Clinics in Surgery

Case Report
Published: 20 Apr, 2021

Hirayama Disease - A Not So Foreign Entity: Case Report and Literature Review

Yuh SJ[1], Guillon LL[2], Lalonde K[1], Boubez G[3], Wang Z[3] and Shedid D[1]*

[1]Department of Neurosurgery, Centre Hospitalier de l'Université de Montréal (CHUM), Canada

[2]Department of Radiology, Centre Hospitalier de l'Université de Montréal (CHUM), Canada

[3]Department of Orthopedics, Centre Hospitalier de l'Université de Montréal (CHUM), Canada

Abstract

Introduction: Hirayama disease, also known as juvenile muscular atrophy of the distal upper limb, is a rare disease that is characterized by an insidious-asymmetric onset of muscular atrophy and weakness in the distal upper extremities.

Case Report: Here we present a case in a young male of Canadian decent, who presented with severe atrophy of both forearms, sparring however the brachioradialis. Motor strength revealed weakness in all muscle groups in the upper extremities, more prominent in the distal muscle groups bilaterally. There was marked difficulty with prehension. He also found his walking to be more difficult.

Cervical flexion MRI demonstrate forward migration of the dura mater wall with an enlarged posterior epidural space, with disappearance of these large flow voids with the neck in neutral position.

Conclusion: Thought typically a benign self-limiting type of cervical myelopathy, early diagnosis of Hirayama disease is essential. It is important to appreciate the typical clinical and imaging features to accurately differentiate this rare disease from other neurological misdiagnoses. Progressive neurological and presence of myelomalacia should warrant aggressive surgical treatment as in our case.

Keywords: Hirayama disease; Cervical myelopathy; Surgery

OPEN ACCESS

Correspondence:
Sung-Joo Yuh, Department of Neurosurgery, Centre Hospitalier de l'Université de Montréal, Montréal, Québec, Canada, E-mail: syuh071@gmail.com

Received Date: *18 Mar 2021*
Accepted Date: *15 Apr 2021*
Published Date: *20 Apr 2021*

Citation:
Yuh SJ, Guillon LL, Lalonde K, Boubez G, Wang Z, Shedid D. Hirayama Disease - A Not So Foreign Entity: Case Report and Literature Review. Clin Surg. 2021; 6: 3137.

Introduction

Hirayama Disease (HD) was first described in 1959 as a cervical myelopathy variant [1]. Also known as juvenile muscular atrophy of the upper extremity [2], it is a rare neurologic condition that involves the inferior motor neurons, most commonly affecting the C7 to T1 spinal nerves and their myotomes.

Characterized by an insidious-onset [3], this disease affects mostly young male of Asian descent, typically diagnosed during the second and third decades of life [4]. There is notable wasting with weakness of the distal muscles of the upper extremity, sparing notably the brachioradialis (oblique amyotrophy). HD may affect C5 to C7 segmental myotomes more commonly in Western countries, whereas the C7 to T1 segment is predominantly affected in Asian countries [5].

Case Presentation

A 22-year-old male of French-Canadian descent, with a past medical history of poorly controlled type 1 diabetes since the age of 1 year old, presents with a slowly progressive weakness of his upper extremities, with a marked difficulty with prehension. He also describes a pins and needle sensation in his fifth digit on the left hand. He also found his walking to be more difficult, presenting with pyramidal symptoms of his lower extremities.

Physical exam demonstrates a very emaciated young male, with a severe atrophy of both forearms, sparring however the brachioradialis. Motor strength revealed weakness in all muscle groups in the upper extremities, more prominent in the distal muscle groups bilaterally. The muscular strength was graded as following (right/left): Brachioradialis and biceps 4/4, interossei and abductor digit minimi 2/2, flexor digitorum profundus 2/2, extensor digitorum communis 2/1, triceps 2/1, flexor pollicis longus 2/3, hip flexors 2/3, knee extensors 3/3, dorsiflexors 2/2, extensor hallucis longus 3/3. His deep tendon reflexes were graded as following: Biceps and supinator 2-/2-, triceps 0/0, patellar

