



Case report

Hirayama disease with proximal upper limb involvement in an adolescent female: A case report

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ABSTRACT

Introduction and importance: Hirayama disease is a rare benign motor neuron disorder that involves a single upper limb. It affects the lower cervical myotomes and presents clinically by muscle wasting and weakness.

Case presentation: We report the case of a 17-year-old female who presented with a four weeks history of progressive weakness of the left upper limb. The blood investigations and image reported unremarkable findings. Patient improved on physiotherapy.

Clinical discussion: Hirayama disease is rarely encountered in clinical settings and should be suspected in female patients presenting with unilateral or asymmetrical bilateral lower motor weakness of hands and forearms.

Conclusion: We present a rare condition in a 17-year-old female with a left upper extremity monomelic amyotrophy, a Hirayama disease.

1. Introduction

Hirayama disease is a rare and benign motor neuron disorder that involves a single upper extremity which was originally reported and called by Hirayama et al. in 1959, as “juvenile muscular atrophy of unilateral upper extremity” [1,2]. In 1984, Gourie-Devi et al. coined the term “monomelic amyotrophy” (MA) [3].

The disease affects generally the lower cervical myotomes and presents clinically as muscle wasting and weakness [4]. Commonly affects young males (male: female ratio of 20:1) between the ages of 15 and 25 years [1]. It is frequently reported in most eastern world countries like in India, Japan, and other Asian countries, but many cases have been reported from other parts of the world as well [5–7].

The etiology of Hirayama disease is still unknown but theories reported that it occurs secondary to chronic compression of spinal cord or atrophy [8]. We reported a 17-year-old female who was managed in surgical department with a clinical diagnosis of Hirayama disease, according to SCARE 2020 criteria [9].

2. Case presentation

A 17-year-old female presented with insidious-onset, slowly progressive weakness and wasting of her left deltoid and shoulder girdle muscles for 4 weeks prior to the presentation. Initially, she noticed difficulty with abducting her left arm and raising it above head level. The weakness progressed slowly over time and then became static. She denied history of trauma but she had shoulder pain on abduction, and she was not on any medications prior to development of symptoms.

No history of chronic disease such as HIV, Diabetes mellitus or any other disease such as syphilis was reported. The neurological examination demonstrated significant atrophy of his left deltoid, with no fasciculations (Fig. 1). The muscle strength, (according to Medical Research Council) was 2. The tendon reflexes and sensory examination were all within the normal range.

All Blood investigations including biochemical and blood count parameters and Brain CT scan were normal. Electromyography (EMG) and magnetic resonance imaging (MRI) studies were not performed due to their unavailability in our hospital at the time of patient admission.

Based on the clinical presentation and the normal findings on the CT scan, we concluded on left upper extremity monomelic amyotrophy, a

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Hirayama disease. The patient was prescribed for regular physiotherapy to strengthen her shoulder muscles and improve range of movement. After two months of physiotherapy there was marked improvement in range of motion and individual muscle power with a strength of 4 (Fig. 2).

3. Discussion

Hirayama disease is a benign form of focal amyotrophy which was first published in 1987 and it's a form of juvenile muscular atrophy of unilateral upper extremity [1]. It is characterized by asymmetric, unilateral weakness and atrophy of the hand and forearm muscles, with sparing of the brachioradialis and several cases of lower limb involvement have been reported [2].

Majority of these cases are reported in Japan and a few in Western World but this is the first case to be reported in Africa and among females since Hirayama is more prevalent in males [10] and symptoms usually start in the 20s and limited in a single extremity; in this case it was the shoulder which was involved and this is unusual and rarely reported in literature [11]. Clinical presentation often occurs insidiously and approximately 70 % of the patients experience disease progression within 3 years, and approximately 95 % stabilize 5 years after disease onset [12] however in this case the disease progression was within 4 weeks.

Presentation is usually asymmetric muscle weakness either unilateral or bilateral and can be atrophic in young males and is usually self-limited [1]. These symptoms can be associated with autonomic dysfunction in distal upper limbs in the form of exaggeration of weakness on exposure to cold (cold paresis), cold skin, excessive sweating, and hair loss over the dorsum of the hands. Our patient presented with a rare form of Hirayama disease, affecting the shoulder muscles with no sensory symptoms and upper motor neuron [11].

Pathophysiology remains unknown and viral infection, atopy, ischemia of anterior horn cells and a tight dural canal are predisposing factors [11]. Neuroradiologic examination often reveals cervical compression and flattening of the lower cervical cord due to forward displacement of the cervical dural sac and spinal cord [8]. In this case CT spine was normal however diagnosis can be confirmed by electrophysiological and radiological studies including MRI which are not available in this setting [13]. EMG findings would be indicative of chronic denervation in the C7, C8, and T1 innervated muscles, with or without acute denervation potentials [4,8]. MRI findings are usually spinal cord atrophy, enhancement of posterior epidural venous and anterior shifting of posterior dural sac on flexion [11].

Treatment using a cervical collar in the acute phase has been advocated for; however, the role of surgery is controversial for patients who progress [13]. In this case conservative management which included physiotherapy was enough to produce encouraging results. Cervical spinal decompression with fusion and duraplasty has shown good outcome in carefully selected patients as it gives a permanent stable



Fig. 2. Normal cervical spine in the neutral position shown on a CT scan.

fixation with a shorter period of stability [12].

4. Conclusions

Hirayama disease is rarely encountered and should be suspected in low-income countries through clinical examination due to lack of investigative radiology and electrophysiological studies. Physiotherapy is still the conservative management type as per our case. The availability of electrophysiological study and magnetic resonance imaging is needed in diagnosis process.

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Ethical approval

Not applicable.

Consent for publication

Written informed consent was obtained from the patient for



Fig. 1. Clinical photograph showing atrophy of the left shoulder.

publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

FKS managed the patient and wrote the first draft. SMK, AAO and AK helped in editing and reviewing the paper. FKS wrote the first draft. All authors read and approved the final version to be published.

Research registration

Not applicable.

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Declaration of competing interest

The authors declare no conflicts of interest.

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