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Case Report

Beyond the Classic Distal Pattern: Proximal Upper Limb Monomelic Amyotrophy in Hirayama Disease

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Abstract

Hirayama disease (HD) is, a benign lower motor neuron disease, characterized by unilateral weakness and wasting of distal upper limb in young adults. We herein report a case of atypical presentation of HD with proximal unilateral upper limb involvement.

Keywords: *Hirayama Disease, Proximal, Monomelic Amyotrophy, Cervical Spine, Flexion*

Introduction

Hirayama disease (HD), also known as brachial monomelic amyotrophy, is characterized by weakness and wasting of distal aspect of unilateral upper limb in young adults[1]. It is a benign lower motor neuron disease affecting lower cervical myotomes. HD is more prevalent in Asian countries usually affecting males[2]. Proximal upper limb involvement in HD is rarely reported in literature[3-7]. We herein report a case of Hirayama disease with proximal involvement of upper limb.

Case Presentation

A 15-year-old boy presented with progressive weakness and atrophy of the right upper limb over the past 3 months. He noticed wasting of right upper limb in biceps brachii and deltoid muscle. He had involuntary twitching of right proximal upper limb muscles. He had weakness in flexion of right arm at elbow joint and abduction of shoulder joint from the last 3 months. There was no history of preceding surgery, vaccination, trauma or any systemic illness prior to the onset of the wasting and weakness. There was no history of similar illness in the family.

On neurological examination, the muscle power of the right elbow flexion was 4-/5 and shoulder abduction were 4/5 with associated atrophy in upper arm. The circumference of right arm was smaller than that of the left arm: 19cm versus 23cm. The intrinsic muscles of hand and forearm were normal. The biceps and brachioradialis jerk were absent. The sensory examinations were normal. Neurological examination of the other extremities was normal. There was presence of irregular tremor of outstretched right hand with wrist in extension (polyminimyoclonus).

Nerve conduction studies (NCS)showed decreased compound muscle action potential (CMAP) of right musculocutaneous and axillary nerve. Electromyography (EMG) showed increased spontaneous activities, large, polyphasic motor unit action potentials (MUAPs) with prolonged duration and reduced recruitment pattern in right deltoid and biceps muscle. EMG of the left arm, cervical paraspinal muscles and right lower limb was normal.

Magnetic resonance imaging (MRI) cervical spine at the level of C4-C5 intervertebral disc in neutral position showed anteriorly displaced posterior dura along with enlarged epidural space at the level of C3, C4 and C5 vertebra in T2 weighted images with further exaggeration of dural detachment and compression of cord on dynamic flexion. High signal intensity was seen on T2 weighted images in the upper cervical cord, accompanied by atrophy of cord and reversal of cervical spine lordosis (Figure 1). He was managed with a supportive neck brace. During follow-up at 2 months, there was neither improvement nor deterioration of motor weakness or muscle atrophy.

