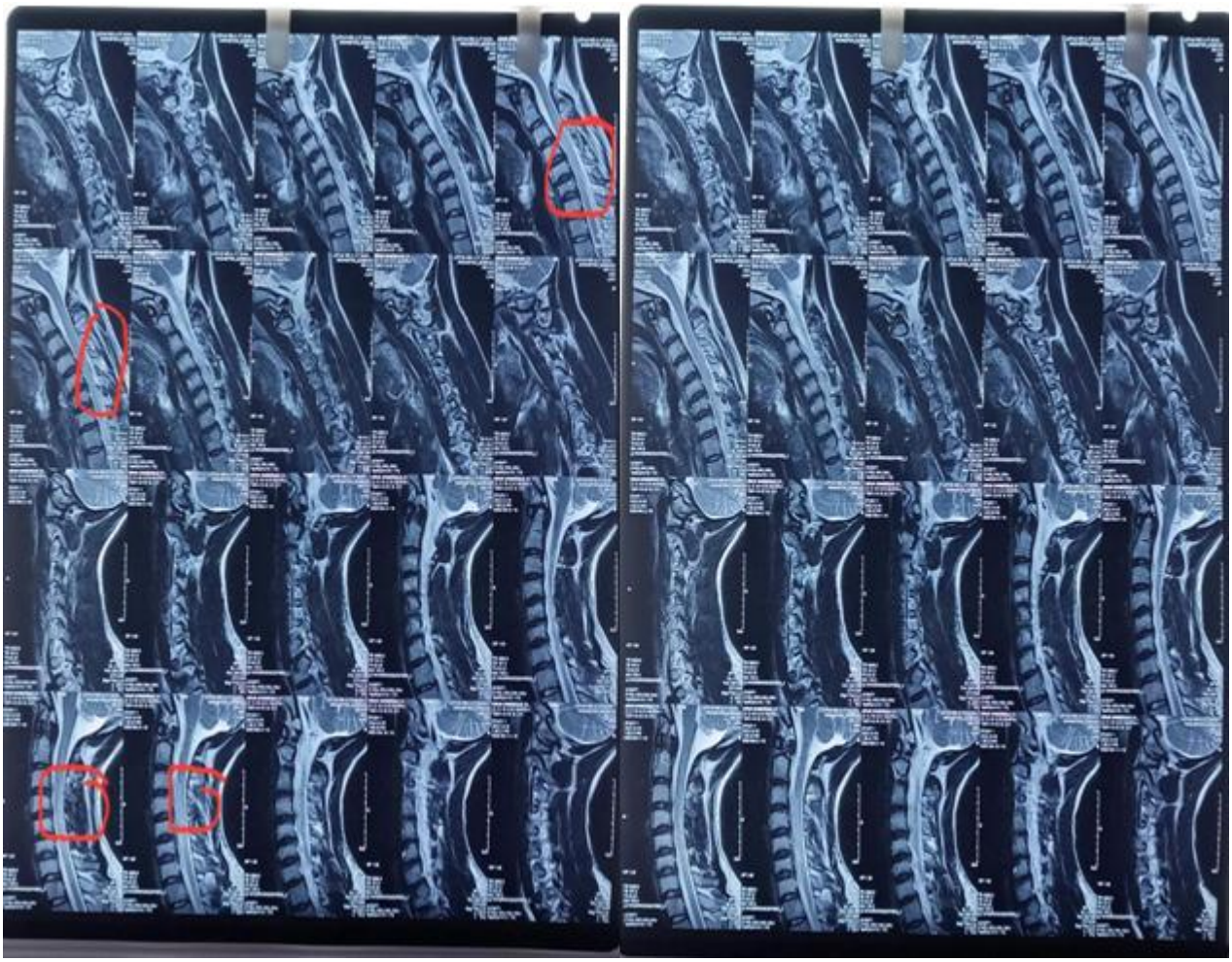
Hb-14.4gm/dl. Leucocyte count-8400/mcgL. Platelets-334000/mcgL

AST-32 U/L. ALT-42U/L. ALP-36U/L. Creatinine-0.7mg/dl. Urea-26 mg/dl

MRI spine in flexion and extension

MRI shows a crescent-shaped lesion in the posterior epidural space of the lower cervical cord with mild asymmetric cord flattening. [2] It is only with specialised flexion MRIs that the diagnosis can become confirmatory. Flexion MRIs have shown a distinct change in the alignment of the soft tissue structures, specifically an anterior shift of the cervical dural sac from the lamina at the C4-C7 region allowing for chronic micro trauma, with a corresponding post-contrast enhancement of the posterior epidural venous plexus indicative of ischemia. [3] following are the MRI Cervical spine images in flexion and extension showing

Crescent formation during flexion which vanishes during extension of neck (marked by Red circle)



3. Discussion

The incidence of Hirayama disease is very rare all over the world with diagnosed cases accounting just above 1400 cases. With confirmatory diagnostic test being MRI in flexion and extension of neck. Spontaneous arrest of progression of weakness in upper limb is characteristic hallmark of Hirayama disease. Physiotherapy, Avoidance of use of pillows during sleep, Neck muscle exercises and counselling is the only available treatment for the disease. With the help of MRI and proper clinical assessment of patient will help patient to get diagnosis and reduce psychiatric burden at the younger age.

The clinical diagnostic criteria for the Hirayama disease include the following:

- Distal upper limb muscle weakness and atrophy
- Onset at between 10-20 years of age with male preponderance
- Unilateral or predominantly asymmetric pattern
- Insidious onset, gradual progression followed by an arrest
- Lack of pyramidal signs or sensory disturbance, and
- Exclusion of other mimics.

The Huashan classification system [4] has been shown to be useful for the diagnosis and treatment of HD and is classified as

- Type 1-atrophy of hand and forearm muscles or asymmetric bilateral atrophy in upper limbs with type 1a (stable) and type 1b (progressive)
- Type 2-atrophy alongside pyramidal tract damage
- Type 3-atypical with atrophy of proximal upper limb muscle or symmetric upper limbs or presence of sensory dysfunction.
- A conservative approach is initially recommended for type 1 and 3 diseases, whereas surgery is recommended for type 2

The surgeries are based on the anterior cervical discectomy with fusion and plating showed the best outcome. Maintaining the physiological local lordosis angle is extremely important.

4. Conclusion

High clinical suspicion with the help of MRI of cervical spine in flexion and extension leads to diagnosis of this rare oblique atrophy which with the help of Neck extension exercises and physiotherapy will render the possibility of power improvement in patients with Hirayama disease.

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