

Case Report

Rare case of Hirayama's disease

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ABSTRACT

Hirayama's disease is a rare benign neurological disorder also known as monomelic amyotrophy, Sobue disease, Juvenile Muscular Atrophy of Distal Upper Extremity (JMADUE). It mainly affects young males in their second or third decades and is most commonly seen in Asian countries like Japan, Malaysia and India. In majority of the cases the cause of the disease is unknown. An 18 year male came with weakness in his right hand and forearm since 1 year. Examination revealed weakness and wasting of muscles of forearm and hand without lower limb involvement and normal deep tendon reflexes. MRI showed focal short segment hyperintense signal in the ventral and right lateral aspect of the cervical cord at C5-C6 level with the involved segment measuring 4x3mm in size. Based on clinical and radiological features a diagnosis of focal amyotrophy was made. Patient is given a cervical collar to prevent flexion at the neck and physiotherapy in the form of hand and forearm exercises were started. Regular follow up of the patient once every 2 months is being done. Hirayama's disease is a rare, benign, self-limiting neurological disorder. Early diagnosis and management by preventing cervical flexion with the help of a cervical collar has shown to halt the progression of the disease.

Keywords: Hirayama's disease, Monomelic amyotrophy, Juvenile Muscular atrophy of distal upper extremity

INTRODUCTION

Hirayama's disease is a rare benign neurological disorder. It is also referred to as Monomelic amyotrophy and Juvenile muscular atrophy of distal upper extremities. This disease was first described by Hirayama et al. in 1959. The disease is usually focal lower motor neuron type of disease and due its benign nature it can be distinguished from other lower motor neuron disorders like Amyotrophic Lateral Sclerosis (ALS). It is most commonly seen in Asian countries like India and Japan affecting young males usually in the second or third decades. Pathogenesis of the disease is debated. Dynamic spinal cord compression due to neck flexion with forward displacement of posterior dura is considered as the

primary mechanism. The cause of the disease is unknown in majority of the people. We report a case of Hirayama's disease having unilateral involvement of the right upper extremity and describe the clinical features & the MR Imaging findings along with the mechanism behind its characteristic appearance.

CASE REPORT

18 year male came with complaints of weakness in his right hand and difficulty in gripping and holding objects since 1 year. Onset was insidious in nature and gradually progressed from difficulty in gripping objects to inability to unbutton the shirt with the right hand. There was no associated pain or loss of sensation. He had also noticed

atrophy of muscles of the right hand which had progressed to the medial aspect of the forearm. There was no history of trauma, febrile illness, poliomyelitis or exposure to heavy metals or toxins in the past (Figure 1).



Figure 1: Atrophy of the right palm as compared to the left.

On examination his higher functions were normal. Motor examination showed weakness & atrophy of thenar, hypothenar, interossei and foreram muscles of the right upper extremity except brachioradialis of the right side which was spared. Deep tendon reflexes were not exaggerated. Coordination and gait are normal (Figure 2).



Figure 2: Wasting of the right thenar, hypothenar & the muscles of the forearm along the ulnar aspect.

Blood investigations, blood urea, serum creatinine, serum electrolytes, liver function tests, thyroid function tests, erythrocyte sedimentation rate and creatine phosphokinase were within normal limits. Plain X-ray of cervical spine was normal except loss of lordosis was seen. Electromyogram showed chronic partial denervation with large amplitude long duration polyphasic potentials in the muscles of the right forearm and hand (Figure 3).

MRI cervical spine in neutral position showed focal short segment hyperintense signal in the ventral and right lateral aspect of the cervical cord at C5-C6 level with the involved segment measuring 4x3 mm in size. On flexion MRI there is mild diffuse widening of posterior epidural space. No evidence of ligament hypertrophy or intervertebral disc prolapse. The facetal joints, paravertebral soft tissues and cranio-cervical junction

were normal. The intervertebral foramina showed normal size and contour (Figure 4, Figure 5).



Figure 3: Plain X-ray cervical spine showing loss of cervical lordosis.



Figure 4: Cervical spine MRI in neutral position.



Figure 5: Cervical spine MRI in flexion.

The patient was given a hard cervical collar to prevent cervical flexion and physiotherapy in exercises of the hand and forearm. He has now been following up once every 2 months and after doing a thorough clinical examination of the involved extremity showing no deterioration, one can conclude that the disease has not progressed further since 4 months.

DISCUSSION

Hirayama's disease came into recognition in Japan in 1959 where it was reported as Juvenile muscular atrophy of unilateral upper extremity. 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. 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 (hypotenar, 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. 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. Since the pathogenesis clearly understood, probable causes suggest imbalanced growth between the patient's 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's disease show 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's disease.

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