Case Report

A case of Hirayama Disease in Pakistan

Mahvish Zahid, Mehmooda Wasim, Yasir A. Ghaffar, Naila Naeem Shahbaz, Muhammad Abdullah, Ismail A. Khatri
Department of Neurology, Dow University of Health Sciences and Civil Hospital, Karachi.

Abstract

We report a case of Hirayama disease in a 23 year old male presenting with weakness of right hand and forearm. The weakness started gradually, progressed over a period of 12 months and stabilized. Few months after the onset of right-sided weakness, he noticed weakness in the left hand and forearm which increased over a period of 3 months before stabilizing. Neurological examination demonstrated moderate to severe atrophy of distal upper extremity muscles, preserved reflexes and normal sensory findings. Electrophysiological studies revealed neurogenic changes in the muscles innervated by lower cervical spinal cord. Magnetic resonance imaging showed atrophy of mid-cervical cord with high signal intensity in the anterior horn cell region. These examination and investigation findings were compatible with the diagnosis of juvenile muscular atrophy of distal upper extremities (JMADUE) also known as Hirayama disease.

Introduction

Hirayama disease is a rare condition.[1] It is also known as juvenile muscular atrophy of distal upper extremities (JMADUE), monomelic amyotrophy, benign focal amyotrophy, juvenile asymmetric segmental spinal muscular atrophy and juvenile muscular atrophy of unilateral upper extremity.[2] Hirayama et al. described this condition in 1959. Several cases have been reported mostly from Asian countries.[3] We report the case of a 23 year old man with weakness of distal upper extremities which progressed for several months before stabilizing. His clinical, electrophysiological and imaging findings were consistent with Hirayama disease. To our knowledge there is no previous published report of Hirayama disease from Pakistan.

Case Report

A twenty three year-old right-handed Pakistani man developed weakness and wasting of the right hand and forearm. The symptoms started at age of 21 and worsened over approximately 1 year. Weakness and wasting of left hand started several months later, which progressed for 3 months. He noticed weakness while carrying cement bags, and later developed difficulty in handling small utensils and handwriting. He had no sensory symptoms. There was no preceding illness, trauma, exposure to toxins or family history.

Clinical examination revealed moderate to severe atrophy of intrinsic hand and forearm muscles except for brachioradialis as seen in oblique atrophy of Hirayama disease (Figure 1a and 1b). The wrist flexors, finger abductors, and finger extensors were the weakest muscles. The right hand was weaker. Deep tendon reflexes were normal except slight hyporeflexia in triceps, the knee jerks were brisk bilaterally. Plantars were flexors. Mild action tremor was seen in fingers. Higher mental functions, gait, cranial nerves, cerebellar functions, and sensory examination were normal.

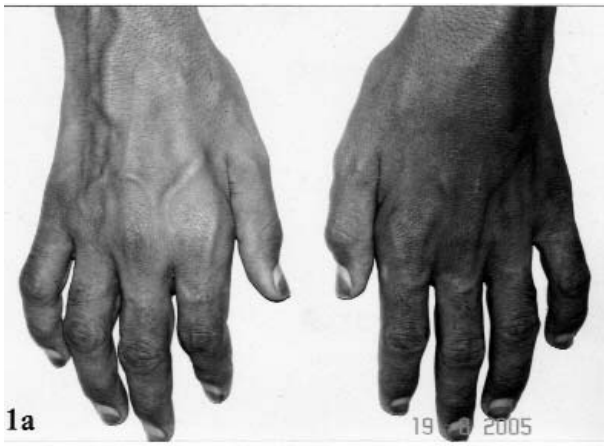

Figure 1a.

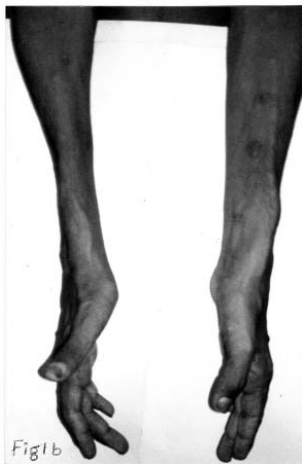

Figure 1b.

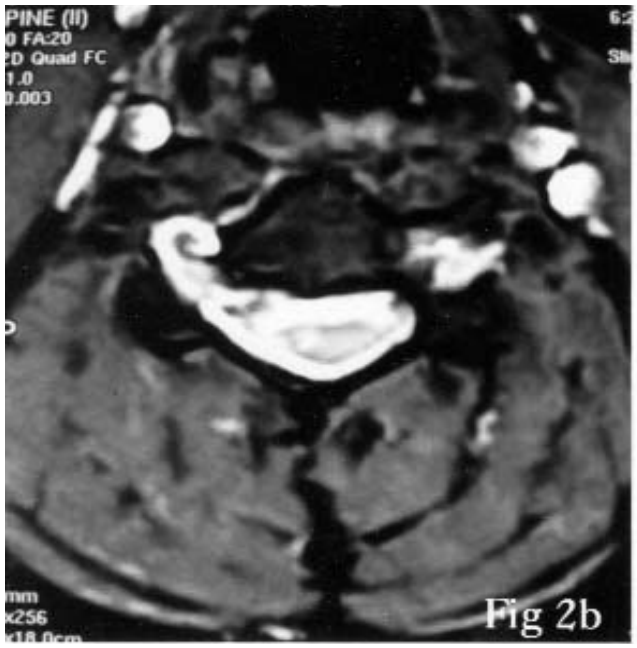

Figure 2a.

