Hirayama's Disease – A Rare Case Report with Review of Literature

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What to Learn from this Article?

Presentation and Diagnosis of Focal Amyotrohic (Hirayama's Disease).

Abstract

Introduction: Hirayama's disease is a rare benign disorder, also referred to as monomelic amyotrophy (MMA), Juvenile non progressive amyotrophy, Sobue disease. It is a focal, lower motor neuron type of disease. Mainly young males in their second and third decades of age are most commonly affected. It is seen most commonly in Asian countries like India and Japan. In majority of people cause of this disease is unknown. MRI of cervical spine in flexion will reveal the cardinal features of Hirayama disease.

Case Report: A 22 year gentleman came with a history of insidious onset of weakness in both the hands begenning with left side followed by right of 4 years duration. On examination he had clawing of both hands with wasting of forearm muscles. Lower limbs had no abnormality with normal deep tendon reflexes. MRI showed thinning of cord from C4 to C7 level suggestive of cord atrophy. Based on these features a diagnosis of focal amyotrophy was made. A cervical collar was prescribed and patient is under regular follow up.

Conclusion: Hirayama disease is a rare self-limiting disease. Early diagnosis is necessary as the use of a simple cervical collar which will prevent neck flexion, has been shown to stop the progression

Keywords: Hirayama's disease, monomelic amyotrophy, Juvenile non-progressive amyotrophy, Sobue disease.

Author's Photo Gallery

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Introduction

Hirayama's disease is a very rare benign disorder, also referred to as monomelic amyotrophy (MMA), Juvenile non progressive amyotrophy, Sobue disease. It is an untreatable, focal, lower motor neuron type of disease. The benign nature of MMA helps to distinguish it from other lower motor neuron disorders like amyotrophic lateral sclerosis (ALS). Mainly young males in their second and third decades of age are most commonly affected. It is seen most commonly in Asian countries like India and Japan [1,2]. The onset of this disorder corresponds to the beginning of the adolescent growth spurt [3]. Hirayama's disease is characterised by gradual onset of muscular dystrophy in distal part of the upper limbs related to flexion movements of the cervical spine [4,6-10]. The pathogenetic mechanism in Hirayama's disease is due to forward displacement of the posterior wall of the lower cervical dural canal when the neck is in flexion, which causes marked, often asymmetric, flattening of the lower cervical cord [4, 10–13]. The muscular atrophy associated with Hirayama's disease reaches a plateau phase between two to five years after the onset of disease. After this period of time, this disease neither improves nor worsens. People with Hirayama's disease do not experience pain or loss of sensation because of the disease. The cause of this disease is unknown in majority of people. MRI cervical spine in flexion will reveal the

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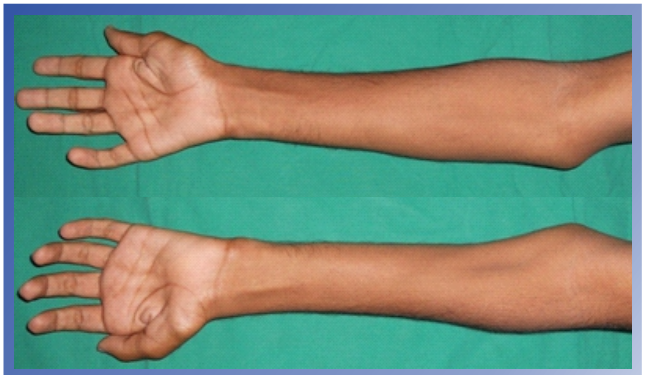

Fig 1: Both forearms showing wasting of muscles

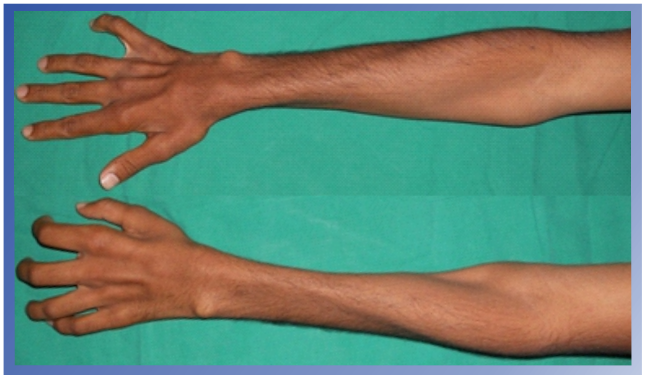

Fig 2: Both forearms showing wasting of muscles and sparing of Brachioradialis

cardinal features of Hirayama disease [3,]

We report a case of Hirayama's disease involving both the upper limbs and describe the characteristic MR imaging findings and also discuss the mechanism behind its characteristic appearance.

