A severe case of Hirayama disease successfully treated by anterior cervical fusion

Case report

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Hirayama disease, or juvenile amyotrophy of distal upper extremity, is a benign, self-limiting cervical myelopathy consisting of selective unilateral weakness of the hand and forearm. The weakness slowly progresses until spontaneous arrest occurs within 5 years of onset. The condition predominantly affects Asian males and is thought to be secondary to spinal cord compression during neck flexion, because of a forward displacement of the posterior dural sac.

The authors present what is to their knowledge the first reported case of a Caucasian male with a severe form of Hirayama disease, suffering from weakness of the leg as well as the forearm. An abnormal range of cervical flexion was observed at the C5–6 level. The patient was successfully treated by anterior cervical discectomy and fusion. (http://thejns.org/doi/abs/10.3171/2013.10.SPINE13508)

Key Words • Hirayama • juvenile amyotrophy of distal upper extremity • cervical flexion myelopathy • cervical flexion MRI • discectomy and fusion • abnormal range of cervical flexion

Juvenile amyotrophy of distal upper extremity, also known as Hirayama disease, is a benign, self-limiting cervical myelopathy first brought to attention by Hirayama11 in 1959. Over the past 50 years, with the advent of MRI, several researchers have established this illness as a different entity from the motor neuron diseases.6–10,13,17,25,26 It has been characterized not as a primary disease of motor neurons, but as a myelopathy secondary to spinal cord compression during cervical flexion.

The clinical features of Hirayama disease are: 1) asymmetrical weakness of the distal upper extremities, with atrophy of the thenar and hypothenar eminence and relative sparing of the brachioradialis muscle (oblique amyotrophy); 2) cold paresis (worsening of the weakness and distal tremor with cold); 3) lack of sensory, autonomic, or cranial nerve signs or symptoms; 4) insidious onset at the second to third decades of life (15–25 years), predominantly in males, with slow worsening over a variable period of time (mean 5 years)31 and subsequent spontaneous arrest of progression; and 5) usually sporadic occurrence (although familial cases have been reported).1,3,14,23

Radiological findings are essential for the diagnosis of Hirayama disease. Neutral and flexion cervical MRI shows asymmetrical atrophy of the spinal cord, forward displacement of the posterior dural sac with neck flexion, and thus, secondary cord compression against the posterior wall of the vertebral body.6,15,16,21,27,30

Since neck flexion was recognized as a possible cause of the disease, several therapies have proved their utility, shortening the progression period and even improving the patient’s strength. Wearing a cervical collar is the most common conservative therapy. For cases in which the disease progresses despite conservative treatment, several surgical options have been used: posterior decompression with dural sac augmentation,2 with and without fusion, anterior decompression and fusion, and combinations of these methods. Which is the most effective is still a matter of debate.

We are presenting a case of Hirayama disease with lower-extremity signs and symptoms, which was successfully treated with anterior discectomy and fusion alone.

Case Report

A previously healthy 19-year-old Caucasian male presented with a 2-year history of insidious progressive
weakness of the right distal upper limb. The weakness was already stable, and it had not progressed over the last several months. He reported a slow progressive weakness of the proximal lower ipsilateral limb, and this was what had brought him to consult a neurologist. He also reported that his condition worsened with the cold. There was no family history of neuromuscular disease. At physical examination, oblique atrophy was obvious (atrophy of the thenar and hypothenar eminences and of the cubital border of the forearm, with relative sparing of the brachioradialis muscle), and the patient had severe weakness of the intrinsic musculature of the hand (0/5). His wrist flexion and extension and his elbow extension were also affected. He had mild weakness of hip flexion and knee extension in the right lower limb but was still able to walk. The patient’s deep tendon reflexes were normal in the upper limb and the left foot, but were brisk in the affected lower limb. He had a positive Babinski sign on the right. No sensory alterations were present, and the cranial nerves were unaffected. No other abnormalities were found on physical examination.

Complete blood counts, blood chemistry analysis, and urinalysis revealed nothing unusual. Electromyography (EMG) showed acute over chronic denervation of the right interossei dorsalis manus muscles and chronic reinnervative changes in the left abductor pollicis brevis, flexor pollicis longus, and flexor digitorum profundus. The ulnar and median nerve conduction velocities were within normal limits. EMG over the tibialis anterior muscle showed mild acute denervation changes, such as spontaneous potentials, without conduction velocity abnormalities.

Cervical MRI in a neutral neck position depicted a normal-size spinal canal, with a loss of normal cervical lordosis. The spinal cord showed atrophic changes at the C5–6 level, with signal hyperintensity on T2-weighted images extending 1 level above and below (Fig. 1). Loss of attachment of the posterior dura was not seen on any image. Images obtained with the neck flexed showed forward displacement of the posterior dural sac and excessive angulation of the spine at C5–6 level, causing compression of the cord (Fig. 2). The forward displacement of the dural sac created a crescent-shaped space between the dura and the laminae, with a large flow void. This space enhanced strongly after gadolinium contrast administration. After considering the findings, a severe form of Hirayama disease was diagnosed.

Considering the clinical course of the disease and that a conservative treatment had been tried unsuccessfully at the referring hospital, the decision was made to treat the patient surgically. Based on the excessive angulation observed at the C5–6 level (Fig. 2), an anterior C5–6 discectomy was performed, a polyetheretherketone (PEEK) cage was placed, and a plate was screwed to the vertebral bodies, completing an anterior fusion procedure. The total surgical time was less than 50 minutes, and the blood loss was insignificant. The patient’s postoperative course was uneventful. The neurologic examination findings were unchanged, and the patient was discharged 48 hours after surgery. The disc was sent to the pathology laboratory, and no abnormality was found.

