CASE REPORT

Late-onset Hirayama disease presenting with ulnar neuropathy: A case report

Meral Bilgilisoy Filiz,[1] Tuncay Cakir,[2] Naciye Füsun Toraman,[3] Sebnem Koldas DoGan,[4] Iclal Erdem Toslak,[5] Hilmi Uysal[6]

Abstract

Hirayama disease (HD) was first reported by Hirayama et al. in 1959. The disease is considered as a type of benign focal motor neuron disease that primarily affects upper limbs of young males. In this case report, we present a man aged 40-years with rapidly progressive weakness and atrophy in his left hand. The findings of nerve conduction studies were consistent with left ulnar neuropathy at the elbow. Flexion magnetic resonance imaging (MRI) revealed minimal enlargement of the posterior epidural space and anterior displacement of the spinal cord. After exclusion of relevant diseases the patient was diagnosed as having Hirayama disease with ulnar neuropathy. Mild ulnar entrapment at the elbow may be considered as a clinical feature of HD. Therefore, it is recommended that young male patients with wasting in upper extremities with findings of ulnar entrapment should not be judged to have ulnar neuropathy before HD has been carefully excluded.

Keywords: Juvenile muscular atrophy, Monomelic amyotrophy, Ulnar entrapment.

Introduction

Hirayama Disease (HD), also known as juvenile muscular atrophy or monomelic amyotrophy, was reported first by Hirayama et al. in 1959.[1] The disease is considered as a type of benign focal motor neuron disease that affects primarily upper limbs of young males. The pathogenesis is unclear. Dynamic compression that results in spinal cord ischaemia caused by cervical flexion is speculated as a causative factor.[1,2] Herein we present the case of a late-onset Hirayama disease in a 40-year old male presenting with ulnar neuropathy and his one year follow-up.

Case Report

A previously-healthy right-handed man aged 40 years who was obese (body mass index: 37 kg/m^2) and worked as a general surgeon, presented to our outpatient clinics of Physical Medicine and Rehabilitation Department of Antalya Training and Research Hospital, Antalya, Turkey, on December 2013 with symptoms of having difficulty grasping small objects and progressive wasting in his left hand, which he had had for two months. He had no trauma, neck or back pain, bulbar dysfunction or relevant familiy history. There was no sensory deficit but the patient had a slight weakness in abduction-adduction of the left hand fingers, which was graded as a Medical Research Council grade 4. Atrophy was prominent in the first dorsal interosseal muscle and less obvious in the hypothenar muscles (Figure-1). There was no evidence of fasciculations or spasticity. The cranial nerves and deep tendon reflexes were normal.

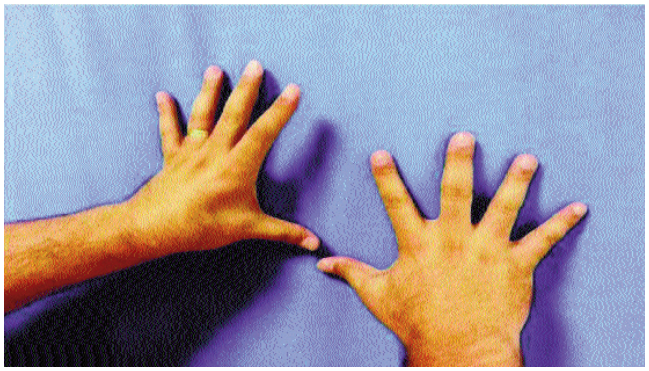

Figure-1: Atrophy of the first dorsal interosseal muscle.

Laboratory tests including complete blood count, electrolytes, blood sugar, liver, renal and thyroid function tests, creatine kinase, protein and immune fixation electrophoresis, vitamin B12, antinuclear antibody, and rheumatoid factor were within normal limits. Electrophysiologic examinations were performed. Nerve conduction velocity of the above elbow segment was >10 m/s slower than the below elbow segment of the ulnar nerve (61.1m/s vs 39.5m/s). An inching study of the ulnar nerve performed in the elbow region with 2 cm intervals revealed a significant delay between the elbow-2 cm distal segment latancies, which suggested ulnar neuropathy at the elbow level (6.70 ms vs 7.80 ms).

