Hirayama Disease

Jennifer Tynan, Evan Frangou, Christopher Voll, Rob Otani, Sheri Harder

Can. J. Neurol. Sci. 2010; 37: 703-705

A previously healthy 16-year-old female with no family history of neuromuscular disorders presented with a seven year history of hand incoordination. On examination, she had wasting of the intrinsic hand muscles bilaterally, right worse than left. There was no sensory loss, muscle cramping, or pain. Magnetic resonance imaging (MRI) of the cervical spine did not reveal any abnormal findings. Nerve conduction studies identified moderate slowing of the conduction velocity in the right ulnar nerve.

One year later, the patient's complaint of hand incoordination continued. Repeat nerve conduction studies revealed conduction blocks in both median nerves in addition to the previously noted decreased conduction velocity in the right ulnar nerve. Follow-up cervical spine MRI was performed (Figures 1, 2).

Magnetic resonance imaging in the neutral position demonstrated subjectively decreased volume and subtly increased T2 hyperintensity in the anterocentral portion of the cervical spinal cord from C5-7 (Figures 1, 2). The cord signal change was non-specific. Imaging obtained in the flexed position elicited severe spinal canal stenosis most marked at C6-7 with a maximal midline anteroposterior diameter of the spinal canal of 6 mm. Prominent flow voids in the posterior epidural space were suggestive of epidural venous engorgement.

Discussion

Hirayama disease (HD) was first reported in Japan by Dr. K. Hirayama in 1959 and initially labeled "juvenile muscular atrophy of a unilateral (later distal) upper extremity."[1] Hirayama disease is sporadic juvenile muscular atrophy of the distal upper extremities, which predominantly affects young men between 15 and 25 years-of-age. Hirayama disease is characterized by insidious onset of unilateral muscular atrophy and weakness of the hand and forearm, in the absence of sensory or pyramidal signs. Bilateral presentation, though less common, is also known to occur.[2] The clinical course is initially progressive for six to nine years, followed by spontaneous stabilization by the middle of the third decade.[1] This disease has had a number of other names some of which include brachial monomelic amyotrophy, benign focal atrophy, benign focal amyotrophy, Sobue's disease, and juvenile segmental muscular atrophy.[1]

