CASE REPORT

A Portuguese case of Hirayama disease

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Dr Mário Correia de Sá,
mario.s.sa@gmail.com**SUMMARY**

Hirayama disease, also known as monomelic amyotrophy or juvenile spinal muscular atrophy of the distal upper extremity features the impairment of the anterior horn cells of the distal cervical spinal cord secondary to dural sac anterior displacement during cervical flexion. We describe a case of a 17-year-old boy with a history of scoliosis, evaluated in the emergency department for decreased muscle strength and atrophy of the left upper limb with progressive worsening for about 6 months. We performed electrophysiological studies that showed severe neurogenic atrophy involving the C7–T1 left myotomes. Brain and spine MRI performed showed flattening of the lower cervical cord and dura mater anterior displacement during cervical flexion. These findings were consistent with the diagnosis of Hirayama disease.

BACKGROUND

Hirayama disease (HD), or monomelic amyotrophy, or juvenile spinal muscular atrophy of the distal upper extremity features the impairment of the anterior horn cells of the distal cervical spinal cord (SC) secondary to microcirculatory changes within the anterior spinal artery territory presumably caused by the anterior displacement of the dural sac during cervical flexion.

This entity, first described in 1959, primarily affects men, teenagers or young adults and is associated with muscle atrophy and decreased muscle strength of the distal upper limb usually unilateral or asymmetric, with slow or none progression after the initial worsening phase.

Most cases were reported in Asian countries, however, several cases have been described in Europe and in the American continent.

In recent years, a significant relationship between this disease and the presence of elevated serum IgE was demonstrated and these patients showed more severe clinical course compared with those with normal serum IgE levels.

CASE PRESENTATION

A 17-year-old boy, with a history of scoliosis was referred to the emergency department for decreased muscle strength of the left hand associated with left upper limb atrophic changes progressively worsening over the past 6 months. History of trauma or a particularly relevant allergic disease was denied.

The physical examination showed thoracic asymmetry with right hemithorax prominence and scoliotic posture. The left upper limb showed atrophy of all the muscle groups and marked asymmetry when compared with the right limb.

On neurological examination, the teenager presented with decreased left upper limb muscle strength noted in the *Barré* test and during grip. The tendon reflexes were normal with bilateral flexor plantar reflex. The patient showed no changes on cranial nerves examination or sensory or gait disturbances.

The analytical evaluation consisting of complete blood count, serum glucose, renal function, liver function, lactate dehydrogenase and creatine kinase was normal. The determination of the serum IgE was elevated (200.6 IU/mL).

Electromyography showed signs of chronic neurogenic injury involving the C7–T1 myotomes bilaterally and more severely in left C8 and T1. The somatosensory evoked potentials showed normal results.

Brain and spine MRIs were performed and showed lower cervical SC thinning, between C5 and C7, more marked on the left side (figures 1 and 2) and, during cervical flexion, dural sac anterior displacement, from C3 to C7 (figure 3).

In this position, the epidural space presented a contrast uptake, suggesting engorgement of the epidural venous plexus thereby compromising the intradural space.

Physical therapy was started immediately as was interdisciplinary discussion about other therapeutic options. The clinical, radiological and electrophysiological findings were compatible with the diagnosis of HD.

Figure 1 T2-weighted sagittal image in the neutral position reveals lower cervical spinal cord thinning between C5 and C7 (arrow).

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Figure 2 T2-weighted axial image in the neutral position shows medullary atrophy at the left aspect of the cord (arrow).

DISCUSSION

HD was first described in 1959 by K Hirayama as juvenile muscular atrophy of the distal upper extremity. Since then, several similar cases were reported not only from Asian countries like Japan, India and Sri Lanka, but also, like the present, from European or American countries. The authors believe that the case here reported is the first described in Portugal.