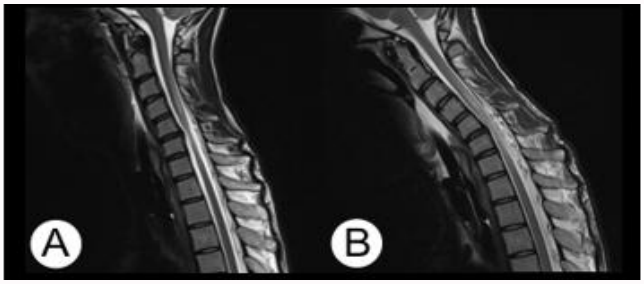

Figure 1: (A) Preoperative MRI neutral position. Preoperative MRI flexion position.

2/2+, Achilles 3/3. There was a slight up going toe on the right, and was indifferent on the left. There was sustained clonus on the right as well. Slight increase of the tone in the right lower extremity. Posterior column involvement was intact.

Imaging Study

Cervical X-ray

Static cervical X-ray (Figure 1A) did not reveal any malalignment, or any listhesis. Furthermore, dynamic cervical X-ray did not reveal any instability (Figure 1B, 1C).

Magnetic resonance imaging (MRI)

Magnetic resonance imaging of the cervical spine in neutral position (Figure 2A) revealed severe cord atrophy of the cord at the C5 to C6 level, with a T2 hypointense signal change. There was slight kyphosis noted as well. MR imaging in flexion (Figure 2B) revealed anterior displacement of the dorsal dura, with enlarged posterior epidural space.

Electrophysiologic study

Initial Electromyogram (EMG) demonstrated chronic moderate to severe denervation in the C7 and C8 distribution, sparring the C5 and C6 myotomes. Some irritability of the muscular fibers of C8 was noted, which may be related to partial reinnervation. This study was compatible with an inferior motoneuron disease of Hirayama type. However, the severity of the disease, as well as the cord signal (which is atypical for this disease) was atypical.

However, repeat EMG studies was compatible with a pure axonal sensory type of polyneuropathy that was slightly deteriorated. However, there was also possible diabetic polyneuropathy that was noted, fitting with the poorly controlled type I diabetes noted.

Treatment

Due to the severity of the clinical weakness at the initial presentation, the patient was offered urgent anterior cervical discectomy and fusion surgery to stabilize the spine. However, he initially refused hoping that his clinical status would stabilize on its own, following the natural course of the disease. As such, he was treated conservatively with an Aspen Vista Collar for a period of 12 months.

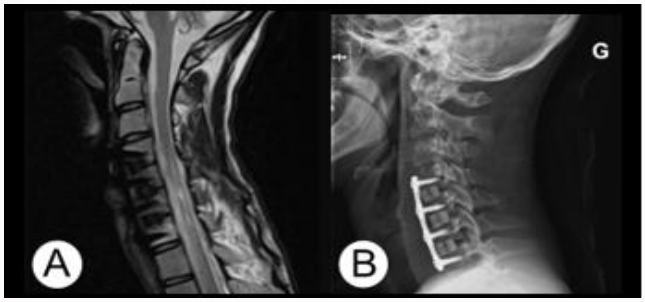

Figure 3: (A) Postoperative MRI neutral position. (B) Postoperative radiograph neutral position.

Unfortunately, on follow up, there was continued deterioration of his neurological function. There was continued marked weakness, till the point that he was unable to ambulate anymore.

As such, he underwent emergent multi-level anterior cervical discectomy at C4 to 5, C5 to 6 and C6 to 7 with instrumented fusion. There were no complications. His hospital stay was unremarkable. He was discharge to rehabilitation for a period of 3 months.

Postoperative imaging

Magnetic resonance: On repeat MRI (Figure 3A), there was no residual compression noted. There was persistence of the myelomalacia signal from C3 to T1 seen preoperatively.

