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Case Report

Hirayama Disease with Rare Presentation in a Young Adult

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I N F O

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A B S T R A C T

Hirayama disease or Monomelic Amyotrophy (MMA) is a type of rare cervical myelopathy which is an asymmetrical, self-limited with progressive atrophy and weakness of the muscles of hands and forearms. It is mainly found in males of young age. There is occurrence of forward dislocation in the posterior dura of the lower cervical Dural canal at the time of flexion of the neck which causes lower cervical cord atrophy along with asymmetric flattening. Here, we report a rare case of this disease in a 21-year-old male who presented with slowly progressive asymmetrical weakness and atrophy of muscles of both hands and forearms associated with unusual characteristics of autonomic disturbances and lesion of upper motor neuron.

Keywords: Atrophy, Cervical Myelopathy, Hirayama Disease, Young Males

Introduction

Hirayama disease, is a neurological entity with rare occurrence. It is characterised by sporadic juvenile muscular atrophy of the distal part of upper limbs due to involvement of the lower cervical cord regions. It develops at the end of teen age and early of twenties predominantly in the male sex. It is mainly characterised by insidious onset and slowly progressing unilateral or bilateral atrophy of muscles along with weakness of the hands and forearms. Presence of sensory abnormality, autonomic dysfunction, and Upper Motor Neuron (UMN) signs such as hyperreflexia and hypertonia are rarely found.[1] Differential diagnosis of Hirayama disease is Motor Neuron Disease (MND). Unlike MND, the disease progresses in beginning and is then spontaneously arrested for long time. Japan and other Asian countries are highly prevalent for Hirayama disease. This case is reported from India.

Case Presentation

A 21-year-old Indian male presented in Medicine outdoor with chief complaints of slowly progressive wasting and distal weakness of right hand and forearm for 6 years. After a period of one and half year, wasting and weakness also involved the left hand. This leads to limitations of several daily activities. There was also tremulousness of hands for the past one and half years. Tremulousness slowly progressed to involve both lower limbs after 3 months. It was present at rest and on an intention to do something. Daily activities such as writing, mixing the food, buttoning and unbuttoning the cloths, clutch and brake application in bike and picking objects hampered grossly. Similar complain

were also noted in left hand. There was also complaint of excessive sweating in the palms of both hands. History of sensory involvement, neck pain, dysphagia, difficulty in walking, diplopia, bowel and bladder involvement was absent. There was no history of trauma, toxin exposure, or drug intake in past. No any similar complaint was found in any member of family.

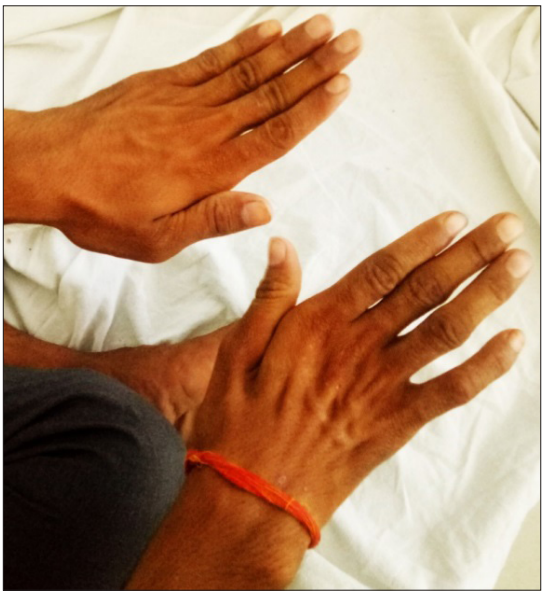

Figure 1.Atrophy of bilateral hands

Figure 2.Atrophy of the right forearm muscles and sparing of brachioradialis muscle

On general examination patient was conscious and oriented, with normal vitals. On neurological examination there were Upper Motor Neuron (UMN) and Lower Motor Neuron (LMN) signs present. It was found that bilateral forearms and hands were atrophied and weak. Brachioradialis muscles were normal (Figures 1 and 2).

