

Hirayama Disease: Will Physiotherapy Help?

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ABSTRACT

Hirayama disease (HD) is a rare benign neurological disorder in the young with a male preponderance and usually affecting one upper extremity. It is characterized by the insidious onset and progressive weakness and wasting of a distal extremity. Generally, this disease is considered as a benign and non progressive motor neuron disease that stabilizes within five years of onset. This case report described a 19-year-old male patient who experienced left distal upper extremity amyotrophy with no sensorial abnormality. Early diagnosis is necessary as the use of a simple cervical collar preventing neck flexion, with postural correction and strengthening of weak muscle of hand and forearm has been shown to stop the progression. Further physiotherapy protocols have been found to be helpful in good prognosis of the condition.

Keywords: Hirayama disease, monomelic amyotrophy, motor neuron disease

INTRODUCTION

Hirayama disease (HD), also known as monomelic amyotrophy (MA) or Sobue disease or juvenile muscular atrophy of distal upper extremity ^[1] (JMADUE), is a rare motor neuron disorder that affects one upper extremity. Hirayama et al. pointed out that these clinical findings do not fit any of the previously known diseases that cause degenerative and progressive muscular atrophy. ^[2,3] Later, these clinical findings were identified in detail in 38 patients by Hirayama et al. This clinical picture was called as juvenile spinal muscular atrophy or juvenile asymmetric segmental spinal muscular atrophy ^[4] or juvenile muscular atrophy of unilateral upper extremity, ^[5] or Madras pattern of motor neuron disease. ^[2,6,7] It has been mostly reported in Japan, India, Sri Lanka, Korea, Hong Kong, Taiwan, and Malaysia. ^[2,4] The number of cases reported from Western countries is very low. ^[8] Men are affected by this disease five times more often than women. ^[2] It is generally seen among young people who are in a period of rapid growth. Clinical

findings appear in the second or third decade and progress slowly. In this disease, unilateral or asymmetric bilateral muscle weakness and atrophy, which progress slowly, occur in the hands and forearm. The disease is limited to upper extremity motor neuron involvement; lower extremity, sensory, or bulbar involvement is not observed. In some cases, muscle weakness becomes evident in cold weather. Patients express that while weakness in fingers increases in cold weather, it gets normal in warm weather. Therefore, patients discover their disease for the first time during winter months. It is rarely seen with hyperhidrosis and abnormal sympathetic skin responses. In 20% of the cases, weakness of the upper arm can be seen. It displays asymmetrical and varying degrees of involvement in the thenar, hypothenar, and interosseous muscles of the hand. The cause of this disease is unknown. However, theories such as chronic spinal cord compression or atrophy have been suggested. In particular, crushed and flattened spine due to excessive forward displacement of the dural sac

during cervical spine flexion and ischemic changes resulting from this situation are thought to be responsible for spinal anterior horn damage.^[9] Further, on pre- and post-contrast neutral and flexion positioned cervical magnetic resonance imaging, localized lower cervical cord atrophy, straightening of the asymmetric cord, abnormal cervical curvature, loss of connection between the basal lamina and posterior dural sac, forward displacement of the posterior wall of the dural canal, and expansion of epidural component symptoms have been observed.^[10] Familial incidence is very low, and very few cases of autosomal recessively inherited disorders have been reported.^[1,2,7,8] This article describes the case of a 19-year-old male patient who came to Institute of Health Sciences, physiotherapy department with complaints of weakness and wasting of the left hand from past 3 years with initiation of right hand weakness from past two months.