Static cervical radiography of the cervical spine (Figure 3B) did not reveal any instrumentation abnormalities.

Discussion

Hirayama Disease (HD), also known as non progressive juvenile spinal muscular atrophy, is a form of juvenile muscular atrophy. It is a rare self-limiting cervical flexion myelopathy with insidious onset of progressive weakness and muscular wasting of the upper extremity [4,6]. It typically affects young individuals in their second to third decade predominantly in Japan and Asian countries.

The disease appears to be self-limiting; however, occurs in two phases with symptoms deteriorating in the first 1 to 3 years, followed by stabilization after about 5 years [7]. The initial symptoms of HD are slowly progressive hand weakness and fatigue, followed by cold paresis, tremors, and atrophy.

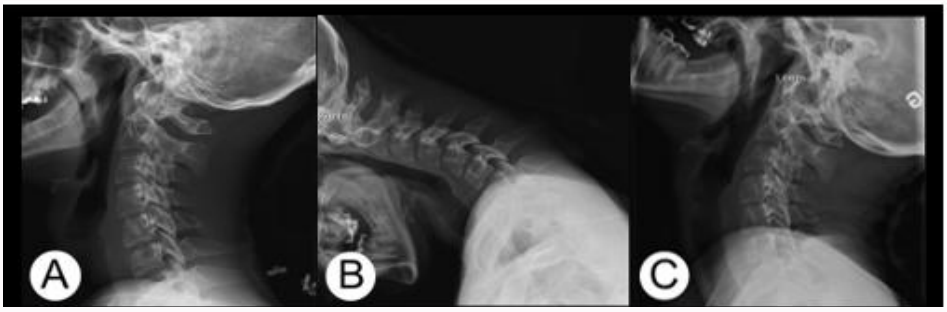

Figure 2: (A) Preoperative radiograph neutral position. (B) Preoperative radiograph flexion position. (C) Preoperative MRI extension position.

The important clinical characteristic is the unilateral or bilateral asymmetric weakness and wasting mainly involves the C7, C8 and T1 myotome [8]. And although the hand and forearms muscles are diffusely involved, the brachioradialis, belonging to C5 to C6 myotomes, is typically spared, giving rise to the characteristic 'oblique amyotrophy' of the forearm [2]. Typically, there is no objective sensory disturbance or lower limb involvement [9].

One theory explaining the pathophysiology of HD is the mechanical factor in flexion myelopathy, which leads to hemodynamic changes in spinal cord micro-circulation. Due to an imbalance between the growth of the vertebral column and that of the spinal canal content [10], there is forward displacement of the posterior dural sac with neck flexion [11], leading to compression and injury of the spinal cord with posterior damage to the anterior horn cells [4]. The compression causes microcirculatory changes in the territory of the anterior spinal artery usually at the C4 to C6 levels. These microcirculatory disturbances and ischemic changes in the cord lead to severe ischemic changes and gliosis at the site of the abutment [12]. Another possible underlying cause is the increased laxity of the dura mater, anchored superiorly at C2 to C3, allows for increased cord movement and concomitant anterior displacement with neck flexion, resulting in repetitive cervical microtrauma that leads to micro ischemia of the anterior horn cells [13]. Nonetheless, ischemic injury of the cervical anterior horn and/or nerve root caused by the excessive forward displacement of the posterior dura during neck flexion has become the main current hypothesis of the pathogenicity mechanism of HD [14].

Electromyogram is an important diagnostic tool that is important for distinguishing Hirayama disease from other similar disease such as ALS and multifocal motor neuropathy [5,15] where denervation and reinnervation confined to the upper limbs is also noted. However, in HD, the brachioradialis is denervated but the root of the C6 nerve is usually retained. The persistent denervation of the C7 to T1 myotomes is also noted [16]. Furthermore, both Compound Muscle Action Potential (CMAP) decrement to repetitive nerve stimulation and split-hand phenomenon quantified by CMAP amplitude were demonstrated to be useful in distinguishing ALS and HD, even in the early stages of the disease [17].

