Hirayama Disease: Case Report*

Doença de Hirayama: Relato de caso

Victor Alves Rodrigues[1] Matheus Rocha Pereira Klettenberg[1] Luciano Farage[2] Lisiane Seguti[1]

[1] Pediatric Neurologist, Hospital Universitário de Brasília, Brasília, DF, Brazil
[2] Radiologist at Incor, Brasília, DF, Brazil

Address for correspondence Victor Alves Rodrigues, MD, Hospital Universitário de Brasília, SGAN 605, Av. L2 Norte, Brasília, DF, Brasil (e-mail: victoralvesrodrigues2017@gmail.com).

Rev Bras Ortop 2022;57(4):697–701.

Abstract

A 26-year-old previously healthy patient who, at the age of 18 years, began progressive loss of distal strength, rest tremor, and muscle atrophy in the left upper limb. Upon examination, the patient presented moderate distal atrophy, degree 4 in muscular strength, and minipolymioclonus. Electromyoneurography revealed (EMNG) chronic preganglionic bilateral involvement of bilateral C7/C8/T1, worse on the left, with signs of active C8/T1 denervation. A cervical spine magnetic resonance imaging (MRI) scan showed spondylodiscal degenerative changes with central protrusions in C4-C5, C6-C7, and right central in C5-C6, which touched the dural sac. The anteroposterior diameter of the medulla in neutral position, in the C5-C6 plane, was of 5.1 mm. There was a reduction of the spinal cord caliber to 4.0 mm after the dynamic maneuver of forced flexion of the spine, as well as signal increase in the anterior horns. The clinical findings and those of the complementary tests were compatible with Hirayama disease (HD), a rare benign motor neuron disease that affects cervical spinal segments and is most prevalent in men, with onset in the early 20s. Unilateral and slowly progressive weakness is typical, but self-limited. Sensory disturbances, and autonomic and upper motor neuron signals are rare.

Management is usually conservative, with the use of a soft cervical collar. Although rare, HD should be considered in young patients with focal asymmetric atrophy in the upper limbs. The early diagnosis of HD depends on the degree of suspicion, as well as on the cooperation and communication among the various specialties involved in the investigation.

Keywords

- Hirayama disease
- muscular atrophy, spinal
- spinal cord
- magnetic resonance imaging

* *Work developed at Hospital Universitário de Brasília, Brasília, DF, Brazil.*

received
May 30, 2021
accepted
October 14, 2021

DOI https://doi.org/10.1055/s-0042-1742339.
ISSN 0102-3616.

Thieme Revinter Publicações Ltda., Rua do Matoso 170, Rio de Janeiro, RJ, CEP 20270-135, Brazil

Resumo

Paciente de 26 anos, previamente hígido, que, aos 18 anos, iniciou perda progressiva de força distal, tremor de repouso, e atrofia muscular no membro superior esquerdo. Ao exame, apresentou atrofia moderada, distal, força muscular de grau 4, e minipolimioclonus. A eletroneuromiografia (ENMG) revelou comprometimento pré-ganglionar crônico de C7/C8/T1 bilateral pior à esquerda, com sinais de desnervação ativa em C8/T1. A ressonância magnética (RM) de coluna cervical mostrou alterações degenerativas espondilodiscais com protrusões centrais em C4-C5, C6-C7, e central direita em C5-C6, que tocavam o saco dural. O diâmetro anteroposterior da medula na posição neutra, no plano de C5-C6, era de 5,1 mm. Houve redução do calibre da medula para 4,0 mm após a manobra dinâmica de flexão forçada da coluna, e aumento de sinal nos cornos anteriores. Os achados clínicos e os dos exames complementares eram compatíveis com doença de Hirayama (DH), uma doença benigna rara dos neurônios motores, que afeta os segmentos espinhais cervicais e é mais prevalente em homens e de início próximo aos 20 anos. É típica a fraqueza unilateral e lentamente progressiva, porém autolimitada. Perturbações sensoriais, sinais autonômicos e do neurônio motor superior são raras.

O manejo geralmente é conservador, com uso de colar cervical macio. Apesar de rara, a DH deve ser considerada em pacientes jovens que apresentam atrofias assimétricas focais de membros superiores. O diagnóstico precoce de DH depende do grau de suspeição, e da cooperação e comunicação entre as diversas especialidades envolvidas na investigação.

