Unusual presentation of more common disease/injury

A 20-year-old female with Hirayama disease complicated with dysplasia of the cervical vertebrae and degeneration of intervertebral discs

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Summary

A 20-year-old female patient was presented with a 1-year history of progressive weakness of the left hand. Examination on admission showed atrophy of the muscles of the left forearm, cold paralysis and minipolymyoclonus. MR images of the cervical cord showed anterior transfer of the cervical cord on anterior flexion and cervical cord compression at the site of cervical kyphosis, confirming the diagnosis of Hirayama disease. Many features of the present case are unusual: the patient is a female (who are rarely afflicted by this disease), with cervical kyphosis and a history of exercise involving cervical vertebral loading, suggesting a potential involvement of the latter two factors in the disease onset. The findings suggest that cervical vertebral dysplasia and intervertebral disc degeneration may influence cervical kyphosis, and be involved in the onset of Hirayama disease.

BACKGROUND

Hirayama disease (juvenile unilateral muscular atrophy of the upper extremity) is a disease entity unrelated to motor neurone diseases.^{1–3} Details of the clinical presentation and pathogenesis of Hirayama disease remain to be clarified. The main manifestation of Hirayama disease is muscle weakness/atrophy localised to the unilateral hand and forearm. The condition is frequent in young men and shows progressive deterioration in the initial stages, but later stabilises, with favourable prognosis.⁴ We present a female patient with the disease who showed cervical vertebral dysplasia and intervertebral disc degeneration and discuss their roles in the onset of Hirayama disease.

CASE PRESENTATION**Case**

A 20-year-old woman.

Complaint

Weakness and tremor in the left hand.

Present illness

At the age of 19 years, weakness of the left hand was noted during exercise. The condition worsened gradually, disabling the patient from holding objects such as tableware. At the age of 20 years, tremor in the left hand began, making it difficult to hit the computer keyboard with the left little finger. However, the patient was able to use the mobile phone with the left thumb. She described occasional difficulty in moving the left fingers in winter.

Medical history

Bronchial asthma, but no recent episodes.

Agents routinely employed

None.

Lifestyle

The patient was a member of a cheerleader team since 18 years of age.

Alcohol

Occasional alcohol consumption.

Smoking

No history of smoking.

Family history

Not significant.

Physical examination on admission

Her height was 148.7 cm and the body weight 46.7 kg. The arterial blood pressure was 120/58 mm Hg. Minipolymyoclonus of the second-to-fifth fingers was observed on muscle contraction, which appeared on the extension of the left finger. A slight muscle atrophy was noted on the ulnar side of the left forearm, but no such atrophy was noted on the radial side. Compared with the right hand, there was marked atrophy of the left dorsal hand muscles, but atrophy of the left hypothenar eminence and left thenar muscles was mild. Atrophy of C₇ muscles with preserved brachioradialis belly was observed. During admission, no deterioration of weakness of muscles of the left upper extremity was noted in relation to postural changes such as cervical anterior flexion. The deep tendon reflex was normal, and no laterality or pathological reflex was evident. No abnormalities were found in the superficial/deep-sensory systems, and no vesicorectal disorders were evident. A manual muscle test of the left

Table 1 Results of the manual muscle test

	Right	Left
Hand spination	5	4
Hand pronation	5	4
Flexor carpi radialis	5	4
Extensor carpi ulnaris	5	4
Extensor digitorum	5	3
Abductor pollicis longus	5	4
Extensor pollicis longus	5	4
Extensor pollicis brevis	5	4
Flexor carpi ulnaris	5	4
Abductor digiti minimi	5	4
Flexor digiti minimi	5	4
First dorsal interosseous muscle	5	3

upper extremity showed marked weakness of the left upper extremity at the C₇, C₈ and Th₁ myelomere levels (table 1). Needle electromyography of the left upper extremity (table 2) showed prolonged high tonic activity in the flexor carpi radialis and extensor carpi digitorum muscle. However, there were no abnormalities in the extensor carpi radialis. Nerve conduction studies of the left median and ulnar nerves showed no conduction block and ruled out multifocal motor neuropathy, neuritis of the brachial plexus or the Lewis–Sumner syndrome (table 3).

Laboratory data

Blood cell counts, blood biochemistry and cerebrospinal fluid examination were within the normal ranges.

