



## Case Report

# Hirayama's Disease in a Young Belgian Male: Case Report and Review of Literature

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### Abstract

Hirayama's disease, also known as juvenile non-progressive amyotrophy, is a rare neurological pathology characterized by an asymmetric distal amyotrophy of the upper limb affecting myotomes C7 to T1 in young males. Hirayama's disease is most commonly observed in Asian population. The aim of this article is to report disease in a young European patient and to describe the clinical presentation and investigations.

**Keywords:** Hirayama's Disease; Cervical Myelopathy; Monomelic Amyotrophy (MMA); Juvenile Non-Progressive Amyotrophy; Flexion MRIA.

### Introduction

Hirayama's disease, also termed juvenile non-progressive amyotrophy, or monomelic amyotrophy, first described by Hirayama et al in 1959 [1], is a rare type of cervical myelopathy related to flexion movements of the neck causing an asymmetric distal weakness or amyotrophy of muscles innervated by C7 to T1. It is most commonly in Asia, and it mainly affects young men in their second or third decade. The pathogenesis of this disease is not fully clarified. Dynamic cervical magnetic resonance imaging (MRI) is fundamental for the diagnosis. Conservative treatment with cervical collar and physiotherapy is effective in most cases but some severe cases require surgical treatment. This article reports a case of a 17-year-old male who was diagnosed with Hirayama's disease, and describe the clinical presentation and investigations.

### Case Presentation

A previously healthy Caucasian 17-year-old male patient

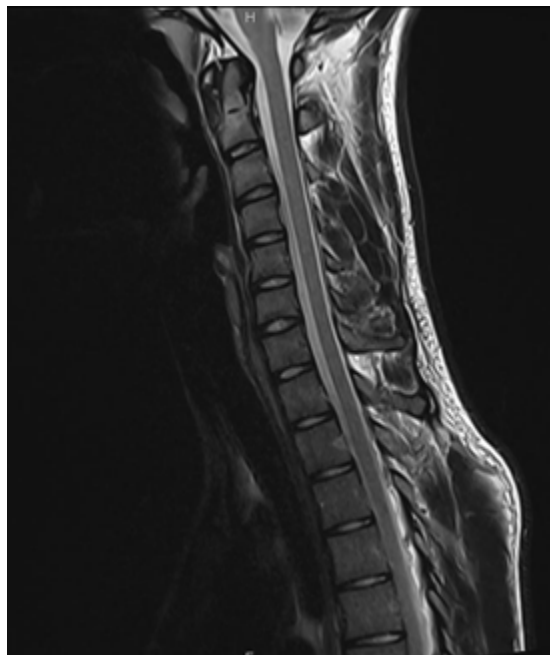
developed for several months progressive weakness in the right upper limb associated with amyotrophy involving the muscles of the hand, forearm and triceps. There was neither context of trauma nor family history of neuromuscular disorders. He did not report any sensory disturbance or associated cramp or fasciculations. He had no complaints of neck pain. Furthermore, he reported no disorders in the left upper limb, nor the lower limbs. Clinical examination revealed an atrophy of the hypothenar, thenar and interosseous muscles. There was no pyramidal syndrome. Examination of the cranial pairs, cervical spine and other limbs was normal. Blood biology was normal including anti-GM1 antibodies. Cerebrospinal Fluid (CSF) analysis showed normal cytochemistry and an absence of specific IgG oligoclonal band

Electromyography (EMG) showed a purely motor neurogenic disease of C7 to D1 associated with signs of acute denervation with fibrillations and fasciculations. There was a reduction in the distal motor amplitude, which was clearly predominant in the first interosseous muscle compared to the short abductor muscle of the thumb. The left upper limb showed more moderate motor abnormalities. The lower limbs results were normal Cervical spine X-Ray was normal. However, we noted a disappearance of cervical lordosis (Figure 1).