Palavras-chave

- doença de Hirayama
- atrofia muscular espinal
- medula espinal
- imagem por ressonância magnética

Introduction

Initially described by Keizo Hirayama in 1959, Hirayama disease (HD) is also called non-progressive juvenile amyotrophy or monomelic amyotrophy.[1] The clinical diagnosis is suspected in view of certain criteria outlined by Tashiro et al.[2] 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.[2] 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".[1]

Usually sporadic, HD has an unknown prevalence, and family occurrence is extremely rare.[1] In general, it is self-limited, initially presenting a progressive phase, which can range from 1 to 5 years, followed by a stationary period.[3]

There is no clear pathophysiological explanation. One of the theories postulated is that the disproportionate growth between the spine and its contents, during the growth spurt phase, could create a distension to the posterior wall of the dural canal that would cause anterior displacement of the dura mater in flexion. The displaced dura mater would lead to compression of the spinal cord, resulting in local disturbances in microcirculation and necrosis of the cells of the anterior horn.[1,4]

Electromyoneurography (EMNG) may demonstrate signs of acute or chronic denervation in the intrinsic musculature of the hand and reduction of the amplitude of the compound muscle action potentials. Denervation may extend to clinically normal muscles in 25% to 50% of the cases.[1,5]

Dynamic cervical magnetic resonance imaging (MRI) is essential for the diagnosis of HD. The main MRI findings in neutral position are: localized atrophy of the lower cervical cord/asymmetric flattening of the spinal cord; intrinsic spinal cord hypersignal on T2-weighted images, and an abnormal cervical curvature with loss of insertion between the posterior dural sac and the underlying lamina. Flexion MRI (30° to 40°) shows: anterior displacement of the posterior wall of the cervical dural sac and dorsal flattening of the lower cervical cord due to the enlargement of the posterior epidural space attributed to congestion of the epidural venous plexus.[6]

The aim of the present study is to report a case of HD diagnosed late, after suspicion by EMNG, emphasizing the importance of the electro-clinical correlation and communication among the professionals involved in the diagnostic "odyssey" (routine) of a rare disease.

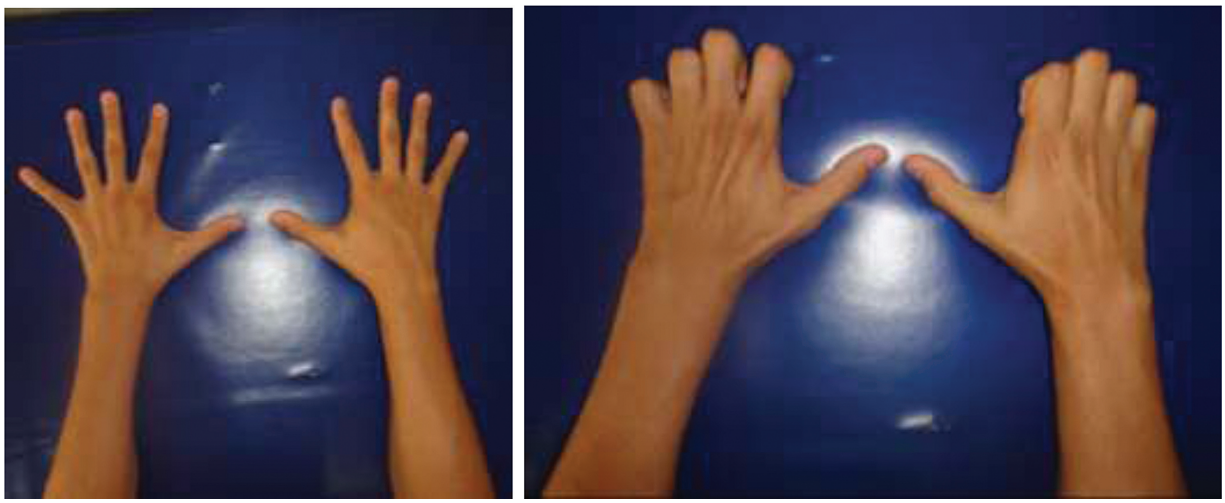

Fig. 1 Asymmetric atrophy affecting the left upper limb.

