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Radiology

A Case Report of Hirayama Disease Revealed by Upper Limb Weakness and Wasting

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Abstract **Case Report**

Hirayama disease is a rare entity characterized by unilateral or asymmetrical bilateral focal weakness and wasting of muscles innervated by C7, C8, and T1. We report the case of a 20-year-old male who presented with gradual left upper limb weakness and wasting, confirmed by electrophysiological and radiological studies showing detachment of the posterior longitudinal ligament with dilatation of the epidural veins. Anterior displacement with flattening of the medullary cord from C4 to C7 was observed, where there was a T2 hypersignal abnormality involving the anterior horns and producing a "snake eye" appearance. The disease is believed to result from the forward displacement of the cervical dural sac and spinal cord induced by neck flexion, and should be suspected in male patients presenting with unilateral or asymmetrical bilateral lower motor weakness of hands and forearms.

Keywords: Hirayama, snake eye appearance, amyotrophy.

INTRODUCTION

Hirayama disease is a rare condition characterized by unilateral or asymmetrical bilateral focal weakness and wasting of muscles innervated by C7, C8, and T1. It shows a gradual onset and benign course that commonly affects young males (male:female ratio of 20:1) between the ages of 15 and 25 years [1]. It was first described by Hirayama *et al.*, [2] in 1959 as "juvenile muscular atrophy of the unilateral upper extremity," and in 1984, Gourie-Devi *et al.*, [3] coined the term "monomelic amyotrophy."

The disease predominantly affects young men in the second to third decades of life and is characterized by an insidious onset, unilateral or bilateral asymmetric (rarely bilateral symmetric) weakness and atrophy of the forearm and hand, with sparing of the brachioradialis muscle, giving the characteristic appearance of oblique amyotrophy [4]. HD is a benign, self-limiting pathology; after a progressive phase of neurological deficits affecting the C7, C8, and T1 myotomes for about 1-5 years, it spontaneously arrests [5].

The incidence of Hirayama disease is low, and it is rarely encountered in clinical settings. Most case reports in the literature are from Asian countries, and it is rarely encountered in the Middle East or Arab countries. As a motor neuron disorder, it leads to atrophy of the involved muscles of the upper extremity due to an imbalance between the vertebral column and spinal canal content growth, making the dural sac thicker. These changes displace the posterior dural sac anteriorly on neck flexion, resulting in compression. Sensory and autonomic involvement is rare [6]. Atrophy slowly progresses and reaches a plateau typically over the course of several years before stabilizing. It is more prevalent in Asia, with a clear male predominance, particularly in the third decade of life [7]. In India, only 279 cases have been reported over a 35-year period [8].

Hirayama disease 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). Magnetic resonance imaging (MRI) features have been described in the literature to aid in the diagnosis of HD. In the present study, we report our experience of HD and review the clinical and MRI features in both neutral and neck-flexed positions.

Conservative management with cervical collars and physiotherapy is usually sufficient to relieve symptoms in early disease, but in advanced and severe

cases, surgical intervention has been found to be useful [9-12].

We report a case of Hirayama disease in a 26-year-old male who presented with gradual left upper limb weakness and wasting, which was confirmed by electrophysiological and radiological findings.

CASE REPORT

A 26-year-old previously healthy patient presented with a history of gradual loss of distal strength in the left upper limb associated with muscle atrophy that began at the age of 18 years. He reported severe cervical pain and the habit of flexing his neck to relieve the pain. There was no significant personal or family history. On physical examination, he showed moderate atrophy involving the distal musculature of both upper limbs, which was worse on the left (►Figure 1).

