Hirayama disease: a frequently undiagnosed condition with simple inexpensive treatment

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SUMMARY

Hirayama disease is a non-progressive asymmetric juvenile muscular atrophy involving C7-T1 myotomes leading to weakness and atrophy of intrinsic muscles of hand and forearm with relative sparing of the brachioradialis muscle. This benign focal cervical poliopathy is believed to be caused by forward displacement of the posterior cervical dural sac. In this case report we highlight the importance of dynamic MRI of cervical spine for diagnosis of the above condition and the use of hard cervical collar to prevent further flexion injury.

BACKGROUND

Hirayama disease (HD) or juvenile muscular atrophy of unilateral upper extremity was first described in 1959. This disease was more commonly reported from Asian countries like Japan and India and was thought to be rare in western countries. This condition usually affects young men in their second or third decade. Its characteristic appearance of oblique amyotrophy and relatively benign non-progressive course differentiate it from motor neuron disease. The most plausible cause of this focal anterior horn cell disease is impairment of microcirculation because of forward displacement of tight dura leading to dynamic compression on flexion. Lack of awareness about this condition and use of flexion cervical MRI for its diagnosis hampers its appropriate treatment in preventing further progression of disease.^{1 2}

CASE PRESENTATION

A 20-year-old college student was brought to our department with complaints of difficulty in holding a pen while writing and breaking morsels. He had also noticed a thinning of both hands since 8 months. The weakness and thinning of small muscles were insidious in the onset and had progressed to an extent that the patient could not perform fine tasks like holding a pen or a cup of tea. There was history of intermittent fasciculations occurring in the thinned-out part of the forearm and right hand. He also complained of tremulousness of his fingers. He had started working out in the gym since a year ago, which included neck exercises and weight lifting up to 50 kg apart from other exercises, for about an hour each day. The neck movements during exercise comprised active as well as passive strain, especially neck flexion during strenuous upper-extremity exercise. He had not exercised with such intensity previously. There was no history of weakness involving proximal muscles of arm and lower

limbs, neck pain, sensory complaints and bladder or bowel features.

Examination revealed normal pulse and blood pressure, a body mass index of 20.7 kg/m², no hypopigmented patch, ulcers, scars or thickened nerve were detected on examination. Cranial nerve examination was normal. On Motor system examination, wasting of small muscles of hand was noted on both sides (right > left; figure 1). There was preservation of brachioradialis muscles on both sides, with wasting noted in the medial aspect of the forearm. Power testing was normal in all the muscle groups in both upper limbs and lower limbs except for intrinsic muscles of hand. Deep tendon jerks and superficial reflex were normal. Sensory system examination showed normal pain, temperature, fine touch and vibration sense. Polyminimyoclonus was seen in the right hand. No cerebellar signs or any other abnormal movements were seen.

INVESTIGATIONS

MRI cervical spine was suggestive of loss of posterior dural attachment and anterior shifting of lower cervical cord on flexion from C3 to C7, subtle hyperintensities along with thinning of the cervical cord (figures 2–4). Nerve conduction studies showed normal sensory nerve action potentials, reduced amplitude of right ulnar compound action potentials due to atrophy of muscles. Needle electromyography (EMG) revealed a neuropathic pattern like large-amplitude Motor Unit Action Potentials. Fasciculation, fibrillations and reduced recruitment was seen in both first dorsal inter-osei,

Figure 1 Photographs of both hands and forearm showing wasting of small muscles of hand (right > left) and medial aspect of forearm with preservation of brachioradialis.

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Figure 2 MRI cervical spine in neutral position. Thinning of the cervical cord. Short TI inversion recovery images show cord hyperintensity from C6 to C7.

abductor pollicis brevis and adductor digiti minimi. Deltoid and Biceps needle EMG was normal.

DIFFERENTIAL DIAGNOSIS

- ▶ C8-T1 radiculopathy
- ▶ Ulnar neuropathy
- ▶ Syringomyelia
- ▶ Hansen's disease
- ▶ Amyotrophic lateral sclerosis
- ▶ Spinal cord tumors

TREATMENT

Despite a time lag of 8 months between the time patient first noticed his symptoms and diagnosis of this condition, a trial of cervical collar was given to the patient.

OUTCOME AND FOLLOW-UP

On follow-up visit after 2 months, the patient had a subjective sensation and better handgrip strength. However, there was no change in the power in affected muscles. Importantly, there was no further deterioration in power or increased atrophy in the affected muscles.

Figure 3 (A and B) Postcontrast image sagittal view of the cervical spine in flexion shows the movement of dura on flexion along with venous enhancement.