Case Report

The present report was approved by the institutional Ethics in Research Committee (CAAE 34383220.9.0000.5558), and the informed consent form was signed.

A 26-year-old previously healthy patient who, at the age of 18 years, started a progressive loss of distal strength in the left upper limb associated with muscle atrophy. He reported severe cervical pain and the habit of flexing his neck to relieve the pain. It evolved without restrictions to his occupation as a computer technician.

There was no significant personal or family history, except for the report of mild head injury after a fall from a height of two meters, with head trauma against the ground, which occurred at the age of 8 years.

Upon physical examination, he presented with moderate atrophy involving the distal musculature of both upper limbs, which was worse on the left (►**Figure 1**). He also presented reduced muscle tone, grade-4 distal and grade-5 proximal muscle strength, bicipital, tricipital, brachioradial and finger flexor hyporeflexia, and presence of minipolymioclonus, all on the left side.

The right brachial plexus MRI was normal. The first cervical MRI showed small disc protrusions from C4-C5 to C6-C7, touching the dural sac. The EMNG performed three years after the initial investigation showed chronic preganglionic involvement and active denervation in C7/C8/T1 which was bilateral and worse on the left. In this examination, HD was hypothesized, and a dynamic cervical MRI was requested.

The new MRI scan of the cervical spine (►**Figure 2**) showed spondylodiscal degenerative changes with central protrusions in C4-C5, C6-C7, and right central in C5-C6, which touched the dural sac. The anteroposterior diameter of the spinal cord in neutral position on the C5-C6 plane was of 5.1 mm. There was a reduction of the bone marrow caliber to 4.0 mm 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.

Discussion

The patient in the case herein reported presented clinical findings and those of the complementary tests compatible with HD, confirmed only eight years after the onset of symptoms. The diagnosis was only possible due to suspicion of the neurophysiologist, who suggested dynamic cervical MRI.

Vitale et al.,[3] in a prospective study, observed that 100% of HD patients presented loss of cervical lordosis on MRI. The sensitivity gain with the MRI in dynamic flexion is of 87% when compared to the sensitivity of 49% of the MRI in neutral position.[3]

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.[7]

The management is usually conservative, with a cervical collar, avoiding sustained or repeated neck flexion.[1,8] The surgical treatment with cervical spinal fusion and duraplasty is reserved for selected cases.

Finally, the case herein reported illustrates the importance of the interaction among professionals in choosing the most appropriate method of investigation for the diagnostic conclusion.

Although rare, HD should be included in the list of differential diagnoses in young patients with focal asymmetric atrophy of the upper limbs. Establishing the diagnosis of HD early depends on the degree of suspicion, cooperation, and communication among the various specialties.