Case Report

A 22 year gentleman came with a history of insidious onset of weakness in both the hands, left side followed by right of 4 years duration. He noticed weakness in the left hand muscles which was gradually progressed to the fore arm muscles [Fig - 1]. Within 6 months he noticed similar complaints in the right hand also, which was progressed gradually to the fore arm muscles. He had also noticed atrophy of muscles of hand and fore arm which was gradually progressive in nature. He did not have any pain, loss of sensation, diplopia, dysphagia, ptosis, muscle cramps, fasciculations, headache or neck pain. There was no history of trauma, febrile illness, poliomyelitis or exposure to toxins or heavy metals in the past. There was no family history of similar complaints or neuromuscular disease.

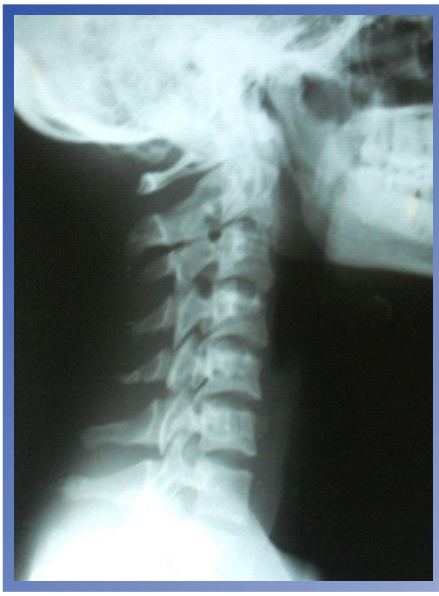

Fig 3: X ray Cervical Spine showing loss of lordosis

On physical examination he was conscious oriented to time, place and person. His cranial nerves and sensory examination was normal. Motor examination showed bilateral complete clawing of fingers with gross atrophy and weakness of thenar, hypothenar, inter osseous, fore arm muscles, biceps, triceps and deltoid muscle of both upper limbs except Brachioradialis, which was spared on both the sides [Fig - 2]. Deep tendon reflexes were not exaggerated. Co ordination and gait were normal.

Blood routine investigations, blood urea, serum creatinine, serum electrolytes, liver function tests, thyroid function tests, erythrocyte sedimentation rate and creatine phosphokinase were within normal limits. Plain cervical spine X - ray was normal except for loss of cervical lordosis [Fig - 3]. Electromyogram showed evidence of denervation in the form of fibrillation and fasciculations in C7 and C8 distribution in both upper limb muscles. Left side was more predominant compared to right. Both Brachioradialis were spared. There was no involvement of the lower limb muscles. Magnetic resonance imaging (MRI) of the cervical spine in neutral position showed thinning of cervical cord from C4 to C7 level, suggestive of cord atrophy [Fig - 4]. There was mild irregularity of the cord at C5-C6 level. Anteroposterior diameter of cervical cord at C5 was 5mm, at C6 was 4.1 mm and at C7 was 5.5 mm. On forward flexion of the cervical spine, there was anterior displacement of the posterior dura from C4 to T1 levels with maximal shift at C6 and C7 [Fig - 5]. There was no evidence of ligamentum flavum hypertrophy or inter vertebral disc prolapse. The facetal joints, paravertibral soft tissues and cranio – cervical junction were normal. The intervertebral foramina showed normal size and contour.

Based on the clinical features and the characteristic findings on flexion MR images diagnosis of Hirayama disease was made. Patient was put on a hard cervical collar to prevent neck flexion and followed up clinically at regular intervals. At the end of 12-month follow-up the

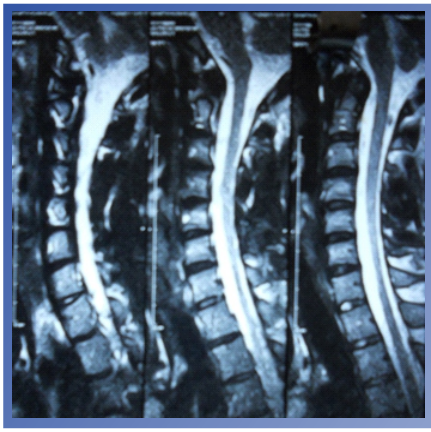

Fig 4: MRI C spine Neutral position T2 images showing thinning of cervical cord from C4 to C7 level, suggestive of cord atrophy

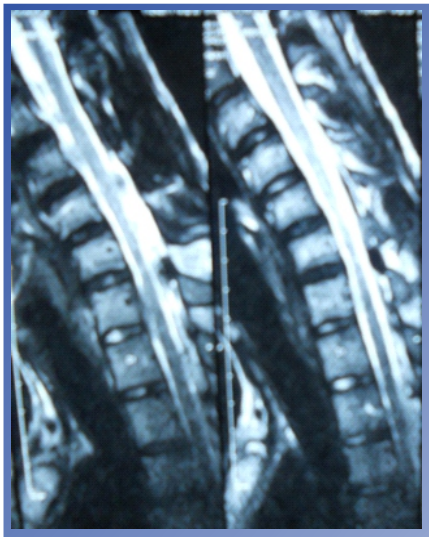

Fig 5: MRI C spine Flexion T2 images showing anterior displacement of the posterior dura from C4 to T1 levels with maximal shift at C6 and C7

patient was doing well, with no further progression of symptoms.