DISCUSSION

HD or juvenile muscular atrophy of distal upper extremity is characterised by insidious onset unilateral or asymmetric weakness of muscles in C7, 8 and T1 myotomes. It usually progresses gradually and only during the initial phases of illness. About two-third of patients do not worsen clinically after about 5 years of disease onset.¹ Men are more commonly affected and it usually manifests during the second or third decade of life. The higher incidence of HD was noted in Japanese and later even Indian men. This was initially thought to have some ethnic or regional significance.¹⁻⁴ However, this condition is not limited to Asian population, as cases have been reported even from Europe and America.⁵⁻⁶ The exact data regarding the epidemiology of this condition are scarce. Tashiro *et al* studied the epidemiology of HD in Japan with the help of two nationwide questionnaire-based surveys, from 1996 to 1998, and identified 333 cases. They found that the peak age was 15–17 years, and was more common in men.⁷ Huang *et al*⁸ presented data of 40 patients in Taiwan. The data on HD in western countries are limited to few case reports only. Ghosh *et al*⁶ described HD in six children from North America. Further, a number of cases of HD remain undetected, probably as a result of lack of diagnostic criteria and underdeveloped health services in developing nations.

In HD, weakness and wasting are noted in small muscles of hand along with the preservation of brachioradialis muscle. The weakness usually involves both extensors and flexors of fingers. The tremulousness of hand on extension of the fingers (polyminiomyoclonus) is often seen. Most patients also report aggravation of weakness in cold weather, that is, cold paresis. The electrophysiological studies by Kijima *et al*⁹ suggested active denervation leading to conduction block of the muscle fibre membrane in reinnervating muscles as a plausible aetiology of cold paresis.¹⁻²

Ever since this condition was first described in 1959 many theories have been postulated regarding its aetiology and associated precipitating factors. The aetiopathogenesis of HD is still unclear. In the first ever autopsy done of patients suffering from HD, Hirayama *et al*¹⁰ concluded asymmetric atrophy, gliosis and destruction of the lower cervical cord was due to ischaemia of the lower cervical cord. In 1980, Singh *et al*³ postulated that these patients had atypical poliomyelitis like viral infection causing localised anterior horn cell disease.

Figure 4 (A and B) MRI cervical spine in extension and (C) T2 axial MRI at C7 level shows atrophy.

According to various imaging and pathological studies, HD seems to be caused by dynamic compression of the lower cervical cord due to repeated or sustained neck flexion.^{1 2 6 11–13} We believe that our patient may also have sustained flexion injury of cervical cord due to strenuous exercise leading to HD. During the growth spurt, there is disproportionate growth of the spinal canal compared to dura, which leads to a tight dural sac surrounding the spinal cord.¹¹ Normally, the loosely attached dural sac provides room for lengthening of cervical spine during flexion. However, the presence of tight dural sac causes anterior displacement of posterior dural wall and cord compression on flexion of neck. The compressed cervical segment may further lead to rise in intramedullary pressure causing impaired perfusion of microvasculature in anterior horn cell leading to ischaemia and necrosis.^{1 2 11 13}

Dynamic MRI of cervical spine (neutral and flexion position) is the imaging modality of choice for the diagnosis of HD. However, due to lack of awareness about this illness, many clinicians fail to advise flexion cervical spine MRI.^{2 6 13} Routine cervical spine MRI will show lower cervical cord atrophy, hyperintensity on T2-weighted imaging in the anterior part of cord in the absence of medullary compression and loss of normal cervical curvature.^{12 13} Loss of attachment of the posterior dura mater in neutral position is considered one of the most valuable signs of HD.¹⁴ Flexion MRI of the cervical spine shows anterior displacement of posterior wall of dura mater and appearance of epidural flow voids which enhance on gadolinium contrast injection. In a recently conducted study, Hasan *et al* concluded that neutral position cervical MRI showing asymmetrical cord atrophy and anterior shift of posterior dural wall along with flow voids in the posterior epidural compartment is highly predictive in HD. All the above mentioned findings were seen in our patient.^{2 12–14}

In our case, the patient developed weakness and atrophy after he started weight training. In our opinion, this was probably the precipitating factor for flexion injury to lower cervical cord. Although his neutral position cervical spine MRI did not show loss of attachment, the flexion cervical MRI studies revealed displacement of posterior dural wall along with enhancement of posterior epidural venous plexus and asymmetrical cord atrophy.

Treatment of HD is targeted to prevent further flexion injury of cervical cord by inadvertent repeated neck flexion.¹ Hence, many authorities recommend the use of hard cervical collar to prevent any further flexion injury of the cervical cord. The

benefits are more likely in patients who have been diagnosed early and have milder cord atrophy.¹⁵

Learning points

- ▶ Hirayama disease is a rare clinical disorder, characterised by the presence of wasting and weakness of hand musculature, polyminimyoelonus and cold paresis.
- ▶ The aetiopathogenetic factors are uncertain, although ischaemia, pressure of engorged vertebral venous plexus and dynamic flexion injury on lower cervical column structures have been postulated.
- ▶ The application of hard cervical collar during the early phase of illness can halt the progression of illness.

Competing interests None.

Patient consent Obtained.

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