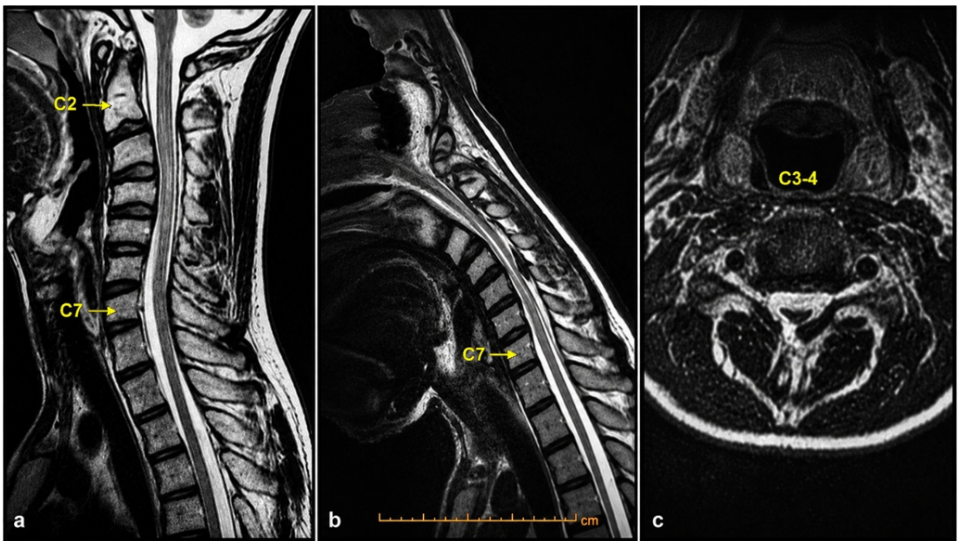

Figure 1. Sagittal T2 weighted MR image in neutral position shows anteriorly displaced posterior dura along with enlarged epidural space at the level of C3, C4 and C5 vertebra (a) and reversal of cervical spine lordosis (kyphosis). Flexion T2 weighted MR image demonstrates exaggeration of dural detachment with compression on cord and prominent flow voids in widened epidural sac. (b) High signal intensity can be seen in the upper cervical cord, accompanied by atrophy of cord. Asymmetric flattening of cord, more on the right side, along with dural detachment can be observed on axial T2 weighted MR images obtained at the level of C3-C4 vertebra (c).

Discussion and Conclusion

Hirayama disease is a benign lower motor neuron disease of only one upper limb which progress in initial years followed by a stationary course, without involving any other limbs. HD was initially named as “juvenile muscular atrophy of unilateral upper extremity” by Hirayama et al in 1959[1].It was initially thought of unilateral disease but later bilateral asymmetrical as well as bilateral symmetrical cases has also been reported[8]. Involvement of the small muscles of hand and medial forearm muscles are common leading to oblique amyotrophy. HD usually affects males but it is reported to occur in females also[9].

The etiology of HD is still not known as even the genetic studies were not able to demonstrate abnormalities[10]. The various mechanism has been proposed for pathogenesis of HD such as viral infections, rigorous physical activity, anterior horn cells ischemia, and atopy[1]. One of the most accepted mechanisms is imbalanced growth of vertebral column and contents of spinal canal leading to dural sac tightening and anterior displacement of posteriordural wall on neck flexion. The spinal cord compressed against the posterior margin of vertebrae causes necrosis of anterior horn cells and cord atrophy due to microcirculatory disturbances in the adjacent cervicalcord[1]. Our patient showed prominent posterior epidural space with myelopathic changes in cord from C3 to C5in neutral position with further prominence in flexed posture which can explain the proximal involvement of upper limb. Spectrum of imaging changes are seen on MRI cervical spine in neutral position i.e., loss of cervical lordosis and cervical kyphosis, C5–C6 cord atrophy, asymmetric flattening of cord on axial images, hyperintense signal in anterior horn cell on axial T2 images (snake-eye appearance)[9].

Electrophysiological tests are one of the essential modalities for the diagnosis of HD. In typical cases of HD there is reduction in CMAP of ulnar- abdutor digiti minimi while in our case, there was reduction in CMAP of right musculocutaneous- biceps brachii and right axillary-deltoid as there was proximal involvement. EMG shows chronic denervation changes more frequently than changes of active denervation in affected muscles[9].

There are only few reported cases of proximal upper limb involvement in HD[3-7]. The previous reported cases have showed predilection for C4 to C6 vertebral level as seen in our patient. This unusual involvement of the relatively higher level of the spinal cord at C4–C5 spinal column is postulated due to upper cervical kyphosis seen in previous reported cases[6]. Some of the previous reported cases of proximal HD showed additional involvement of distal muscle, bilateral involvement or no abnormality on cervical MRI[5-7]. Isolated unilateral involvement of upper limb with MRI changes is seen in only two previous reported cases as seen in our patient[3,4].

The management of HD mainly rests on the use of cervical collar. Surgical methods like spinal fusion surgery and laminectomy plus tuboplasty can be used in selected patients[9]. Rehabilitation in form of limb-strengthening exercises, splint support and tendon transfer help in reducing disability in later stage of the disease. The prognosis of HD is good as it progresses only for an initial period of 3–4 years. However, if left untreated, it leads to severe atrophy of involved muscles. Hence, early diagnosis and management are required to limit the degree of disability.

Conflict of Interest

The authors declare that they have no conflict of interest.

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