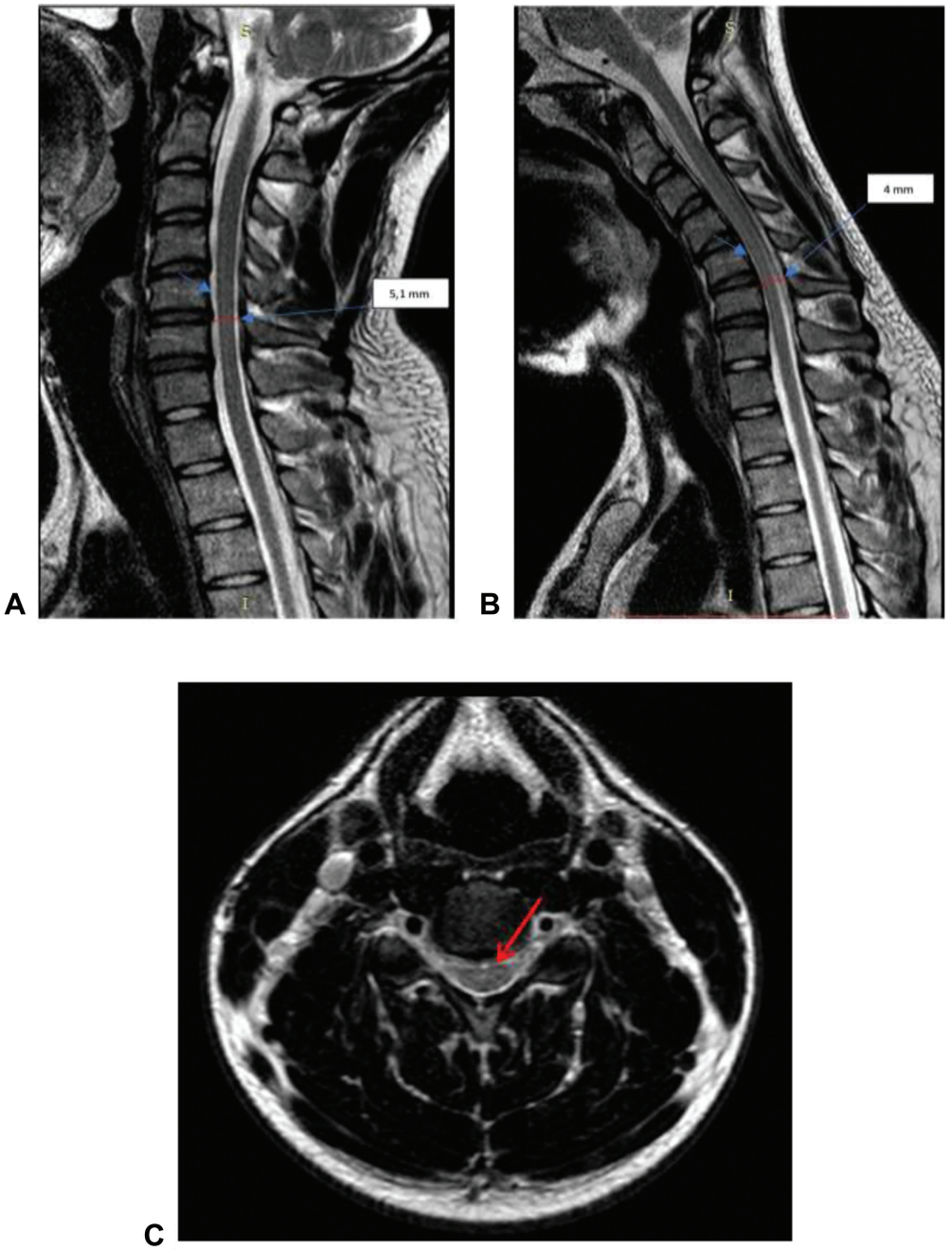

Fig. 2 Magnetic resonance imaging scans of the cervical spine of the patient in sagittal neutral position (**A**), in axial sagittal flexion (**B**) (**C**). T2-weighted images on sagittal views showing anterior displacement of the dura mater posterior to vertebral level C5-C6, suggesting an increase in mobility, which determines compression on the spinal cord. T2-weighted axial view (**C**) demonstrating asymmetric spinal cord flattening.

Financial Support
This present study received no financial support from public, commercial, or non-profit sources.

Conflict of Interests
The authors have no conflict of interests to declare.

References

1 Hirayama K. Juvenile muscular atrophy of distal upper extremity (Hirayama disease). Intern Med 2000;39(04):283–290
2 Tashiro K, Kikuchi S, Itoyama Y, et al. Nationwide survey of juvenile muscular atrophy of distal upper extremity (Hirayama disease) in Japan. Amyotroph Lateral Scler 2006;7(01):38–45
3 Vitale V, Caranci F, Pisciotta C, et al. Hirayama's disease: an Italian single center experience and review of the literature. Quant Imaging Med Surg 2016;6(04):364–373
4 Kandukuri GR, Acosta NR. Significance of Dynamic Imaging in Diagnosis of Hirayama Disease: A Rare Case Report and Literature Review. Kans J Med 2020;13(13):58–60
5 Hassan KM, Sahni H. Nosology of juvenile muscular atrophy of distal upper extremity: from monomelic amyotrophy to

Hirayama disease–Indian perspective. BioMed Res Int 2013; 2013:478516
6 Raval M, Kumari R, Dung AA, Guglani B, Gupta N, Gupta R. MRI findings in Hirayama disease. Indian J Radiol Imaging 2010;20 (04):245–249
7 Huang YL, Chen CJ. Hirayama disease. Neuroimaging Clin N Am 2011;21(04):939–950, ix–x
8 Tokumaru Y, Hirayama K. [Cervical collar therapy for juvenile muscular atrophy of distal upper extremity (Hirayama disease): results from 38 cases]. Rinsho Shinkeigaku 2001;41(4-5):173–178