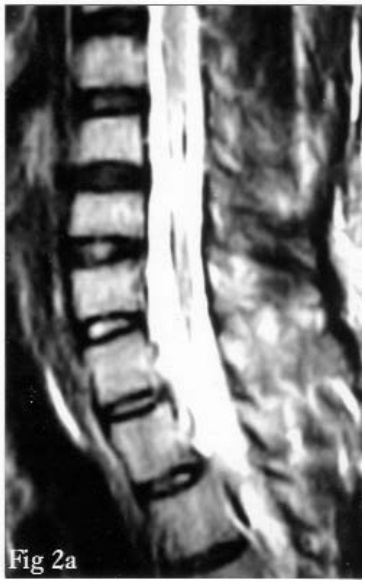

Figure 2b.

The haematological investigations were normal. Cerebrospinal fluid showed glucose - 51 mg/dl, proteins - 15 mg/dl, and white cells - 5/mm^3. Electrophysiologic studies showed decreased amplitude in ulnar nerves with normal latencies and borderline low velocities; giant motor unit action potentials were seen in the muscles innervated by C7, C8 and T1 nerve roots, with fibrillations and positive waves. The magnetic resonance imaging (MRI) of the cervical spine showed high signal intensity in mid-cervical cord at

C5-C6 (Figure 2a). The axial sections showed anteroposterior flattening of the cord with intrinsic signal changes. (Figure 2b) Clinical, radiological and electrophysiologic findings in our patient were consistent with the diagnosis of Hirayama disease.

The patient was advised to wear a soft cervical collar. On three month follow up the patient had shown no progression of his disease. The role of surgery is debatable and as the progression had stopped, he was not offered surgical intervention.

Discussion

Hirayama disease was initially recognized in Japan in 1959 and reported under the name of juvenile muscular atrophy of unilateral upper extremity.[3] Since then similar cases have been described from many countries, mostly from Asia. In a report in 1991, Chan et al. estimated 150 cases from Japan, 37 from India, and 102 from Sri Lanka.[4] There are reports from several other countries including Singapore, Taiwan, Hong Kong, Nigeria, Denmark, Holland, USA, Austria, UK, France, Canada, Poland and Germany.[3,5] Our patient is to our knowledge, the first report from Pakistan.

The disorder has distinctive features of male predominance, asymmetric involvement of upper extremities and self limiting course.[4] The atrophy and weakness predominantly involves the intrinsic hand muscles (interossei, thenar, and hypothenar muscle groups) as well as the ulnar side of the forearm.[1,3] There is sparing of brachioradialis muscle giving the impression of an 'oblique atrophy'.[1,2,6] There is unilateral involvement in majority, but asymmetric and symmetric bilateral involvement is also observed.[2] In the 18 cases of Kikuchi et al.[3] 71% patients had unilateral and 29% had bilateral involvement. There is no relationship between the patient's handedness and side of greater muscular atrophy.[3,5] Deep tendon reflexes in upper extremities are normal, however, hypoactive triceps reflex and mild hyperreflexia in legs have been reported.[6] Sensory symptoms and signs are conspicuously absent.[5,7] A distal irregular jerky tremor (minipolymyoclonus) is also seen in some patients.[1,5,6] Another interesting phenomenon is exacerbation of weakness in a cold environment.[3,6,8] Our patient also had sparing of brachioradialis muscles, mild hyporeflexia in triceps muscles, slightly brisk knee jerks, preservation of sensations and mild action tremor.

Clinically, these symptoms are seen in 15 to 25 years old males with insidious onset and unilateral muscular atrophy. Motor deficit and atrophy may progress for 1 to 3 years and has self limiting course.[7] In the series of Kikuchi et al,[3] there were 17 males and 1 female and the progression of symptoms arrested within 5 years. They also noted some improvement in strength after arrest of progression.

The nerve conduction studies frequently demonstrate low-amplitude compound muscle action potentials (CMAPs) commensurate with degree of weakness and atrophy.[3,8] The electromyographic (EMG) findings usually show loss of motor unit potentials (MUPs) which are rapidly firing, high amplitude, and polyphasic, as seen in anterior horn cell disorders.[1,3,5] These changes are localized to lower cervical and T1 myotomes. Fibrillation potentials and sharp waves may be seen.[6] Our patient had low amplitude CMAPs with giant MUPs and active denervation.