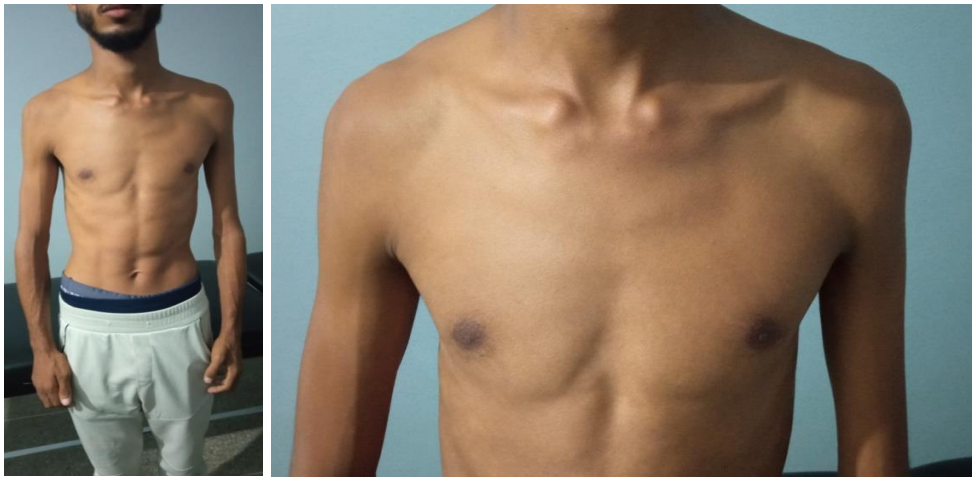
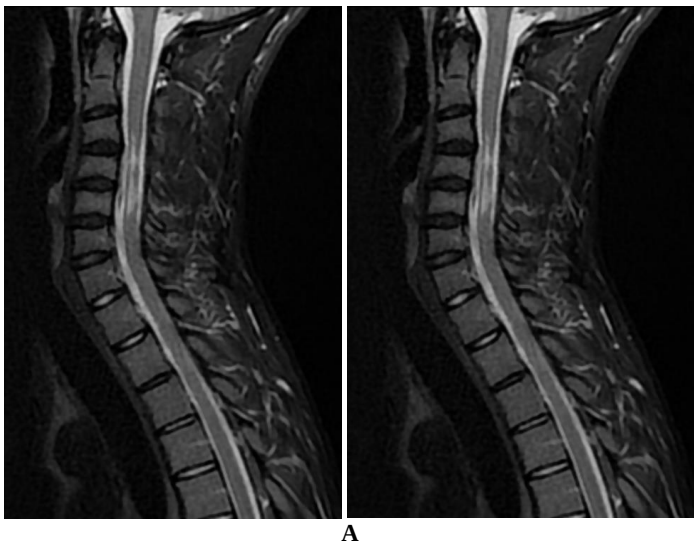
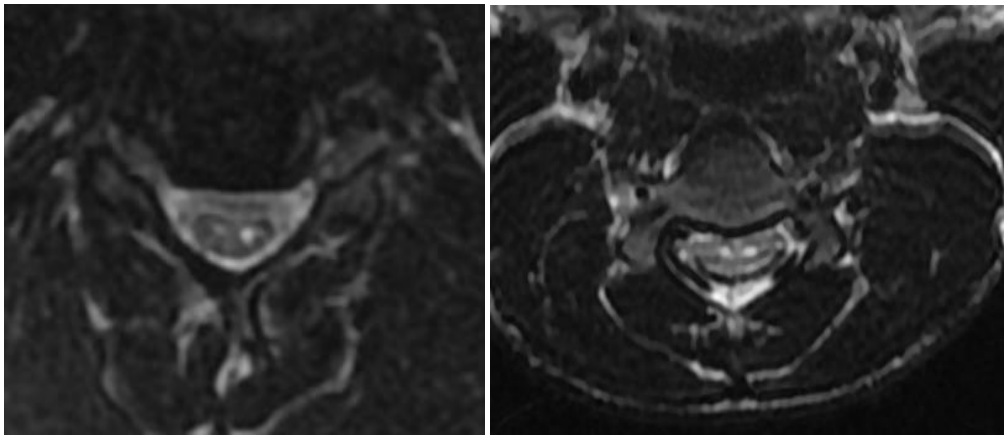


Figure 1: Pictures of the patient illustrates moderate atrophy involving the distal musculature of both upperlimbs, which was worse on the left

