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Case Report

Unilateral Phrenic Nerve Palsy Secondary to Hirayama Disease: First Description of Two Cases

Marcos Polanco[1*], Jorge Madera[1], Ana Lara Pelayo[1], Natalia Valle[2], Elena Valle[3], Julio Pascual[1]

[1]Service of Neurology, University Hospital Marqués de Valdecilla, Santander, Spain

[2]Service of Radiology, University Hospital Marqués de Valdecilla, Santander, Spain

[3]Service of Neurophysiology, University Hospital Marqués de Valdecilla, Santander, Spain

[*]**Corresponding author**: Marcos Polanco, Service of Neurology, University Hospital Marqués de Valdecilla, Santander, Spain

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Introduction

Hirayama disease is a rare cause of cervical myelopathy, with male predominance, which typically presents as distal, painless and asymmetric weakness and amyotrophic of one upper extremity, usually corresponding to C7-T1 myotomes. Its etiology is not entirely clear, although it is believed to be due to a compression of the cervical spinal cord produced during neck flexion, characteristically presenting in the cervical MRI (performed in flexion) an abnormal anterior displacement of the posterior dura mater. This detachment would cause the compression of motor neurons located in the anterior horn of the spinal cord being responsible for the weakness and amyotrophic characteristic of this disease. This disease typically begins in young adulthood and usually stabilizes in the first 5 years after its onset [1,2].

Although Hirayama disease characteristically presents as exclusive second motor neuron involvement restricted to the C7-T1 territory, atypical clinical manifestations have recently been described, such as the association of sensory or first motor neuron symptoms and even shoulder girdle amyotrophic, secondary to a detachment of the dura mater in more proximal segments. [1-3] here we describe two cases of phrenic nerve palsy secondary to Hirayama disease.

Keywords: Hirayama's disease, Phrenic nerve palsy, diaphragmatic palsy, weakness.

Case Reports

Case 1

A 40-year-old male with no history of interest except allergic bronchial asthma consulted due to a 1-year history of progressive, painless, distal atrophy of the right upper extremity with thenar predominance and loss of strength in the right hand (2nd and 3rd fingers and interosseous muscles). Neurophysiological studies showed denervation restricted to muscles depending on C8-T1. A cervical MRI disclosed a slight increase in the posterior epidural space at C5, C6, and C7 with a detachment of the posterior dura mater more striking in flexion. Over the next two years, he progressively developed proximal right upper limb weakness with difficulty in shoulder elevation. In the context of dyspnea attributed to an asthmatic exacerbation he was evaluated in the Emergency Department where an elevation of the right diaphragmatic dome was detected, confirmed by chest X-ray (Figure 1) and thoracic CT. He was referred to our Respiratory Service, where a right diaphragmatic palsy was confirmed. Complementary tests were completed with a spirometry that showed a forced vital capacity of 86% with a fall of 16% in decubitus.

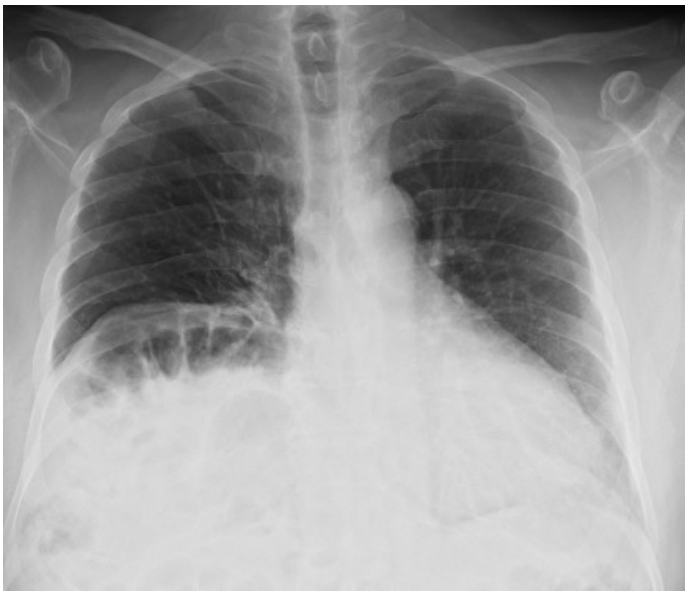

Figure 1: Chest X-ray of Case 1 showing elevation of the right diaphragm.

There were no alterations in the routine laboratory tests. In addition, a neurophysiological study of both phrenic nerves revealed a 50% reduction in the amplitude of the phrenic nerve potential of the right side versus the left side.

