Navigating Atypical Hirayama Disease: Insights from a case presentation

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Introduction

Hirayama disease, HD (previously known as juvenile muscular atrophy of the distal upper extremities first reported by Hirayama in 1959) is a rare neurological disorder that presents in early adolescence, typically males, affecting their hand and forearm muscles exhibit weakness and amyotrophy that is unilateral or asymmetric. The compression associated with HD is usually restricted to the lower cervical spinal cord, mainly at the C7-T1 level.

Unlike the classic variant, atypical Hirayama disease often exhibits a slower progression (in some cases it has been reported that it can progress over 10 years) and may involve additional neurological symptoms, such as sensory disturbances, upper motor neuron signs or more widespread muscle weakness(proximal and distal).

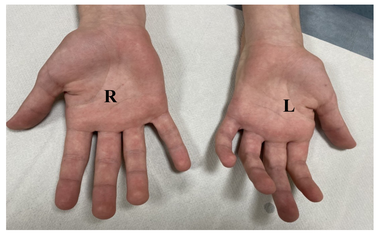
Case Presentation

- 15-year-old presented with progressive weakness in the left hand, poor grip, tremors, and reduced sensation (except for heightened temperature), and muscle wasting in his left hand. No useful function in the left hand
- Unremarkable past medical history. No history of preceding viral illness, trauma or back pain.
- No use of recreational drugs
- No bulbar dysfunction or gross motor difficulties
- Born of non consanguineous relationship and normal birth and early development.
- Nil significant past medical or family history of note.

Case Presentation (continue)

Examination findings:

- Wasting of the small muscles (thinner, hypothenar, and dorsal interossei muscles) of the left hand and forearm associated with both flexion and extension weakness of the fingers.(Figures 1,2)
- Normal power at the wrist, elbow and shoulder and both lower limbs.
- Reflexes was within normal limits in both biceps and was absent the supinator and triceps and the left.
- Lower limb showed brisk reflexes bilaterally associated with ill-sustained clonus and flexor plantar response.
- No fasciculation, normal gait, cranial nerves and cerebellar signs.
- Romberg's test was negative.



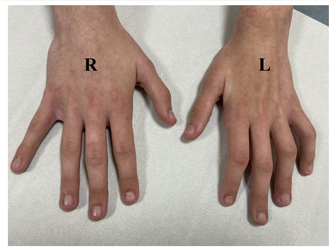
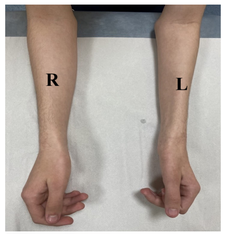


Fig 1: Muscle wasting in left thenar, hypothenar, interossei



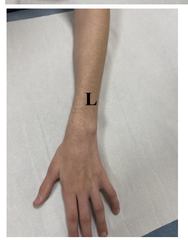
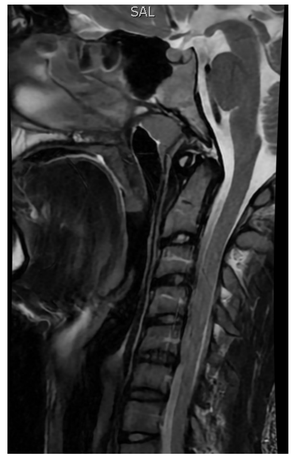


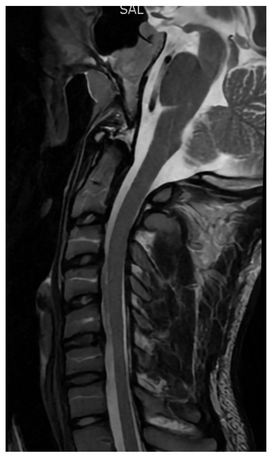
Fig 2: Muscle wasting in left forearm

Investigations

- Unremarkable blood tests, apart from low vitamin D and b12. Treated with adequate supplements.
- Normal genetic tests- SNP array, PMP22, FSHD gene
- Normal MRI brachial plexus
- **MRI cervical spine with flexion extension view**: focal myelomalacia at C5/C6 more so on the left with increase in the retrodural space during flexion and extension
- **NCV**: Sensory nerve conduction studies were normal in the upper and lower limbs, while motor studies revealed reduced amplitude in the left upper limb compared to the right
- **EMG**: chronic neurogenic changes observed in the several muscles of the left hand and forearm. Normal EMG of the right upper limb and both lower limb



MRI cervical spine Flexion view



MRI cervical spine Extension view

Discussion

- Hirayama disease primarily affects young males, causing unilateral or asymmetric weakness and muscle wasting of the hand and ulnar forearm.
- It typically stabilizes within five years, with factors like cervical cord compression during neck flexion and potential immunological elements contributing to its onset.
- Clinical manifestations include gradual weakness, coarse finger tremors, and sensory deficits, while imaging reveals fforward displacement of the posterior dura on flexing the neck, leading to cord compression.
- Atypical cases have pyramidal signs, proximal limb weakness, sensory involvement and long disease progression.
- Electrophysiological examinations show segmental neurogenic damage.
- Treatment aims to prevent neck flexion, initially with a cervical collar and potentially through surgery. Future research should focus on understanding HD's root causes and refining treatment strategies.

References

1. Hirayama K, Toyokura Y, Tsubaki T. Juvenile muscular atrophy of unilateral upper extremity: a new clinical entity. Psychiatric Neurol Jpn. (1959)
2. Juvenile muscular atrophy of unilateral upper extremity. Hirayama K, Tsubaki T, Toyokura Y, Okinaka S. *Neurology.* 1963
3. Sitt CM, Fung LW, Yuen HY, et al. Hirayama disease in a 17-year-old Chinese man. Singapore Med J 2014

Acknowledgement

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