Impairment of full abduction, palmar grasps, opposition of the thumbs, adduction of the digits was found. In both the hands tremor and sweating were also noted. Orthostatic hypotension was absent. Quadriceps and calf muscles of both lower limbs were having minipolymyoclonus, hypertonia, and sustained bilateral ankle clonus was also noted. Bilateral plantar reflexes were exaggerated. In all four limbs power of the proximal muscles was normal. There was no Evidence of dysfunction of cerebellum, posterior column, or cranial nerves. All other systems were within normal limits.

Compound Muscle Action Potential (CMAP) amplitude was decreased in right median and ulnar nerve in Nerve Conduction Velocity (NCV) test. It was possibly because of severe wasting of the tested muscle. Sensory system was intact. There was incomplete recruitment pattern in Electromyography (EMG) without any sign of fibrillation, fasciculation, positive sharp wave, and no spontaneous insertion activity. All cerebellar signs were absent. Action potential of motor unit was found to be raised along with increase in time duration which revealed neurogenic concern. We were not able to do the autonomic sweat test due to non-availability of this test. Lab parameters such as complete blood count, Erythrocyte Sedimentation Rate (ESR), thyroid profile, kidney function tests, creatine kinase, Liver Function Test (LFT), vitamin D3 level and vitamin B12 all were found normal. Antinuclear Antibody (ANA), extractable nuclear antigens, rheumatoid factor, and Antiphospholipid Antibody (APLA) were negative which rules out vasculitis syndrome. Human Immunodeficiency Virus (HIV), hepatitis B, and hepatitis C were also found negative.

Multi planar brain and cervical spine Magnetic Resonance Imaging (MRI) in neck flexion was done on 3-tesla magnet system using dedicated CP array head coils. The MRI study revealed symmetrical atrophy and thinning with flattening

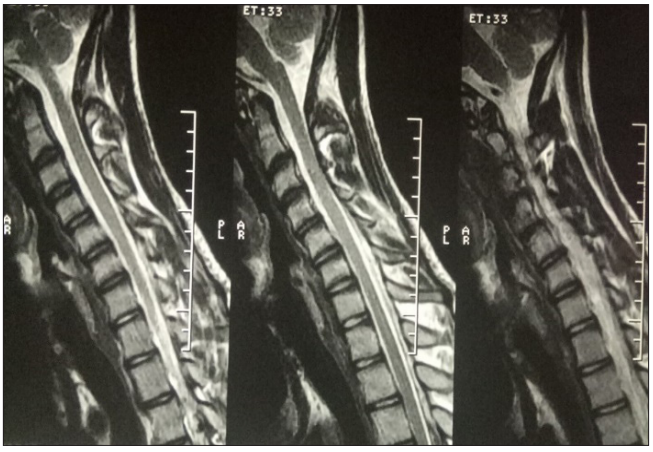

Figure 3.MRI neck showing anterior migration of posterior dura during flexion study along with enlarged posterior epidural space showing flow voids

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of cervical cord at C6 and C7 vertebral level. Subtle T2/ STIR hyperintensities noted in the cord at the level. There was anterior migration of posterior dura during flexion study with enlarged posterior epidural space showing flow voids. Loss of cervical lordosis also noted. Brain scanning was normal in structure and signal pattern.

After detailed history, local examinations, NCV and MR imaging findings we reached at the diagnostic point of Hirayama disease. The patient was advised for cervical collar to prevent neck flexion, and reducing further spinal cord injury.