**Figure 1:** X-ray of the Cervical Spine Revealing a Loss of Cervical Lordosis.

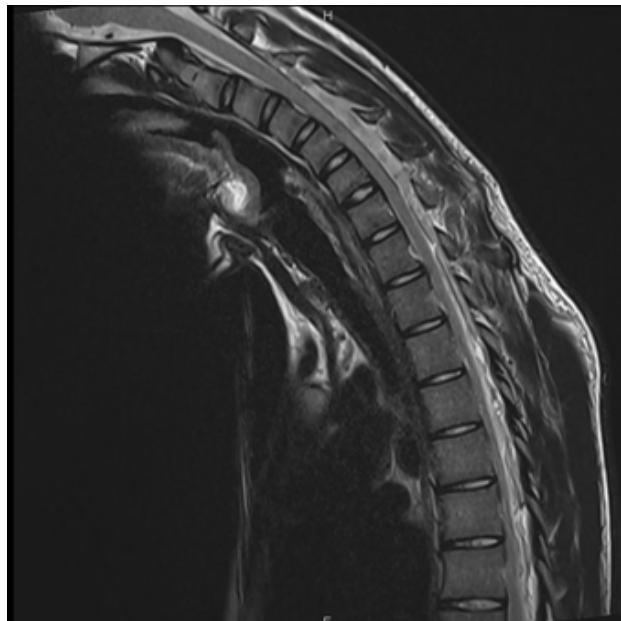
The initial cervical MRI in neutral position was normal (Figure 2).



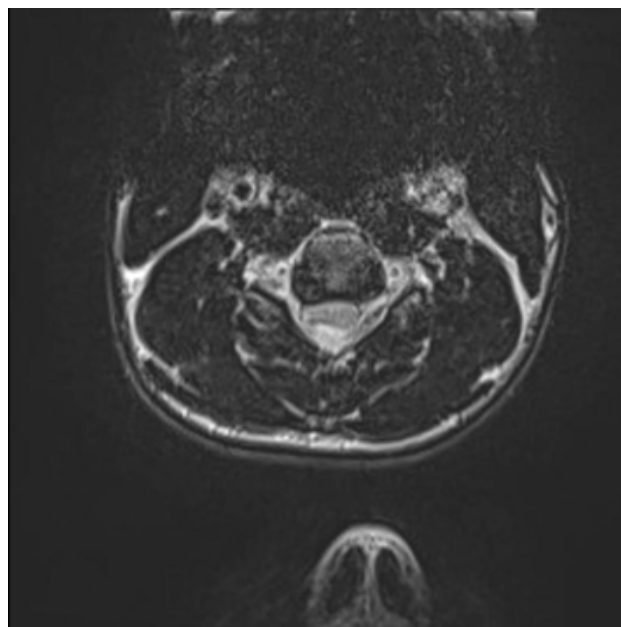
**Figure 2:** T2 Sagittal Cervical Spine in Neutral Position.

In forced flexion of the cervical spine, MRI showed an enlargement of the posterior lamino-dural space from C3 to C7 (7.4 mm) along with an anterior displacement of the dural sac, a

collapse of the subarachnoid spaces and a small compression of the spinal cord predominant on the right side (figure 3, figure 4)



**Figure 3:** T2 Sagittal Cervical Spine in Flexion.



**Figure 4:** T2 Axial Cervical Spine in Flexion.

According to the clinical findings and the analysis of flexion cervical MRI, the diagnosis of Hirayama's disease was confirmed. The treatment consisted in wearing a cervical collar to avoid flexion of the neck. It was followed regularly on the clinical and electrophysiological level. We observe a stable evolution at 10

months without progression of complaints.

## Discussion

Hirayama's disease was first described by Keizo Hiramaya in 1959 as a juvenile muscular atrophy of unilateral upper extremity [1]. Thereafter, the same pathology has been reported under several names; benign focal amyotrophy [2]; monomelic amyotrophy [3]; and juvenile asymmetric segmental spinal muscular atrophy [4], all these terms refer to the same disease. Most cases have been reported in Asia. Cases reported in Europe are still rare where it probably remains underdiagnosed [5-11].

The course of Hirayama's disease is progressive with an insidious onset, followed by spontaneous resolution over 2 to 5 years.

It usually affects young men in their second to third decade.

The clinical features include unilateral or asymmetric weakness and amyotrophy in the forearm and the hand, with sparing of brachioradialis, giving the appearance of oblique amyotrophy. It can be associated with autonomic dysfunction causing cold paresis and muscle cramps.

Tashiro recently defined the criteria requirements for diagnosis of Hirayama's disease [12]:

1. Distal predominant muscle weakness and atrophy in forearm and hand
2. Involvement of the unilateral upper extremity
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 (e.g., motor neuron disease, multifocal motor neuropathy, brachial plexopathy, spinal cord tumors, syringomyelia, cervical vertebral abnormalities, anterior interosseous, or deep ulnar neuropathy)

Other disorders may cause upper limb muscle weakness or amyotrophy and should be considered in the differential diagnosis of Hirayama's disease : Motor Neurone Disease (MND's) including Amyotrophic lateral sclerosis (ALS), Spinal Muscular Atrophy (SMA) and Primary Lateral Sclerosis (PLS), Cervical Spondylotic Myelopathy (CSM), Syringomyelia, Myotonic Dystrophy, Ossification of the Posterior Longitudinal Ligament (OPLL), Intramedullary spinal cord tumors, Peripheral nerve compression syndromes (carpal tunnel syndrome, thoracic outlet syndrome).