He also presented with reduced muscle tone, bicipital, tricipital, brachioradial, and finger flexor hyporeflexia, as well as the presence of minipolymioclonus, all on the left side. The EMNG performed showed pluriradicular involvement on the left side. The MRI (►Figure 2) showed spondylodiscal degenerative changes with central protrusions in C3-C4 to C6-C7, which were touching the dural sac. There was a reduction in the bone marrow caliber after the dynamic maneuver of forced flexion of the spine, as well as signal increase in the anterior horns. The anterior displacement of the posterior dura mater suggested an increase in mobility, which determines compression on the spinal cord. Additionally, there was detachment of the posterior longitudinal ligament with dilatation of the epidural veins. The anterior displacement also caused flattening of the medullary cord from C3 to C5, where there is a T2 high signal abnormality involving the anterior horns, producing a "snake eye" appearance.

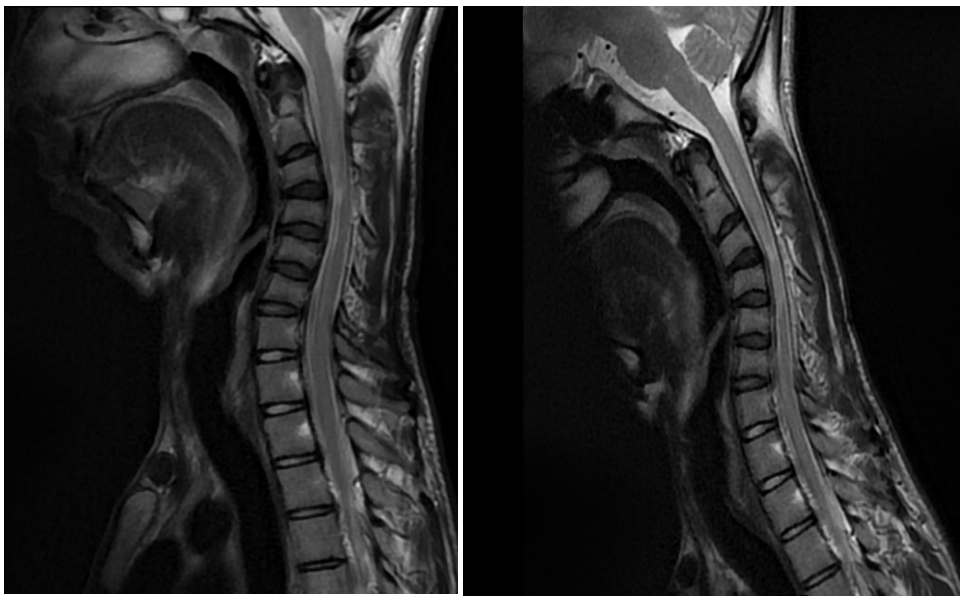


A



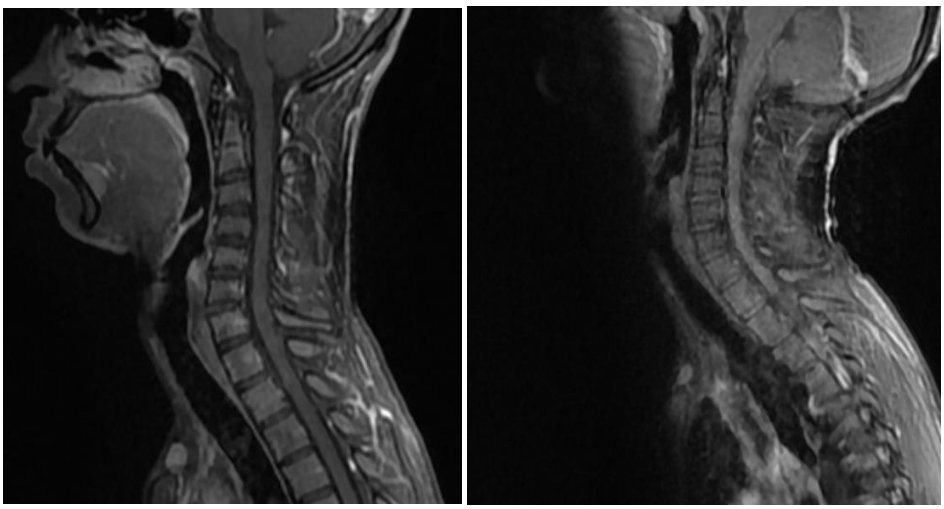
B

Figure 2: Sagittal view of STIR Sequence (A) Axial view of T2W sequence (B): demonstates a High signal changes on both STIR and T2W sequence involving the anterior horns and producing a "snake eye" appearance