Discussion

Hirayama disease was first diagnosed by Hirayama K et al. in Japan in 1959. It was called by name "juvenile muscular atrophy of unilateral upper extremity".[2] It was also described by several other names like "juvenile muscular atrophy of the distal upper extremity, juvenile asymmetric segmental spinal muscular atrophy, and benign focal amyotrophy or monomelic amyotrophy".[3] HD is featured by insidious start of asymmetrical weakness and atrophy of muscles of hands, involving mainly C7, C8, and T1 myotomes predominantly in males with age of 15 to 25 years. At the initial level HD disease progresses for 1-3 years and after that no progression occurs, which shows its benign course. Irregular coarse tremor (minipolymyoclonus) of the digits of involved limb which increases after cold exposure is another feature of HD. No abnormality is noted in the sensory, reflex, and cranial nerves. Pyramidal tract involvement in lower limb, autonomic dysfunctions, and cerebellar involvement are rarely found. Finding of chronic denervation is there in EMG of involved muscles. However, apparently healthy muscles may be having abnormal EMG results.[4]

Tashiro K et al.[5] defined diagnostic criteria of HD as : (1) Distal predominant muscle weakness and atrophy in forearm and hand (2) Involvement of the unilateral upper extremity almost always all the time (3) Onset between the ages of 10 to early 20s (4) Insidious onset with gradual progression for the first several years, followed by stabilization (5) No lower extremity involvement (6) No sensory disturbance and tendon reflex abnormalities (7) Exclusion of other diseases like motor neuron disease, multifocal motor neuropathy, brachial plexopathy, spinal cord tumours, syringomyelia, cervical vertebral abnormalities, anterior interosseous, or deep ulnar neuropathy. Beyond these features, many authors say about sparing of brachioradialis muscle, which gives the impression of an "oblique atrophy".[6]

In this case most of the features defined by Tashiro K et al., are found present, there was also presence of excessive sweating of both palms (which reveal autonomic disturbances) and Babinski's sign (which suggests UMN lesion). Presence of these autonomic dysfunctions and UMN lesions are rarely found in HD. Autonomic disturbances was found in 36% and 46% of the patients in the case series by Hassan KM et al.[7] and Gourie-Devi M et al.[8], respectively. In the same way, UMN lesion were reported in 18% and 12% of the patients observed by Hassan et al.[7] and Sonwalkar H et al.[9], respectively.

The main cause and pathogenesis describing HD is still far away. In a research by Hirayama K et al.[10] reported degeneration of nerve cells, cell shrinkage and necrosis, mild gliosis, and ischemia in the anterior horns of the spinal cord mainly at the C7 and C8 levels. HD also occurs as a result of atopy and increased serum IgE level said by some authors.[11] Kikuchi S et al. gave widely accepted hypothesis explaining HD as a cervical myelopathy along with neck flexion.[12] Repeated neck flexion causes several episodes of ischaemia and chronic injury to the spinal cord, leading to myelopathy. Pathophysiology of presence of UMN signs, was explained by Kao Y et al. as different distribution of stress in the cervical cord.[13]

Differential Diagnosis

It includes Amyotrophic Lateral Sclerosis (ALS), multifocal motor neuropathy with conduction block, distal form of spinal muscular atrophy, post-polio syndrome, and toxic neuropathy as well as syringomyelia. All of them have their different characteristic clinical, radiological, and electrophysiological features.[2]

Typical characteristic features and dynamic MRI study in neck flexion is the main point of diagnosis. While in neutral position of neck we can find loss of attachment between the posterior Dural sac and subjacent lamina, asymmetrical cord flattening, localised lower cervical cord wasting and non-compressed intramedullary high T2 signal intensity in MRI.

HD is a self-limiting neurological entity and there is no consensus on its definitive management, but early diagnosis may prevent progression of the disease by advising for cervical collar which hampers neck flexion. Regular physiotherapy may restrict complications because of decreased movement due to joint stiffness and muscle atrophy.[1]

Conclusion

This case report is rare because of presence of autonomic dysfunction and UMN signs. Young male patient presenting with weakness and wasting of muscles of the hand and forearm should be kept in mind for HD. Early diagnosis may prevent further progression of the disease by prescribing cervical collar.

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Conflict of Interest: None

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