Although the amplitude of compound muscle action potentials (CMAP) of the left ulnar nerve were within normal limits, they were lower than the asymptomatic side. The other conduction studies of ulnar, median, and

[1-4]Department of Physical Medicine and Rehabilitation, [5]Department of Radiology, Antalya Training and Research Hospital, [6]Department of Neurology, Akdeniz University, School of Medicine, Antalya, Turkey.
Correspondence: Meral Bilgilisoy Filiz. Email: mbilgilisoy@gmail.com

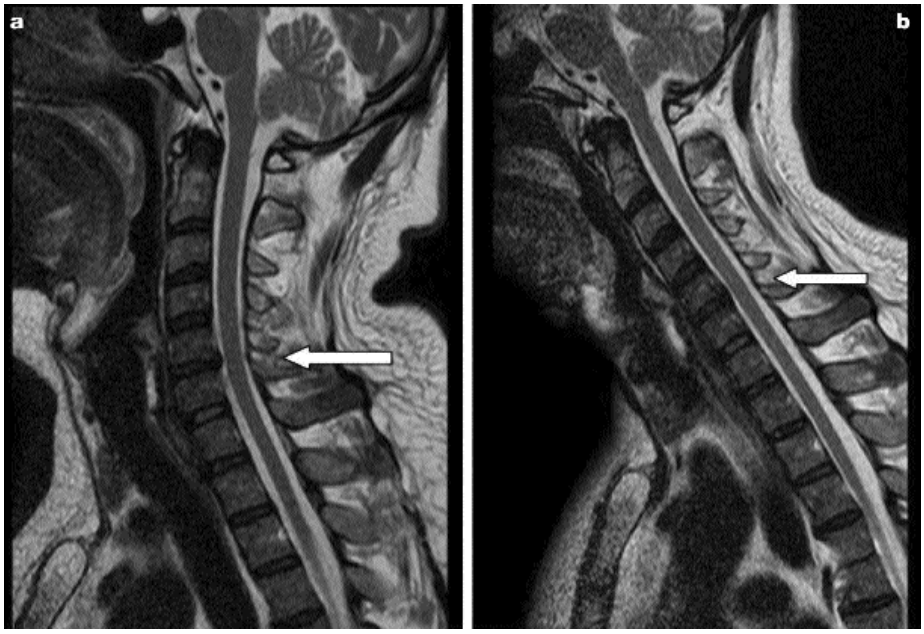

Figure-2: a) Cervical magnetic resonance imaging T2 TSE, loss of normal cervical lordosis and mild degenerative changes of the intervertebral discs with a protrusion at C5-C6 level (white arrow). b) Flexion magnetic resonance imaging -T2 TSE, minimal enlargment in posterior epidural space (white arrow).

medial antebrachial cutaneous nerves of both upper extremities, radial nerve of left upper extremity, peroneal, tibial and sural nerves of left lower extremity, F waves of right and left median and ulnar nerves were considered normal. In a needle-electrode electromyography (EMG) study, abnormal spontaneous potentials (fibrillations, positive sharp waves) were detected in the left first dorsal interosseous, abductor pollicis brevis, extensor carpi radialis longus, deltoid, and C5-C8 left cervical paraspinal muscles, and an increased rate of polyphasic potentials of the left first dorsal interosseous, abductor pollicis brevis, extensor carpi radialis longus consistent with acute-subacute denervation. There was no evidence of neurogenic injury in the lower limbs or the asymptomatic contralateral upper limb. Amplitudes and latencies of motor-evoked potentials of upper and lower extremities on both sides were normal, which showed that there was no dysfunction of the corticospinal motor system.

Magnetic resonance imaging (MRI) of the cervical spine revealed a loss of normal cervical lordosis and mild degenerative changes of the intervertebral discs with a protrusion at the C5-C6 level. In the dynamic flexion MRI,which was performed owing to the suspicion of HD, there was minimal enlargement of the posterior epidural space and anterior displacement of the spinal cord, but no obliteration of anterior subarachnoid space or myelopathy were detected (Figure-2). MRI of the elbow was also performed, which showed no abnormalities that affected signal intensity or thickness of the ulnar nerve at the elbow segment. MRI of the brain with diffusion tensor imaging (DTI) was performed to exclude dysfunction of the corticospinal tracts and the results were considered normal.

Ultimately, after diseases in the differential diagnosis of HD such as amyotrophic lateral sclerosis (ALS), dysfunction of the corticospinal tracts, syringomyelia, radiculopathy, brachial plexopathy were excluded, the patient's clinical, electrophysiologic, and imaging features were found to be consistent with the diagnosis of HD. The patient was recommended to rest and given a neck collar to prevent neck flexion. A physical therapy programme was prescribed including mild strengthening exercises and therapeutic electrical stimulation to the affected muscles.

During his one-year follow-up, no disease progression was observed except for a slight atrophy in the medial border of the left forearm, which added to the interosseal and hypothenar muscle wasting. In his follow-up electrophysiologic tests at three months, there was still a reduction in motor conduction velocity of the ulnar nerve at the across elbow segment (60.8 m/s vs 39.5 m/s), but no significant localization was detected in an inching study. He was able to return to work, but reduced the frequency of operations.

Consent of the patient was taken prior to the writing of the manuscript.