A B

Figure 3: Sagittal view of T2W Sequence in neutral positon (A) in flexion position (B) demonstates: a reduction of the bone marrow caliber after the dynamic maneuver of forced flexion of the spine, as well as signal increase in the anterior horns



A B

Figure 4: Sagittal view of T1W FS Sequence before injection of Gadolinium (A) after injection of Gadolinium (B) demonstates a dilatation of the epidural veins

DISCUSSION

Initially described by Keizo Hirayama in 1959, Hirayama disease (HD) is also called non-progressive juvenile amyotrophy or monomelic amyotrophy [13]. The clinical diagnosis is suspected in view of specific criteria outlined by Tashiro *et al.*, [14] based on an epidemiological survey conducted in Japan. The criteria are: predominant distal muscle weakness and atrophy in the forearm and hand; involvement of the unilateral upper extremity in most cases; onset between the ages of 10 and 20 years; insidious onset with gradual progression during the first years, followed by stabilization; absence of involvement of the lower limbs; absence of sensory disturbances or abnormalities in the stretch reflexes; and exclusion of other diseases [14]. Distal atrophy of the forearm and hand, predominantly in the muscles of the Thenar eminence, hypothenar and interosseous musculature, causes an appearance typical of "oblique amyotrophy". Usually sporadic, HD has an unknown prevalence, and familial occurrence is extremely rare. In general, it is self-limited, initially presenting a progressive phase, which can range from 1 to 5 years, followed by a stationary period [15]. The pathological physiology is still debated. It has been suggested that Hirayama disease is a variant of motor neuron disease. Some studies have reported that there may be electromyographic damage to the muscles of the thoracic wall and lower limbs, lending weight to the hypothesis of a degenerative etiology [16]. But this has been refuted by autopsy data, which found ischemic phenomena. Indeed, pathological anatomy studies have revealed moderate gliosis, large and small neuron loss and central medullary necrosis of the lower cervical cord [11]. The pathophysiological hypothesis currently most widely accepted is that there is disproportionate growth of the spine relative to the dural sac [17].

The dural sac is attached to the spine at two fixed points : proximally at the foramen magnum and at C2 and C3, and distally at the coccyx. In a healthy subject, there is a certain laxity that allows it to adapt to flexion movements of the neck. However, in subjects with the disease, the dural sac is too short compared to the length of the spine, resulting in abnormal stretching in a neutral position, which causes poor juxtaposition with the posterior vertebral laminae [18].

During flexion, the dural sac cannot withstand the strain induced by the lengthening of the cervical spine, and as a result, it compresses the spinal cord against the posterior wall of the vertebral bodies, leading to an increase in intramedullary pressure. This pressure is responsible for local disturbances in the microcirculation of the anterior horns of the anterior cervical cord, which is the region most sensitive to ischemia [19]. It has been suggested that congestion of the epidural venous plexuses, aggravating this increased pressure, may play a role in the pathogenesis of the disease.

A combination of three pathophysiological factors is thought to be responsible for this congestion. Firstly, anterior displacement of the posterior dural sac would cause negative pressure in the posterior epidural space, resulting in increased flow in the posterior venous plexus. Secondly, anterior displacement of the dura mater would compress the anterior epidural venous plexus, resulting in an increased load on the posterior venous plexuses. Finally, the reduction in drainage of the jugular veins during flexion of the neck would impede the return of blood in the epidural veins [20].

Electromyoneurography (EMNG) may demonstrate signs of acute or chronic denervation in the intrinsic musculature of the hand, and a reduction in the amplitude of the compound muscle action potentials. Denervation may extend to clinically normal muscles in 25% to 50% of cases [21-22]. Routine MRI in the neutral position is often reported as normal, but it can also show lower cervical cord atrophy or abnormal cervical curvature (straight or kyphotic).