Two months after the procedure the patient presented with subjective improvement of his lower leg strength. A cervical MRI with the neck flexed and extended was performed 1 month after the procedure (Fig. 3). The posterior dural sac no longer moved forward with neck flexion, and
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there was no contact between the spinal cord and posterior wall of the vertebral bodies; thus, no compression was observed. Six months after the surgery a CT scan was performed, showing bone bridge formation at the surgically treated level (Fig. 4). As of this writing, a year after the surgery, the patient’s leg has regained its full strength, gait is normal, and upper limb is unchanged. EMG shows no abnormalities in the lower extremities, but does show chronic denervation changes in the upper limb.

Discussion

Hirayama disease was initially described in 1959 by K. Hirayama.11 The author described it as a monomyelic disease, affecting only one limb, without impairment of sensory pathways or cranial nerve or brain function. However, the largest series reported in the English literature31 describes electrophysiological studies depicting a bilateral impairment (although asymmetrical) in up to 95% of the cases, and a lower limb impairment in 3 of 195 cases. Nevertheless, patients presenting with bilateral paresis are rare, and patients presenting with lower ipsilateral limb paresis are even rarer.24 In fact, to the best of our knowledge, this is the first report of a Caucasian male with this severe form of Hirayama disease.

The first autopsy of a patient with Hirayama disease was performed in 1982 by Hirayama,8 and the findings suggested chronic ischemic injury to the anterior horn of the cord. He described the condition as a flexion myelopathy and proposed a misbalance between the growth of the spinal cord and the spine as the cause. The anterior horn is the area most vulnerable to ischemic insults; thus it was proposed that chronic repetitive compressions of the cord due to the forward displacement of the posterior dural sac during neck flexion caused perfusion impairment and permanent injury. The male predominance would support the idea of a misbalance in growth, because the growth spurt is usually more marked in males than in females, and the typical age at onset of Hirayama disease is 2 years after the beginning of the growth spurt. The observation that in older patients, once the progression is arrested, the forward shifting of the dura is no longer appreciated supports it as a causative factor.

Evidence from EMG studies helps to differentiate Hirayama disease from other motor neuron diseases. EMG typically shows the presence of neurogenic lesions in all affected muscles with spontaneous potentials, a prolonged duration or augmentation of amplitude in motor unit potentials, or an incomplete pattern of recruitment.3,18–20,25 Electrophysiological studies performed during neck flexion failed to prove abnormalities related to...
neck posture,

The diagnosis of the disease is strongly based on MRI findings in the flexed position of the neck. There are some subtle changes in the neutral MRI that should raise the suspicion of the condition, as described by Huang and Chen: asymmetrical atrophy of the cord and loss of attachment of the posterior sac to the lamina.

It is also important to note that the changes in the flexed position vary with the degree of flexion, and they might not be remarkable with the neck flexed less than 25°. Even though the forward shift of the dura in the flexed position is the most striking finding, it might be found in up to 40% of healthy young males, so the diagnosis cannot be made exclusively on this basis. It is also common to find large flow voids in the crescent-shaped space created due to the forward shifting of the dura. Those flow voids correspond to an engorged venous epidural plexus. Patel et al. reported a case in which venography was performed and the epidural pressure was monitored. The lack of pressure change with neck flexion suggested that the engorgement of the plexus was a passive event rather than a causative factor.

Xu et al. have recently shown that there is a marked abnormality in the range of motion of the cervical spine in patients with Hirayama disease, both globally and by segment. They compared findings in 31 patients with Hirayama disease with findings in 40 healthy subjects of the same age. They found that the angle of flexion was greater in the patients with Hirayama disease than in the controls, especially at the C5–6 level, where they found a mean angulation of 11.95° compared with the 5.36° that was the mean value obtained in the healthy subjects. (To measure the angle an imaginary line is formed by linking the middle point of the posterior and anterior walls of the vertebral body. The angle is measured where the extended lines of adjacent levels cross.) Our patient had an angulation of 11.78° at C5–6, 6.1° at C4–5, and 5.8° at C3–4 (Fig. 2). This finding drove us to operate on the C5–6 segment, and the good result obtained suggests that an excessive range of motion of the cervical spine could be a causative factor in the disease.

Treatment of Hirayama disease is a controversial issue. Because the progression will arrest spontaneously within 5 years of symptom onset in the majority of cases and the use of a cervical collar has been shown to shorten that period, some authors advocate conservative treatment. In some cases, however, the patient’s condition worsens despite the use of the cervical collar. Moreover, the use of a cervical collar has only proven effective in stabilizing the patient’s condition, not improving it. Therefore, an increasing number of physicians advocate surgical treatment. Anterior and posterior decompressive procedures—with or without duraplasty and with or without fusion—have been used, producing arrest of the progression and even improvement of patients’ previous symptoms. However, a randomized controlled trial is lacking, and no recommendations about the type of surgery, its timing, or even its adequacy can be made. The good result obtained in our case suggests that an excessive range of flexion of the cervical spine might play a key role in the pathophysiology of the disease. Thus, a simple and safe anterior fusion procedure could be the best choice.

Conclusions

We present what is to our knowledge the first case of a Caucasian male with a severe form of Hirayama disease. The disease presented as stabilized paresis of the right upper limb and progressive paresis of the right lower limb. The patient was treated with anterior disectomy and fusion. His lower limb function improved, with resolution of symptoms in that limb, and his upper limb function stabilized. Therefore we believe that anterior fusion might be the best choice of treatment.

Disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author contributions to the study and manuscript preparation include the following. Conception and design: Paredes. Acquisition of data: Paredes, Esteban, Ramos. Analysis and interpretation of data: Paredes, Rivas. Drafting the article: Paredes. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Paredes.

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