Discussion

Hirayama disease is characterised by focal amyotrophy with unilateral or asymmetric bilateral weakness and wasting of muscles innervated by C7, C8, and T1. It's an insidious onset, chronic, often self-limiting disorder with male preponderance, seen between the ages of 15 and 25 years [4,5-7]. Hirayama et. al first reported this disease in the year 1959 [5], but pathologic study was not done till 1982 [7], because of its benign course. At pathologic examination, these authors found the lesions only in the anterior horns of the spinal cord from C-5 to T-1, particularly marked at C-7 and C-8 [7]. Most commonly seen in Japan and other Asian countries like India and Malaysia [7]. The pathogenesis of the disorder is unknown – probable causes suggest that an imbalanced growth between the patient's vertebral column and spinal canal contents. This imbalanced growth will cause disproportional length between the patient's vertebral column and the spinal canal contents, which will cause a "tight dural sac" or "overstretch of the cord" in the neutral position and an anteriorly displaced posterior dural wall when the neck is flexed [9]. Current neuroradiologic techniques are able to show forward displacement of the posterior wall of the lower cervical dural canal in neck flexion, which is presumed to be a primary pathogenetic mechanism of Hirayama disease [9].

In neck extension, the dura mater of the cervical spine is slack and thrown into transverse folds [13]. In neck flexion, the dura becomes tighter, because the length of the cervical canal increases as the neck moves from extension to flexion. The difference in length between extension and flexion from T-1 to the top of the atlas is 1.5 cm at the anterior wall and 5 cm at the posterior wall [13]. Normally, the slack of the dura can compensate for the increased length in flexion; therefore, the dura can still be in close contact with the walls of the spinal canal without anterior displacement. In Hirayama disease, the dural canal is no longer slack in extension, because of an imbalance in growth of the vertebrae and the dura mater. Therefore, a tight dural canal is formed, which cannot compensate for the increased length of the posterior wall during flexion. This causes an anterior shifting of the posterior dural wall, with consequent compression of the cord. This compression may cause microcirculatory disturbances in the territory of the anterior spinal artery or in the anterior portion of the spinal 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 ischemia [4]. In patients with Hirayama disease, conventional X ray of the cervical spine usually show no specific abnormalities except straight alignment or scoliosis [11]. Myelography may show the forward movement of the posterior dural wall when the neck is flexed [11]; however, myelography is difficult to perform, as it is difficult to retain the contrast medium in the cervical subarachnoid space when the neck is flexed, regardless of the patient's position. MRI studies in neck flexion, which are easy to obtain, will show the forward displacement of the posterior wall and a well-enhanced crescent-shaped mass in the posterior epidural space of the lower cervical canal [11]. This mass is thought to represent congestion of the posterior internal vertebral venous plexus rather than vascular malformations or tumors, because it vanishes once the neck returns to a neutral position [9]. MRI shows atrophy of the lower cervical cord in a neutral position and there will be abnormal cervical curvature (straight or kyphotic) and loss of attachment between the posterior dural sac and subjacent lamina, which is a most valuable in Hirayama disease.

Conclusion

Even though Hirayama disease is a rare self-limiting disease, early diagnosis is necessary. Use of a simple cervical collar to prevent neck flexion, has been shown to halt the progression of the disease. Diagnosis of Hirayama disease is mainly based on flexion MRI of cervical spine. Asymmetry is one of the most characteristic findings of this disease, both clinically and radiologically. Thus, in cases of adolescent onset slowly progressive distal upper limb weakness followed by stabilization, with neurogenic changes in the EMG and the findings of asymmetric cord atrophy on regular nonflexion cervical spine MRI studies, especially at the lower cervical cord, one should keep in mind the diagnosis of Hirayama disease. When this sign is seen, a flexion MRI study should be performed to confirm the diagnosis.

Clinical Message

Hirayama disease is a very rare self-limiting clinical condition associated with weakness in the upper limb muscles. Early diagnosis is necessary as it is seen in younger people in their second and third decades. It is diagnosed only on flexion MRI of the cervical spine. It may be missed in many occasions as we usually advise just cervical spine MRI without mentioning flexion MRI. Use of a simple cervical collar to prevent neck flexion, has been shown to stop the further progression of the disease.

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