Routine MRI in a neutral position is often reported as normal, but it can also show lower cervical cord atrophy or abnormal cervical curvature (straight or kyphotic) [23]. However, the classical MRI findings of the cervical spine in neck flexion (30° to 40°) include forward displacement of the posterior wall, loss of the posterior dural sac attachment with adjacent lamina, and a well-enhanced crescent-shaped mass in the posterior epidural space of the lower cervical canal. This is thought to represent congestion of the posterior internal vertebral venous plexus as it disappears on neutral neck position [23]. The increase in the laminodural space and the presence of cervical cord flattening during flexion are essential for diagnosis, as described by Boruah *et al.*, [24] in 45 patients with clinically definite "Hirayama disease." They found that the laminodural space at maximum forward shifting of the posterior dural sac ranged from 3 to 9.8 mm, with a mean distance of 5.99 mm.

Vitale *et al.*, [16] suggested an MRI protocol that includes sagittal T1-weighted and T2-weighted sequences and axial T2 or T2*-weighted sequence in a neutral position; sagittal T2-weighted sequences and axial T2 or T2*-weighted sequence in a neck flexion of 25–35 degrees in addition to sagittal T1-weighted sequences in neck flexion before and after gadolinium intravenous administration.

The "snake-eyes" appearance is a radiological finding described as a symmetrical bilateral small high-signal-intensity lesion on axial T2-weighted MRI, which appears during the late stage of "Hirayama disease" and is proposed as an indicator of irreversible damage and poor prognosis [25].

The fact that MRI in flexion may show normality does not exclude the diagnosis, as it has been demonstrated that the disease normalizes after several years of evolution [4]. Several conditions can be considered in the differential diagnosis of "Hirayama disease," including amyotrophic lateral sclerosis, spinal muscle atrophy, C8-T1 radiculopathy, compressive myelopathy, cervical spondylotic myelopathy, syringomyelia, post-polio syndrome, multifocal motor neuropathy, and toxic neuropathy [26].

As "Hirayama disease" is considered a self-limited disease and often stops progressing after 1–5 years of onset, the mainstay of treatment consists of preventing neck flexion using a cervical collar to halt further progression. Its application at an early stage of the disease for 3–4 years has been advocated with a good response [27]. EMG can be used as an indicator to start and stop cervical collar therapy depending on changes of latency and amplitude of motor evoked potentials and persistence of F wave on neck flexion [28]. Good prognosis is seen in patients with a shorter duration of illness and no or mild cord atrophy in neutral neck position [29].

Surgical intervention has been proposed by some authors; however, patients with “Hirayama disease” usually stabilize with conservative treatment, and surgery should be limited to severe cases that have progressed quickly.

Cervical spinal decompression with fusion and duraplasty showed good results in selected patients because it gives a permanent stable fixation with a shorter period of immobilization [30]. Controlled clinical trials are needed to establish firm guidelines for these therapeutic options.

Syringomyelia, amyotrophic lateral sclerosis, cervical spondylosis associated with myelopathy, spinal cord tumors, and traumatic myelopathy are differential diagnoses of HD, and should be ruled out [5]. The management is usually conservative, with a cervical collar, avoiding sustained or repeated neck flexion.The surgical treatment with cervical spinal fusion and duraplasty is reserved for selected cases. Finally, the case here in reported illustrates the importance of the interaction among professionals in choosing the most appropriate method of investigation for the diagnostic.

CONCLUSION

Although rare and probably under-diagnosed, Hirayama disease should be included in the list of differential diagnoses in young patients with focal asymmetric atrophy of the upper limbs.

Establishing the diagnosis of hirayama disease early depends on the degree of suspicion, cooperation, and communication, the diagnosis can only be made based on the clinical picture and neutral position MRI.

Wearing a rigid cervical collar significantly slows progression of the disease.

An alternative has been proposed of surgical decompression, with encouraging results. Early recognition of this disease is important, given the existence of effective preventive measures.

PATIENT CONSENT

The authors confirm that a written informed consent in the local language was obtained from the patient for publication of the case report, on the conditions of maintaining anonymity of identity

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