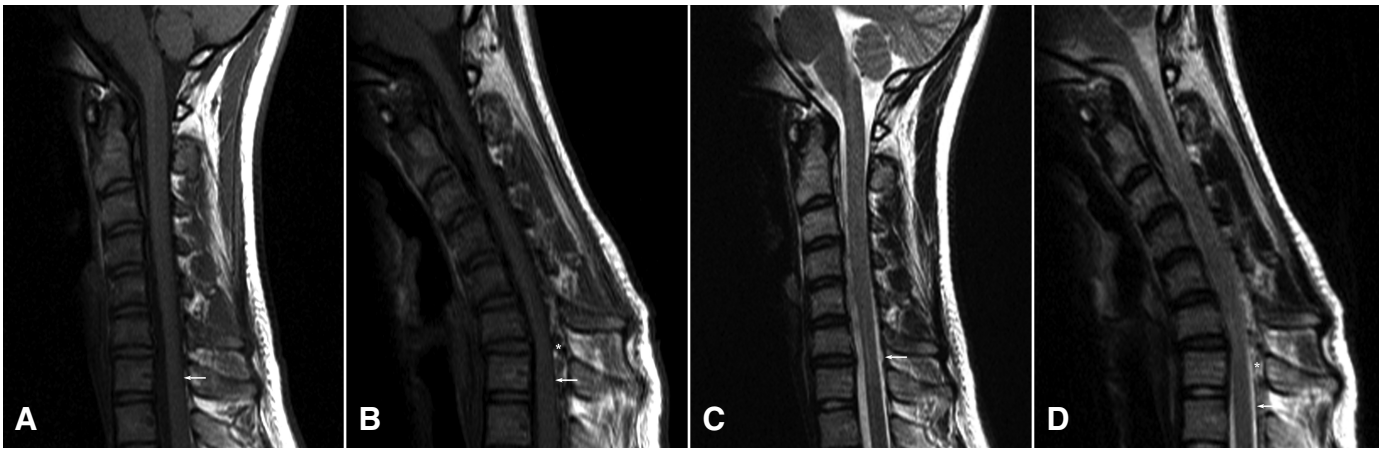

Figure 1: *A - D: Sagittal T1 (TR = 677; TE = 15) and T2 (TR = 2880; TE = 101-105) weighted images in neutral (A, C) and flexed (B, D) positions. Severe spinal canal stenosis and decreased volume in the mid to lower cervical spinal cord is seen in flexion. * denotes enlarged posterior epidural space with venous engorgement. Arrow indicates dura.*

From the Academic Department of Medical Imaging (JT, RO), Division of Neurosurgery, Department of Surgery (EF), Division of Neurology, Department of Medicine (CV), University of Saskatchewan, Royal University Hospital, Saskatoon, Saskatchewan, Canada; Department of Radiology (SH), Loma Linda University Medical Center, Loma Linda, California, USA.

Received November 26, 2009. Final Revisions Submitted March 4, 2010.

Correspondence to: Evan Frangou, Division of Neurosurgery, Royal University Hospital, 103 Hospital Drive, Saskatoon, Saskatchewan, S7N 0W8, Canada.

https://doi.org/10.1017/S0317167100010969 Published online by Cambridge University Press

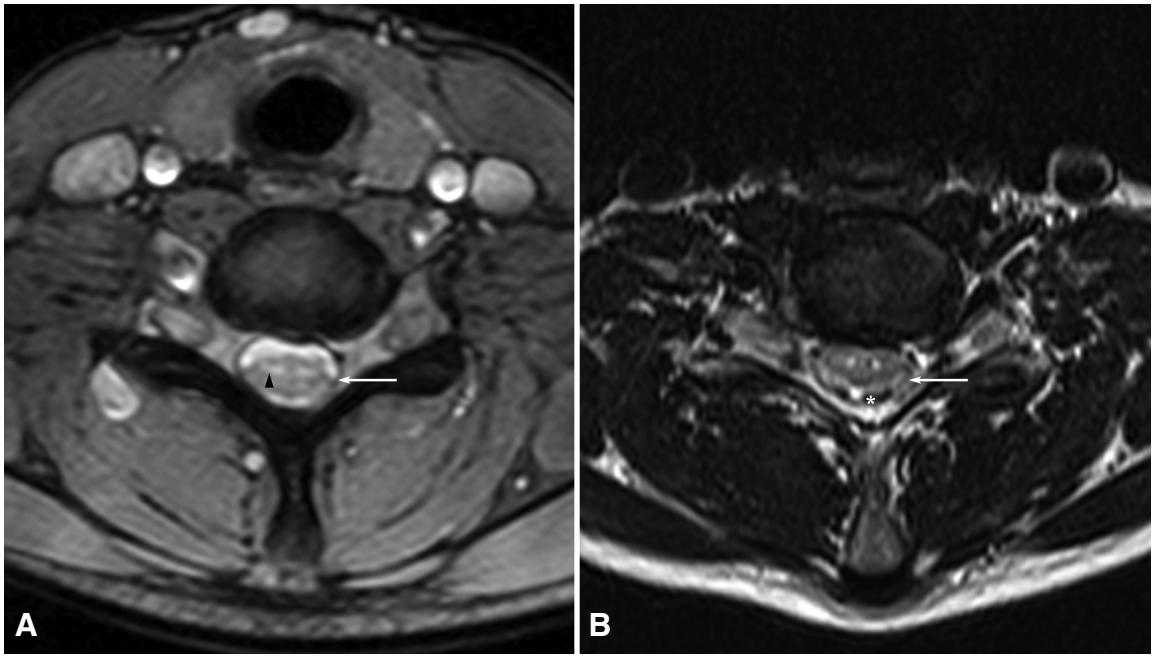

Figure 2: *A) Axial multiple-echo data image combination (MEDIC) (TR = 794; TE = 24) in neutral position. Abnormal intramedullary increased T2 signal intensity is seen within both the left and right (arrow head) anterocentral portions of the cord in the expected location of the anterior horns. B) Axial T2 (TR = 5000; TE = 96) in flexed position. * indicates posterior epidural space enlargement by venous engorgement. Arrow indicates dura.*