Imaging findings

Median-position radiographs of the cervical vertebrae showed cervical kyphosis. Alignment was irregular at the C₄, C₅ and C₆ levels. The anteroposterior intervertebral diameter was within the normal range. Only mild narrowing of the intervertebral disc space was noted. Neither anterior nor posterior flexion was markedly limited. Kyphosis with peaks at the C₅ and C₆ levels became more marked in the anterior flexion position. MRI of the cervical cord (T₂-weighted images) showed cervical kyphosis with a peak at the C₆ level on the median sagittal plane (figure 1a). Marked restriction was noted visually at the C₂–C₇ levels. In addition, the intervertebral disc showed low-signal intensity. Kyphosis with a peak at the C₆ level became more marked in the anterior flexion position

Table 2 Results of the needle electromyography

Voluntary contraction	Muscles tested		
	lt. FCR	lt. ECR	lt. ECD
Normal NMU	+	+++	-
Low-amplitude NMU	-	-	-
High-amplitude NMU	++	-	+++
Polyphasic NMU	-	-	-
Long duration	++	-	+++
Short duration	-	-	-
Grouping	-	-	-
Waning/waxing	-	-	-
Interference	↓↓	N↓	↓↓
Spike number decrease	++	-	+++

ECD, extensor carpi digitorum; ECR, extensor carpi radialis; FCR, flexor carpi radialis; lt, left; NMU, neuromuscular unit; N, normal.

Table 3 Results of the nerve conduction study

Nerve	MCV (m/s)	TL (m/sec)	CMAP (mV)		SCV (m/sec)		SNAP (μV)		FCV (m/sec)
			D	P	D	P	D	P	
Median lt.	64.2	3.0	16.1	13.1	55.6	68.0	93.0	39.4	69.7
Rt.	64.3	2.9	16.4	16.3	58.6	70.5	83.2	40.2	61.2
Ulnar lt.	57.0	2.7	8.2	7.5	47.6	56.5	48.0	22.4	64.5
Rt.	53.0	2.7	11.7	9.0	42.7	57.5	59.1	24.2	62.5

CMAP, compound muscle action potential; D, distal; FCV, F-wave conduction velocity; lt., left; MCV, motor nerve conduction velocity; P, proximal; rt., right; SCV, sensory nerve conduction velocity; SNAP, sensory nerve action potential; TL, terminal latency.

(figure 1b). The posterior dural wall showed anterior transfer at C₅ and C₆ levels. There was a flow void between the posterior dural wall and posterior wall of the spinal canal. This flow void disappeared in the posterior flexion position (figure 1c). In horizontal sections, there was a crescent-type flow void between the posterior dural wall and posterior wall of the spinal canal in the anterior

Figure 1 T₂-weighted MRI images of the cervical cord. Top row: Sagittal images. Bottom row: Axial images at the C₆ vertebral body level. (a) Median position, (b) anterior flexion position, (c) posterior flexion position. (a) Cervical kyphosis with peak at the C₆ level. Vertebral body alignment was irregular at the C₂–C₇ levels. In addition, vertebral body dysplasia was noted. Note the small size of the C_{5,6} vertebral bodies compared with other cervical vertebral bodies. Note also the reduced intensity of the intervertebral discs, suggestive of degeneration. (b) Note the marked cervical kyphosis with a peak at the C₆ level in the anterior flexion position. At the C₅ and C₆ levels, the posterior dural wall showed anterior transfer, and there was a flow void between the posterior dural wall and posterior wall of the spinal canal. This flow void disappeared in the posterior flexion position. On axial images, there was a crescent-type flow void between the posterior dural wall and posterior wall of the spinal canal in the anterior flexion position. The cervical cord adhered to the anterior vertebral body, predominantly showing aplation on the left side, that is, the affected side. (c) In the posterior flexion position, there was no anterior transfer of the posterior dural wall. Note the disappearance of the flow void between the posterior dural wall and posterior wall of the spinal canal.

flexion position (figure 1b). The cervical cord adhered to the anterior vertebral body, showing predominant apposition on the left side, that is, the affected side. The asymmetric cord-flattening provided a further support for the diagnosis of Hirayama disease.

INVESTIGATIONS

OUTCOME AND FOLLOW-UP

We prescribed the use of a cervical collar to reduce the loads to the cervical spine. The progression of the symptoms was not seen for 1 year.

DISCUSSION

Hirayama disease is frequent among men⁴ and only 10.8% of the patients in Japan are women.⁵ Similarly, reports from other Asian countries, with a high prevalence of the disease, estimate the proportion of female patients to be approximately 10–15%.^{6–8} The reason for the marked gender difference remains to be clarified. However, several factors that may be involved in the onset of the disease were identified in the present case.