CASE REPORT

A 19 year old male patient came to the physiotherapy department with a typical presentation of a slowly progressive weakness of left hand with an insidious onset and starting of involvement of right hand from past two months, He complaint of inability to lift heavy objects and hold a glass of water but able to do all his activities of daily living as it is his non dominant hand, further with a notable wasting near the anatomical snuff box (fig 1) and the thenar eminence region and eminent slenderness of forearm on ulna side. In his history, it was revealed that he experienced occasional numbness and chills in his left arm and that he was unable to carry any load using that hand for a long time this had started before 3 years. Nothing significant was found in his family history but he stated that in his residential origin other people too suffered from similar complaint. Physical examination findings showed minimyoclonus, brisk reflexes whereas sensations were intact. Apparent atrophy was observed in thenar, hypothenar,

interosseous muscle and forearm muscles but sparing brachioradialis (fig 2 (a)). The strength of his left hand hypothenar abductor digiti minimi and opponens digiti minimi muscles was 3/5, and that of his left hand thenar abductor pollicis brevis, opponens pollicis, flexor pollicis, adductor pollicis and left hand palmar interosseous and lumbrical muscles was 3/5. The strength of his left forearm flexor pollicis longus, flexor digitorum superficialis and profundus, abductor pollicis longus, extensor indicis, extensor pollicis brevis and longus, and extensor carpi ulnaris and radial muscles was evaluated to be 3/5. Measurements of girth of forearm showed a difference of 4 cm from the unaffected forearm. No atrophy was detected in the upper arm measurements. All values in hemogram and biochemical tests were found to be within normal limits.



Fig.1: This figure shows wasting of muscles near anatomical snuff box

MRI showed thinning of cord at C6-C7 level with separation of dura noted at C7-C8 & C8-T1 exaggerating on flexion study, pushing the cord anteriorly against the vertebra (fig 2). Loss of normal cervical lordosis was noted with early desiccation of C2-C3 and C3-C4 discs. EMG revealed left ulna axonal neuropathy with neurogenic pattern. It was detected that symptoms consistent with anterior horn cell involvement were limited in the left C7, C8,

and T1 myotomes. The patient was diagnosed with MA based on these findings. When the patient was functionally assessed, except for skills requiring advanced skills, his left hand was independent in daily life activities. A difficulty was observed in his left hand grip. The patient was advised to use cervical collar further was taught postural correction exercises of neck and to maintain the condition patient was taught with various exercise which uses hand and forearm muscles. The patient was informed that his clinical information would be used for scientific purposes, and his consent was obtained.

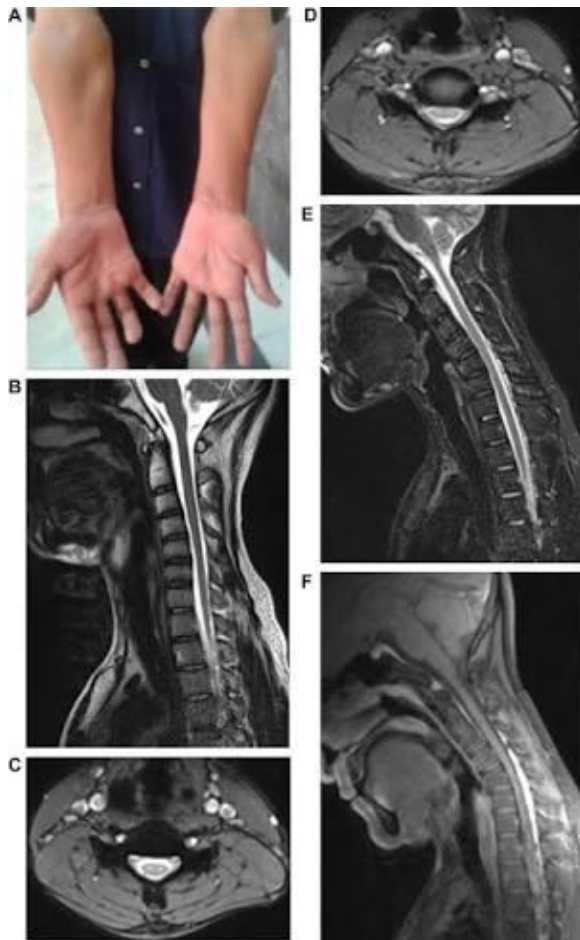


Fig.2(A): This figure shows the wasting of hypothenar muscles and slenderness of forearm on ulnar side
 Fig.2(B),(C),(D),(E),(F): MRI showed thinning of cord at C6-C7 level with separation of dura noted at C7-C8 & C8-T1 exaggerating on flexion study, pushing the cord anteriorly against the vertebra