Conventional radiographic studies of the cervical spine show no specific abnormalities. Myelography can show forward movement of posterior dural wall when neck is flexed; the examination is difficult to perform as contrast medium is not easily retained in the flexed cervical subarachnoid space.[9]

The pathogenesis is debated. Dynamic spinal cord compression due to neck flexion with forward displacement of posterior dura is considered as the primary mechanism.[2] When neck is flexed, the cervical cord is longitudinally stretched. In Hirayama disease, the lower cervical cord moves forward in flexion and contacts the posterior surface of the vertebrae; the lower cervical cord becomes flattened at contact point.[3] Additionally, the posterior wall of the dural tube moves forward, the posterior epidural space expands forming a crescent shaped mass in the posterior epidural space. This mass is formed by the congestion of posterior internal vertebral venous plexus.[3,9] In a report of 73 patients, Hirayama and Tokumaru concluded that dynamic cord compression in flexion with forward displacement of posterior dura is an unequivocal finding in progressive stage. The mechanism of myleopathy may involve ischemic changes or chronic trauma by repeated neck flexion affecting anterior horn cells along with spinal cord thinning, termed as flexion myelopathy.[2]

The neutral-position MRI may show lower cord atrophy, asymmetric cord flattening, noncompressed intramedullary high signal intensity on T2-weighted imaging and abnormal cervical curvature (straight or kyphotic).[7] The MRI of our patient also showed anteroposterior cord flattening and intramedullary high signal. Cervical MR studies in neck flexion can show anterior displacement of posterior wall, a well enhanced crescent shaped mass in the posterior epidural space.[9] Although flexion myelopathy has been proposed as the underlying mechanism, cases of JMADUE without evidence of flexion myelopathy on MRI have been reported. Moreover, even on flexion MRI not all cases necessarily show forward displacement of dural sac.[6,10]

There is no definitive treatment of this condition. The primary principle of treatment is restriction of neck flexion. Posture with long-term neck flexion must be avoided. Low pillows are recommended. A neck collar is recommended for an average of 3 to 4 years.[3,9] Anterior fusion of cervical vertebrae and duraplasty, with or without anterior fusion has been performed. The indications and methods of surgical treatment remain controversial.[3]

In young males with distal upper extremity weakness, atrophy, preserved reflexes and normal sensory examination, Hirayama disease should be considered. Initial unilateral involvement with or without later bilateral affection, unilateral or bilateral tremor, absence of cerebellar and sphincter involvement, and EMG findings of anterior horn cell disease further support the diagnosis. An MRI of cervical spine in neutral position and flexion should be obtained. Syringomyelia, spinal cord tumors, poliomyelitis, multifocal motor neuropathy, and toxic neuropathies should be excluded.

References

1. Nascimento OJ, Freitas MR. Non-progressive juvenile spinal muscular atrophy of the distal upper limb (Hirayama's disease): a clinical variant of the benign monomelic amyotrophy. Arq Neuropsiquiatr 2000; 58:814-9
2. Gandhi D, Goyal M, Bourque PR, Jain R. Case 68: Hirayama disease. Radiology 2004; 230:692-6
3. Kikuchi S, Tashiro K. Juvenile muscular atrophy of distal upper extremity (Hirayama disease). In Jones HR Jr., De Vivo DC, Darras BT (eds), Neuromuscular Disorders of Infancy, Childhood and Adolescence-A clinician's approach. Butterworth-Heinemann; UK 2003; pp 167-81.
4. Chan YW, Kay R, Schwartz MS: Juvenile distal spinal muscular atrophy of upper extremities in Chinese males: A single fiber electromyographic study of arms and legs. J Neurol Neurosurg Psychiatry 1991; 54:165-16
5. Peiris JB, Seneviratne KN, Wickremashighe HR, Gunatilake SB, Gamage R. Nonfamilial juvenile distal spinal muscular atrophy of upper extremity. J Neurol Neurosurg Psychiatry 1989; 52:314-9.
6. Ochi H, Murai H, Osoegawa M, Minohara M, Inaba S, Kira J. Juvenile muscular atrophy of distal upper extremity associated with airway allergy: two cases successfully treated by plasma exchange. J Neurol Sci 2003; 206:109-14.
7. Chiba S, Yonekura K, Nonaka M, Imai J, Matumoto H, Wada T, et al. Advanced Hirayama disease with successful improvement of activities of daily living by operative reconstruction. Intern Med 2004; 43:79-81
8. Chen CJ, Hsu HL,Tseng YC, Lyu RK, Chen CM, Huang YC, Wang LJ, et al. Hirayama flexion myelopathy: neutral position MR imaging findings-importance of loss of attachment. Radiology 2004; 231:39-44
9. Chen CJ, Chen CM, Wu CL, Ro LS, Chen ST, Lee TH. Hirayama disease: MR diagnosis. AJNR 1998; 19:365-8.
10. Willeit J, Kiechl S, Kiechl-Kohlendorfer U, Golaszewski S, Peer S, Poewe W. Juvenile asymmetric segmental spinal muscular atrophy (Hirayama's disease): three cases without evidence of 'flexion myelopathy'. Acta Neurol Scand 2001; 104:320-2.