Conventional radiographic of the cervical spine may show loss of lordosis but is often aspecific [13]. MRI is the best diagnostic tool to confirm the diagnosis and exclude other spinal cord disorders.

Cervical MRI in neutral position is not always contributory but may reveal localized atrophy of the cervical cord, T2 hyperintensity signal, loss of lordosis, and loss of attachment of the dorsal dural sac to the subjacent lamina.

MRI in flexion is critical to confirm diagnosis and shows *anterior migration* of the posterior wall of the dura and an enlarged, crescent shaped and enhancing posterior epidural space, best appreciated with T2-weighted [14].

Electromyography (EMG) and Nerve Conduction Studies (NCS) reveal chronic denervation changes in C7-T1 myotomes and absence of sensory involvement. The reduction of the distal amplitudes predominates in the cubital territory compared to the Median [15], which was also noted in this case.

Autopsy findings demonstrate that the main pathology is asymmetrically located in the cervical anterior horn and anterior roots, with a central necrosis and decrease in nerve cells without macrophage infiltration at the C7-C8 levels [16].

The exact pathogenesis of this disease is still controversial. Many theories have been formulated. The causal factor is the forward displacement of the posterior lower cervical dura during neck flexion. The pathophysiological theories leading to Hirayama's disease include:

1. Compressive cervical myelopathy in flexion: The most commonly accepted theory describes the repetition of these flexion movements causing micro ischemia in the cells of the anterior horn of the lower cervical cord in the C7-T1 areas. This condition leads to myelopathy and degeneration, as evidenced by the asymmetric atrophy of the lower cervical cord seen on cervical MRI [17].
2. Dysplasia of the dural sac: Ito proposed that dural sac dysplasia result from imbalance between the growth of cord and the growth of spinal canal during puberty, leading to tightness of dura. This explains the spontaneous resolution after the end of growth [18].
3. Nerve root dysplasia: Toma suggests that nerve root dysplasia may result from a insufficient length of the cervical nerve roots. During neck flexion, the posterior roots are unable to extend, which results in anterior traction of the spinal cord on the affected side [19].
4. Structural abnormalities of the spinal ligament: Loss of the elastic fibers of the spinal ligament and the epidural ligament in relation to the normal wavy structure of the dura may also lead to tightness of the dura [20].

- Engorged posterior epidural venous plexus: engorgement of the posterior epidural venous plexus is also considered another possible cause of compression of the cervical cord [21].
- Immunologic mechanisms: Several studies have observed a correlation between high Ig E immunoglobulin levels and the development of Hirayama disease, as well as a correlation with clinical manifestations [22,23].

The first-line treatments is mainly conservative and consist on applying a cervical collar to avoid neck flexion and preventing compression injury to the spinal cord during flexion [24]. Surgery is rarely required. Indications for surgery include failure of conservative treatment, poor compliance to the cervical collar, rapid progression of severe symptoms, a significant degree of spinal cord atrophy, and positive pyramidal signs [25].

Different surgical techniques have been described, based on the pathophysiology. The goal of the surgery is to avoid further cord compression by preventing neck flexion or by decompressing the cord.

Posterior approaches are most commonly practiced, including laminectomy with resection of the posterior venous plexus [26], laminoplasty with duraplasty [27], posterior fixation in situ [28].

Anterior approaches may also be performed, as Anterior Cervical Discectomy and Fusion (ACDF) [29] or anterior cervical corpectomy and fusion (ACCF) [30].

The prognosis of Hirayama's disease is good compared to other motor neuron disorders such as SMA, ALS, PLS, with longer survival and fewer morbidity. The complications remain rare and involve chronic atrophy of the muscles and permanent contractures sometimes associated with spasticity.

#### **Declarations**

#### **Ethical Approval and Consent to Participate**

Not applicable

#### **Consent for Publication**

Patient consent for case report

#### **Availability of Data and Materials**

The datasets analysed during the current case report are available in the GHDC XCare repository

#### **Competing Interests**

The authors declare that they have no competing interests

#### **Authors' Contributions**

OA wrote the article and browsed the literature. ST and MG reviewed and approved the article all authors read and approved the final manuscript.

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