DISCUSSION

Hirayama disease came into recognition in Japan in 1959 where it was reported as Juvenile muscular atrophy of

unilateral upper extremity. [3] In a report in 1991, Chan et al. estimated 150 cases from Japan, 37 from India, and 102 from Sri Lanka. The disorder has distinctive features of male predominance between the age of 15-25 years, asymmetric upper limb involvement and a self-limiting course. [12] There is unilateral involvement in majority but asymmetric and symmetric bilateral involvement is also observed. The weakness and atrophy predominantly involves the intrinsic hand muscles (hypothenar, thenar and interossei muscle groups) as well as the ulnar side of the forearm. There is sparing of the brachioradialis muscle giving the impression of an “oblique atrophy”. Motor deficit and atrophy may progress for 1-3 years. In the series of Kikuchi et al, there were 17 males and 1 female and the progression of symptoms arrested within 5 years. [11] They also noted some improvement in strength after arrest of progression. Sensory symptoms and signs are conspicuously absent. Deep tendon reflexes are normal in both upper and lower extremities. On pathologic examination Hirayama et al. found the lesions predominantly in the anterior horns of the spinal cord particularly marked at C-7 & C-8. [13] Since the pathogenesis clearly understood, probable causes suggest imbalanced growth between the patients’ vertebral column and spinal cord causing disproportional length between the vertebral column and the spinal canal contents. Hence, a “tight dural sac” in the neutral position and an anteriorly displaced posterior dural wall when the neck is flexed. In neck extension, the dura matter of the cervical spine is slack and thrown into transverse folds while in neck flexion, the dura becomes tighter because the length of the cervical canal increases as the neck moves from extension to flexion. Normally, the slack of the dura can compensate for the increased length in flexion and so the dura can still be in close contact with the walls of the spinal canal without anterior displacement. In Hirayama disease, the dural canal isn’t slack in extension due to

imbalance in growth of vertebrae and dura matter and therefore, the tight dural canal cannot compensate for the increased length of the posterior wall during flexion. This leads to anterior shifting of the posterior dural wall, consequently, compressing the cord which may lead to microcirculatory disturbances in the anterior spinal artery or in the anterior portion of the cord. The chronic circulatory disturbance resulting from repeated or sustained flexion of the neck may produce necrosis of the anterior horns which are most vulnerable to ischaemia. Conventional X-rays in Hirayama disease shows no abnormality except loss of the normal lordosis. MRI studies with neck in flexion are easy to obtain and show forward displacement of the posterior wall and a well enhanced crescent-shaped mass in the posterior epidural space of the lower cervical canal representing the congestion of the posterior internal venous plexus which vanishes once the neck returns to neutral position. MRI shows atrophy of the lower cervical cord in a neutral position with abnormal cervical curvature and loss of attachment between the posterior dural sac subject lamina, significant in Hirayama disease.

Hirayama disease is a self-limiting disorder and there is no consensus on the definitive treatment. However, early diagnosis is necessary because a cervical collar may arrest the progression of the disorder by limiting the neck flexion. Physiotherapy is also helpful in preventing complications resulting from immobility such as joint stiffness and muscle wasting. [14]

CONCLUSION

Hirayama disease should be considered in the differential diagnosis of young men with forearm and hand muscle atrophy. As early diagnosis is crucially important for the treatment of suspected patients, dynamic flexion MRI should be performed. With the help of early treatment modalities, use of cervical collar, postural correction and exercises of hand and

forearm muscles the prognosis of the can be better off even helping to stop progression.

Informed Consent: Informed consent was obtained from patient who participated in this case.

Conflict of Interest: No conflict of interest was declared by the author.

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