The pathogenesis of HD is unclear and has been attributed to several factors, including trauma, ischemia, autoimmunity, familial or hereditary factors, toxins, and infection.[1,3] Japanese investigators have suggested that HD may be due to microvascular changes following chronic trauma to the spinal cord during neck flexion, especially during adolescence when there is rapid growth.[4] However, not all studies have confirmed this theory and some authors have suggested that HD cannot be explained by the neck flexion mechanism alone.[1] Konno and coworkers postulated another mechanism whereby an inelastic dura constricts and compresses the cervical spinal cord when the neck is in either a neutral or a flexed position.[5] Cord ischemia at C5-T1, in the territory of the anterior spinal artery, is another proposed mechanism; however, the absence of corticospinal tract involvement opposes this hypothesis.[6] Currently, there is no consensus regarding the specific pathogenesis of this disease. Nonetheless, diagnostic criteria of HD have been established (Table).[1,6]

Magnetic resonance imaging can be very useful in the diagnosis of HD. Conventional radiographic studies of the cervical spine generally do not demonstrate any specific abnormalities apart from straight alignment or scoliosis.[7] Myelography can demonstrate the forward movement of the posterior dural wall when the neck is flexed. However, this procedure is difficult to perform as the contrast medium is expelled from the cervical subarachnoid space when the neck is flexed, regardless of patient position.[7]

Like myelography, MRI in neck flexion can show anterior displacement of the posterior dural wall.[4,7-9] In addition, the posterior epidural space of the lower cervical canal becomes enlarged with flexion and is seen as a crescent of high signal intensity on T1 and T2 weighted MRI, with variable epidural flow voids.[4,7] In normal individuals, the epidural venous plexus mostly lies in the anterior and anterolateral epidural space and is unremarkable posteriorly. The anterior plexus is non-distensible under normal venous pressure while the posterior system may have some engorgement with increases in pressure. In patients with HD, the posterior epidural space not only increases in size during flexion, but there is also uniform enhancement on contrast administration, suggesting increased vascularity in this space. These findings suggest that dilated blood vessels occur in the posterior epidural space as a result of negative pressure created by forceful forward movement of the dural sac during neck flexion.[7,9] Compressive flattening of the spinal cord also accompanies the forward shifting of the posterior dura, and in the majority of cases spinal cord flattening is asymmetric.[2,7,8] At late stages of the disease, MRI has occasionally documented asymmetric atrophy of the distal cervical spinal cord.[8]

While a diagnosis of HD on flexion MRI is straightforward, diagnosis on neutral-position MRI is more challenging.[7,10] Chen and coworkers have identified loss of attachment (LOA) between the posterior dural sac and subjacent lamina as the most effective finding in the diagnosis of HD, with sensitivity and specificity greater than 93.5%. Asymmetric cord atrophy, especially at the lower cervical cord, is another neutral-position MRI finding with 59% sensitivity and 100% specificity.[10] Thus, in patients with adolescent onset of distal upper limb weakness, the LOA sign on neutral-position MRI should raise suspicion for

https://doi.org/10.1017/S0317167100010969 Published online by Cambridge University Press

Table: Diagnostic criteria for Hirayama disease[1,6]

1. Distal predominant weakness clinically confined to one limb although electromyelogram studies may demonstrate clinically asymptomatic contralateral neurogenic findings.
2. Almost always involves one upper extremity.
3. Insidious onset between age 10 to early 20s.
4. Gradual progression for a few years, with subsequent stabilization.
5. No lower-extremity involvement.
6. No sensory symptoms or findings.
7. Exclusion of other disorders.