Imaging plays a very important role in the diagnosis of HD. Plain cervical radiograph may show only may only show loss of cervical lordosis, which is quite nonspecific. Conventional neutral position MR imaging may show an atrophied lower cervical cord and asymmetric cervical cord flattening with or without abnormal T2-weighted hyperintensities in the cervical cord, especially the anterior horn cells.

Dynamic cervical MRI in the flexed position demonstrates the classic finding of a forward shifting of the posterior dura mater wall with an enlarged posterior epidural space. There is disappearance of these large flow voids with the neck in neutral position [18]. There is also asymmetric cord flattening in the neutral position and forward displacement of the dura during flexion [19]. Post-gadolinium flexion MR images demonstrate moderate-to intense enhancement in the engorged posterior epidural venous plexus, forming a crescent-shaped epidural mass, which exerts a compression effect over the cord with or without flow voids [20]. Nonetheless, simple dura mater shifting on neck flexion is not enough to support an imaging-based diagnosis of Hirayama disease as this finding can also be seen in 46% of young asymptomatic controls [21].

The key debate for patients with Hirayama disease is how to proceed with treatment as it does have a relatively good prognosis compared to other motor neuron disease [22].

Conservative treatment, in the form of a cervical collar is typically employed to control progressive symptomatology by preventing the repetitive injury to the spinal cord with neck flexion. It has been shown to reverse disease progression, by avoiding repeat neck flexion during the progressive stage of the disease process [23]. Other mainstays for management are physical and occupational therapy.

Spinal decompression and instrumented fusion surgeries from either anterior or posterior approaches are beneficial by limiting dynamic mobility, which could help interrupt and progression of Hirayama disease [24]. Cervical duraplasty with laminoplasty is an alternative treatment for the disease, as it prevents the abnormal forward displacement of the posterior dura mater while preserving flexion of the cervical spine [14]. Nonetheless, surgery results in stabilization of symptoms only, except for one describing neurological improvements in only 3 out of 5 patients who underwent surgery [25]. As such, surgery should be offered to patients with particularly severe forms of the disease, presenting with severe spinal cord atrophy, amyotrophy extending to unusual segments (T1), highly active denervation with fasciculations, and clinical signs of pyramidal deficit.

Conclusion

In conclusion, we present a rare case report of "Hirayama disease" that is rarely encountered in clinical practice in the western world.

It should be part of the differential diagnosis for young male patients complaining of upper extremity weakness associated with muscle wasting, or in males presenting with unilateral or asymmetrical bilateral lower motor weakness of hands and fore-arms. It is a benign entity and no treatment other than cervical collar is usually needed in most cases. EMG can be used as an indicator to start and to stop cervical collar therapy.

Early recognition of this entity and differentiation from other causes of focal cord atrophy is important, because limitation of neck flexion by using a simple neck collar can usually prevent its further progression.

Electrophysiological examinations are indispensable for the diagnosis of HD, especially when differentiating it from other neurological disorders.

Accurate diagnosis and regular close clinical follow up are essential, as any rapid neurological deterioration should warrant aggressive surgical intervention to preserve neurological status.

References

1. Hirayama K, Tsubaki T, Toyokura Y, Okinaka S. Juvenile muscular atrophy of unilateral upper extremity. Neurology. 1963;13:373-80.
2. Hirayama K, Tokumaru Y. Cervical dural sac and spinal cord in juvenile muscular atrophy of distal upper extremity. Neurology. 2000;54(10):1922-6.
3. Ay H. Hirayama disease (monomelic amyotrophy) clinically confused for carpal tunnel syndrome. Neuropsychiatr Dis Treat. 2017;13:1385-8.
4. Foster E, Tsang BK, Kam A, Storey E, Day B, Hill A. Hirayama disease. J Clin Neurosci. 2015;22(6):951-4.
5. de Carvalho M, Dengler R, Eisen A, England JD, Kaji R, Kimura J, et

al. Electrodiagnostic criteria for diagnosis of ALS. Clin Neurophysiol. 2008;119(3):497-503.