Discussion

Hirayama disease is a rare condition that typically affects the distal upper extremities of young males aged between 15-25 years.[1,2] However, cases of HD up to 53 years have also been reported.[2] The disease is usually sporadic and incidence is greater in East Asian countries.[3]

The pathophysiologic mechanisms of HD remain unclear. Although it was considered as a special type of motor neurone disorder, recent studies and case reports indicate that it may be associated with dynamic changes such as

ischaemia, myelopathy or atrophy of the spinal cord induced by neck flexion.[2,4] In our patient, flexion MRI revealed minimal enlargment of the posterior epidural space and anterior displacement of the spinal cord without myelopathy or cord atrophy. These dynamic findings may be seen in 87% of patients with HD. However, inadequate flexion of the neck during MRI may be reported as "normal" with absence of dynamic changes even though the patient has the disease.[1,4] The presented patient was obese with an excessive amount of fat tissue around his neck; therefore, we are not sure if he was able to flex his neck sufficiently. Also, it has been reported that dural displacement and cord atrophy were more prominent in younger patients with progressive disease.[3] The age of the patient and short duration of symptoms might be other reasons that account for the lack of myelopathic changes.

The electrophysiologic investigations in HD show normal sensory conduction velocities (SCV), sensory nerve action potentials (SNAP), and terminal latencies.[5] Amplitude of CMAP may be reduced in affected and atrophied muscles.[1] Guo et al. reported that 50% of their patients with HD showed a reduction of ulnar CMAP amplitude.[5] In another study, abnormally low median and ulnar CMAP amplitudes were found in 22% and 78% of patients, respectively.[6] Our patients's SNAP, CMAP amplitude, and SCV values of median, ulnar and medial antebrachial nerves, and motor conduction velocity (MCV) of the median nerve were normal, but CMAP amplitudes of the left ulnar nerve were lower than the right side and there was an abnormal focal slowing of the ulnar MCV across the elbow, which may be considered as ulnar neuropathy. Similarly, Lyu et al.[6] reported that there was electrophysiologic evidence of ulnar motor entrapment at the elbow in 23% of patients with HD, and 9% of patients with ALS, and they suggested that patients should not be judged to have ulnar neuropathy before the diagnosis of HD had been carefully excluded. Also, Chaudhry and Clawson[7] reported that observation of mild ulnar entrapment at the elbow might be considered as an important clinical feature in HD. In our patient, MRI of the elbow revealed no alterations in signal intensity or thickness of the ulnar nerve at the elbow segment. Thus electrophysiologic findings of the ulnar nerve were considered to be a feature of HD.

Conservative treatments such as avoidance of neck flexion by using a cervical collar is always the first-line treatment because the progression of the disease will generally spontaneously arrest within 5 years.[1,8] Even if randomized controlled trials and consensus about the type of the surgery are lacking, surgical interventions such as anterior or posterior decompression with or without fusion may be reasonable for pateints who do not respond to conservative treatment.[9]

Conclusion

Hirayama disease is considerably rare and can therefore be easily overlooked or misdiagnosed. Although HD generally affects young males, the disease should also be kept in mind for middle-aged patients with progressive asymmetric hand atrophy. Overdiagnosis of ulnar entrapment or unnecessary surgical interventions should be avoided in these patients, even if electrophysiologic studies give the impression of ulnar neuropathy at the elbow.

Disclaimer: Our case report was presented as an abstract at "8th World Congress of Neuro Rehabilitation" April 8-12, 2014, Istanbul-Turkiye.

Conflict of Interest: None to declare.

Funding Disclosure: None to declare.

References

1. Hirayama K. Juvenile muscular atrophy of distal upper extremity (Hirayama Disease). Intern Med 2000; 39: 283-90.
2. Patel DR, Knepper L, Jones HR Jr. Late-onset monomelic amyotrophy in a Caucasian woman. Muscle Nerve 2008; 37: 115-9.
3. Sarawagi R, Narayan S, Lakshmanan PR, Chakkalakkoombil SV. Hirayama Disease: Imaging profile of three cases emphasizing the role of flexion MRI. J Clin Diagn Res 2014;8: 3-4.
4. Cortese R, Gerevini S, Dicuonzo F, Zoccolella S, Simone IL. Hirayama disease: the importance of an early diagnosis. Neurol Sci 2015; 36: 1049-50.
5. Guo XM, Qin XY, Huang C. Neuroelectrophysiological characteristics of Hirayama disease: report of 14 cases. Chin Med J (Engl) 2012; 125: 2440-3.
6. Lyu RK, Huang YC, Wu YR, Kuo HC, Ro LS, Chen CM, et al. Electrophysiological features of Hirayama Disease. Muscle Nerve 2011;44:185-90.
7. Chaudhry V, Clawson LL. Entrapment of motor nerves in motor neuron disease: does double crush occur? J Neurol Neurosurg Psychiatry 1997; 23: 189-92.
8. Sitt CM, Fung LW, Yuen HY, Ahuja AT.Hirayama disease in a 17-year-old Chinese man. Singapore Med J 2014; 55: 87-9.
9. Paredes I, Esteban J, Ramos A, Gonzalez P, Rivas JJ. A severe case of Hirayama disease successfully treated by anterior cervical fusion. J Neurosurg Spine 2014; 20:191-5.