Case 2

This 20-year-old male with no history of interest consulted due to progressive, painless, proximal atrophy and weakness of the upper limbs of clear right predominance. The initial clinical examination showed amyotrophy of the right deltoid and infraspinatus muscles, with decreased muscle balances in the right deltoid (3/5), shoulder external rotation (3/5), biceps (4/5) and supinator longus (4/5). Reflexes were present and symmetrical and there were no sensory alterations. A neurophysiological study found a neurogenic pattern in the muscles corresponding to C5-C6 of severe intensity on the right side and moderate intensity on the left. The study was completed with a cervical MRI that showed a posterior thickening and detachment of the dura mater in the C2-C5 territory (Figure 2).

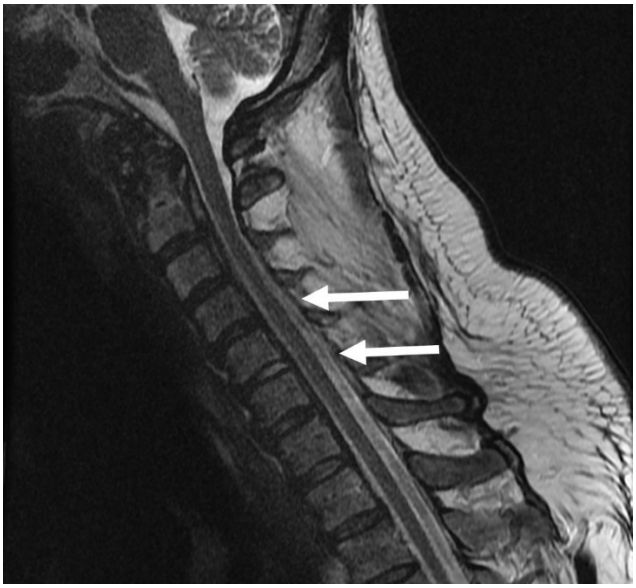

Figure 2: Sagittal MRI flexion section (T2 sequence) of Case 2 showing posterior dural detachment at C2-C5 segment (white arrows).

The patient attended the Emergency Department due to chest discomfort, and a chest X-ray showed an elevated right hemidiaphragm, which was later confirmed with a thoracic CT scan. Spirometry showed a 21% decrease in forced vital capacity when he lied down. An electroneurographic study of both phrenic nerves showed a decreased amplitude with normal latency in the right side, compatible with axonal involvement of this nerve.

Discussion

To the best of our knowledge, this is the first description of two patients with imaging-confirmed Hirayama disease presenting with unilateral phrenic nerve palsy. The phrenic nerve arises from the ventral branches of the C3-C5 roots of the cervical plexus. Lesions at this level can cause unilateral or bilateral diaphragmatic paralysis. Classically Hirayama disease affects lower cervical spinal segments (C7-T1), but cases have been described with the involvement of two segments above. This might affect the C5 root, which contributes to the formation of the phrenic nerve. [4,5] C5 root involvement can lead to weakness of the deltoid, external rotator cuff and forearm flexors as happened in case 2 [3].

The most frequent causes of phrenic nerve injury are secondary to thoracic or cardiac surgery, trauma, tumors, metabolic diseases such as diabetes, or infections. Other less frequent etiologies include multiple sclerosis, cervical spondyloarthrosis, myopathies, and autoimmune diseases, such as Guillain-Barré syndrome. [4] Our two cases, show, first, that Hirayama disease should be considered as a possible etiology in the differential diagnosis in patients with unilateral phrenic palsy of unknown origin and, second, that diaphragmatic palsy should be ruled out in this disease, especially in the presence of respiratory symptoms.

Conclusion

Hirayama's disease should be included in the differential diagnosis of unilateral phrenic paralysis of unexplained cause. This palsy would be explained by compression at the level of the C3-C5 roots due to the dural detachment characteristic of this pathology. Likewise, phrenic neuropathy should be ruled out in patients diagnosed with Hirayama's disease.

Acknowledgments

None

Ethics and patient consent

Informed consents were obtained from the patients for the publication of this manuscript.

Conflict of Interest Statement

The authors report no conflict of interest in connection with this article.

References

1. Hirayama K. (2000) Juvenile muscular atrophy of distal upper extremity (Hirayama disease): focal cervical ischemic poliomyelopathy. Neuropathology. 20: S91-S94.
2. Wang H, Tian Y, Wu J, Luo S, Zheng C, et al. (2022) Update on the Pathogenesis, Clinical Diagnosis, and Treatment of Hirayama Disease. Front Neurol. 12: 811943.
3. Wang H, Tian Y, Wu J, Sun C, Nie C, et al. (2022) The radiological and electrophysiological characteristics of Hirayama disease with proximal involvement: A retrospective study. Front Neurol. 13: 969484.
4. O'Toole SM, Kramer J. (2023) Unilateral Diaphragmatic Paralysis. In: StatPearls. Treasure Island (FL): StatPearls Publishing.
5. Nason LK, Walker CM, McNeeley MF, Burivong W, Fligner CL, et al. (2012) Imaging of the diaphragm: anatomy and function. Radiographics. 32: E51-E70.