6. Xu H, Shao M, Zhang F, Nie C, Wang H, Zhu W, et al. Snake-Eyes Appearance on MRI occurs during the late stage of hirayama disease and indicates poor prognosis. Biomed Res Int. 2019;2019:9830243.
7. Agundez M, Rouco I, Barcena J, Mateos B, Barredo J, Zarranz JJ. Hirayama disease: Is surgery an option? Neurologia. 2015;30(8):502-9.
8. Kuo YH, Kuo CH, Huang WC, Wu JC. Anterior cervical discectomy and fusion for hirayama disease: A case report and literature review. Neurospine. 2019;16(3):626-30.
9. Hirayama K. [Juvenile muscular atrophy of unilateral upper extremity (Hirayama disease)--half-century progress and establishment since its discovery]. Brain Nerve. 2008;60(1):17-29.
10. Kieser DC, Cox PJ, Kieser SCJ. Hirayama disease. Eur Spine J. 2018;27(6):1201-6.
11. Chen CJ, Chen CM, Wu CL, Ro LS, Chen ST, Lee TH. Hirayama disease: MR diagnosis. AJNR Am J Neuroradiol. 1998;19(2):365-8.
12. Chen CJ, Hsu HL, Tseng YC, Lyu RK, Chen CM, Huang YC, et al. Hirayama flexion myelopathy: Neutral-position MR imaging findings--importance of loss of attachment. Radiology. 2004;231(1):39-44.
13. Lay SE, Sharma S. Hirayama disease. StatPearls. Treasure Island (FL) 2020.
14. Ito H, Takai K, Taniguchi M. Cervical duraplasty with tenting sutures *via* laminoplasty for cervical flexion myelopathy in patients with Hirayama disease: Successful decompression of a "tight dural canal in flexion" without spinal fusion. J Neurosurg Spine. 2014;21(5):743-52.
15. Behnia M, Kelly JJ. Role of electromyography in amyotrophic lateral sclerosis. Muscle Nerve. 1991;14(12):1236-41.
16. Dejobert M, Geffray A, Delpierre C, Chassande B, Larrieu E, Magni C. Hirayama disease: Three cases. Diagn Interv Imaging. 2013;94(3):319-23.
17. Lyu RK, Huang YC, Wu YR, Kuo HC, Ro LS, Chen CM, et al. Electrophysiological features of Hirayama disease. Muscle Nerve. 2011;44(2):185-90.
18. Raval M, Kumari R, Dung AA, Guglani B, Gupta N, Gupta R. MRI findings in Hirayama disease. Indian J Radiol Imaging. 2010;20(4):245-9.
19. Lehman VT, Luetmer PH, Sorenson EJ, Carter RE, Gupta V, Fletcher GP, et al. Cervical spine MR imaging findings of patients with Hirayama disease in North America: A multisite study. AJNR Am J Neuroradiol. 2013;34(2):451-6.
20. Gandhi D, Goyal M, Bourque PR, Jain R. Case 68: Hirayama disease. Radiology. 2004;230(3):692-6.
21. Hou C, Han H, Yang X, Xu X, Gao H, Fan D, et al. How does the neck flexion affect the cervical MRI features of Hirayama disease? Neurol Sci. 2012;33(5):1101-5.
22. Aundhakar SC, Mahajan SK, Chhapra DA. Hirayama's disease: A rare clinical variant of amyotrophic lateral sclerosis. Adv Biomed Res. 2017;6:95.
23. Lee KH, Choi DS, Lee YS, Kang DH. Clinical experiences of uncommon motor neuron disease: Hirayama disease. Korean J Spine. 2016;13(3):170-2.
24. Huang YL, Chen CJ. Hirayama disease. Neuroimaging Clin N Am. 2011;21(4):939-50.
25. Lin MS, Kung WM, Chiu WT, Lyu RK, Chen CJ, Chen TY. Hirayama disease. J Neurosurg Spine. 2010;12(6):629-34.