In the present case, cervical cord MRI showed cervical kyphosis, with a peak at the C₆ level, in the median portion of the cervical spine (figure 1a). On the anterior cervical flexion, cervical kyphosis became more marked, and an anterior transfer of the cervical cord and cervical cord compression at the site of cervical kyphosis were observed (figure 1b). The disappearance of physiological lordosis of the cervical vertebrae was reported by Hashimoto *et al*⁹ in 11 of 15 patients, Sonwalkar *et al*¹⁰ in 6 of 8 patients, Raval *et al*¹¹ in 9 of 9 patients and Hassan *et al*¹² in 10 of 11 patients. Abnormal cervical curvature is an important finding on the cervical MRI for the diagnosis of Hirayama disease.¹³ Masaki *et al*¹⁴ performed posterior fixation in patients with cervical kyphosis, which resulted in the relief of symptoms. Based on these studies, cervical vertebral alignment abnormalities, such as cervical kyphosis, as observed in the present case, seem to enhance anterior compression of the cervical cord on cervical flexion, thus contributing to the pathogenesis of this disease. In addition, our patient showed abnormalities in the cervical vertebral bodies and intervertebral discs. The size of the C_{5,6} vertebral bodies was smaller compared to other cervical vertebral bodies on the cervical MRI. Furthermore, low-signal intensity on MRI T₂-weighted images indicated degeneration of the intervertebral discs. Concerning cervical vertebral dysplasia, Takahashi *et al*¹⁵ reported a patient with Hirayama disease and spondyloschisis. It is not clear whether the cervical vertebral dysplasia and intervertebral disc degeneration in our patient are congenital or acquired. However, these factors may be involved in the development of cervical kyphosis, and are likely to be important factors that influence the pathogenesis of this disease.

The role of exercise must be considered in this patient. Hashimoto *et al*⁹ examined the relationship between sports-related and traffic-accident-related head/neck injury and Hirayama disease. In the present case, Hirayama disease developed 6 months after the start of cheerleading. Cheerleaders often make several jumps/landings from high places, as well as lift other team members on both shoulders; thus potentially causing heavy cervical load,

implicating cheerleading activities at the onset of Hirayama disease.

Autopsy findings of Hirayama disease suggest that the disease aetiology is necrosis of the motor neurones in the anterior cornua of the spinal cord.¹⁶ The anterior cervical flexion-related anterior transfer of the spinal cord may result in anterior compression of the spinal cord by the cervical vertebrae, and congestion of the venous plexus may affect microcirculation, leading to necrosis of the cells of the anterior cornua of the spinal cord, which are most markedly influenced in the presence of ischaemia,¹⁷ through repeated transfer.¹⁸ Based on the findings of several studies, the reasons for the repeated anterior transfer of the spinal cord include (i) the overstretch hypothesis (growth imbalance between the cervical vertebrae and cervical cord/pachymeninges may lead to overextension of the spinal cord in the anterior flexion position of the cervical vertebrae, resulting in compression of the cervical cord at the site of cervical kyphosis);¹⁹ (ii) reduction in the extension of the dural canal;²⁰ (iii) loss of the epidural ligament²¹ and (iv) unfavourable extension of the nerve roots.²² Based on these studies, the aetiology of Hirayama disease may differ among patients. The present case showed cervical vertebral dysplasia, intervertebral disc degeneration, cervical kyphosis and a history of exercise. To our knowledge, cervical vertebral dysplasia (small cervical vertebral bodies) and intervertebral disc degeneration have not been reported in patients with Hirayama disease. The present case may be important as it clarifies certain aspects of the aetiology and pathogenesis of Hirayama disease. The above two abnormalities may be involved in the marked cervical kyphosis, thus influencing the onset of Hirayama disease. In addition, in the present case, exercise with cervical load may also have contributed to the disease onset. We believe that several aetiological factors, rather than a single factor, are involved in the onset of Hirayama disease.

Learning points

- ▶ To our knowledge, no case of this disease has been reported with cervical vertebral hypoplasia and intervertebral disc degeneration.
- ▶ These factors may be involved in the marked cervical kyphosis, thus influencing the onset of Hirayama disease.
- ▶ In addition, in the present case, exercise with cervical load may also have contributed to the disease onset.
- ▶ We believe that several aetiological factors, rather than a single factor, are involved in the onset of Hirayama disease.

Competing interests None.

Patient consent Obtained.

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