HD and a flexion MRI study should be performed to confirm the diagnosis.[10] Early detection of this disease is essential as avoidance of neck flexion has been shown to prevent disease progression.[8]

The differential diagnosis for these findings may include vascular malformations, tumors or other pathology. However, posterior venous engorgement on flexion which dissipates in neutral position is not seen in other pathologies and, is generally considered pathognomonic for HD.[7,11]

Current treatment options are somewhat controversial. Wearing a soft cervical collar may prevent the progression of symptoms.[12] Observation and surgical decompression have also been attempted, however no treatment has been extensively studied. The majority of patients show a spontaneous arrest of progression within three to five years.[13]

In summary, patients with symptoms of HD should be considered for flexion MRI, even if neutral-position imaging appears normal. Magnetic resonance imaging should be performed in neutral as well as completely flexed neck positions to detect abnormalities. Flexion MRI studies demonstrate the pathognomonic picture of anterior shifting of posterior dura at the lower cervical spinal canal with engorgement of the posterior epidural venous complex. Also, in non-flexion MRI the finding of asymmetric cord atrophy or LOA, especially at the lower cervical cord, is highly suggestive of Hirayama disease and should prompt a flexion MRI study to support the diagnosis. Treatment with soft collar may be considered.

References

1. Patel DR, Knepper L, Jones HR, Jr. Late-onset monomelic amyotrophy in a Caucasian woman. Muscle Nerve. 2008;37(1): 115-19.
2. Hirayama K. Non-progressive juvenile spinal muscular atrophy of the distal upper limb (Hirayama's disease). In: De Jong J, editor. Handbook of clinical neurology. Amsterdam: Elsevier; 1991. p. 107-20.
3. Misra UK, Kalita J, Mishra VN, Kesari A, Mittal B. A clinical, magnetic resonance imaging, and survival motor neuron gene deletion study of Hirayama disease. Arch Neurol. 2005;62(1): 120-3.
4. Hirayama K, Tokumaru Y. Cervical dural sac and spinal cord in juvenile muscular atrophy of distal upper extremity. Neurology. 2000;54(10):1922-6.
5. Konno S, Goto S, Murakami M, Mochizuki M, Motegi H, Moriya H. Juvenile amyotrophy of the distal upper extremity: pathologic findings of the dura mater and surgical management. Spine (Phila Pa 1976). 1997;22(5):486-92.
6. Tashiro K, Kikuchi S, Itoyama Y, Tokumaru Y, Sobue G, Mukai E, et al. Nationwide survey of juvenile muscular atrophy of distal upper extremity (Hirayama disease) in Japan. Amyotroph Lateral Scler. 2006;7(1):38-45.
7. 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.
8. Gandhi D, Goyal M, Bourque PR, Jain R. Case 68: Hirayama disease. Radiology. 2004;230(3):692-6.
9. Pradhan S, Gupta RK. Magnetic resonance imaging in juvenile asymmetric segmental spinal muscular atrophy. J Neurol Sci. 1997;146(2):133-8.
10. 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.
11. Kikuchi S, Tashiro K, Kitagawa M, Iwasaki Y, Abe H. [A mechanism of juvenile muscular atrophy localized in the hand and forearm (Hirayama's disease)--flexion myelopathy with tight dural canal in flexion]. Rinsho Shinkeigaku. 1987;27(4):412-9.
12. Tokumaru Y, Hirayama K. [A cervical collar therapy for non-progressive juvenile spinal muscular atrophy of the distal upper limb (Hirayama's disease)]. Rinsho Shinkeigaku. 1992;32(10): 1102-6.
13. Hirayama K. Juvenile muscular atrophy of distal upper extremity (Hirayama disease). Intern Med. 2000;39(4):283-90.

https://doi.org/10.1017/S0317167100010969 Published online by Cambridge University Press