The causes that account for the geographical variability in the number of reported cases are still unknown. Hypotheses that could explain this discrepancy are the presence of genetic or environmental factors that predispose to the disease in certain populations and/or the lack of awareness of this disease by many non-Asian physicians resulting in the underdiagnosis of HD, which increases the relevance of this case report.

This entity has maximum prevalence in adolescents and young adult males (male : female ratio 20 : 1).¹ It has an insidious presentation and the amyotrophy is unilateral in most cases, sometimes asymmetric and rarely symmetrical.^{1 2} Weakness develops predominantly in the extensor and flexor muscles of

Figure 3 Contrast-enhanced flexion sagittal T1-weighted MRI shows enhancement of the epidural space (arrow) suggesting engorgement of the epidural venous plexus thereby compromising the intradural space.

the fingers and the wrist sparing the brachioradialis muscle.¹ It has a slow progressive initial course followed by spontaneous arrest after a maximum of 5 years.

For an HD definitive diagnosis a spinal MRI is essential in neutral as well as during cervical flexion. MRI shows thinning of the SC in its lower cervical segment associated, during cervical flexion, with anterior displacement of the dural sac and enlargement of the epidural venous plexus^{3 4} shown as a well-enhanced crescent-shaped mass after contrast administration. It is believed that these findings represent engorgement of the posterior vertebral venous plexus since they are not detected when the patient is in cervical neutral position.

Electromyographic studies should also be conducted since the findings of acute or chronic denervation of the affected muscles support the HD diagnosis.

Ito *et al*⁵ described a significant correlation between the presence of elevated serum IgE and HD and that these patients tend to have more severe disabilities than those with normal IgE levels. Changes in the connective tissue components of the dural sac in patients with elevated IgE levels may be related to these findings but more studies are required.

HD's pathogenic mechanism is still under discussion but it is believed that it is secondary to compression of the lower cervical SC by the posterior dural sac on neck flexion. This repeated injury causes increased intramedullary pressure and microcirculatory changes within the anterior spinal artery territory.⁶ It is also speculated that the disproportionate growth between the vertebral column (and its dural sac) and SC during adolescent growth spurt is associated with this disease.⁷ Other authors believe that HD is explained by different mechanisms including the presence of an inelastic dural sac compressing the SC in neutral position as well as during cervical flexion.⁸

The primary principle of the treatment includes avoidance of long-standing cervical flexion positions including the use of high pillows that cause significant cervical flexion. The use of a neck collar may not only stop HD's progression but also, in certain cases, may lead to partial paresis regression.⁹

Learning points

- It is important to perform a spine MRI in cervical flexion in patients with symptoms consistent with HD even if the spine MRI in neutral shows no abnormalities
- More studies are needed to determine the role of elevated IgE in the pathophysiology of HD.
- HD's early diagnosis will permit early treatment thus allowing disease's control or even regression and may also prevent unnecessary investigations.

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REFERENCES

- 1 Hirayama K. Juvenile muscular atrophy of distal upper extremity (Hirayama disease). *Intern Med* 2000;39:283–90.
- 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;7:38–45.

- 3 Raval M, Kumari R, Dung AA, *et al.* MRI findings in Hirayama disease. *Indian J Radiol Imaging* 2010;20:245–9.
- 4 Yin B, Liu L, Gent D-Y. Features of Hirayama disease on fully flexed position cervical MRI. *J Int Med Res* 2011;39:222–8.
- 5 Ito S, Kuwabara S, Fukutake T, *et al.* Hyperlgaemia in patients with juvenile muscular atrophy of the distal upper extremity (Hirayama disease). *J Neurol Neurosurg Psychiatry* 2005;76:132–4.
- 6 Hirayama K, Tokumaru Y. Cervical dural sac and spinal cord in juvenile muscular atrophy of distal upper extremity. *Neurol* 2000;54:1922–6.
- 7 Kohno M, Takahashi H, Yagishita A, *et al.* 'Disproportion theory' of the cervical spine and spinal cord in patients with juvenile cervical flexion myelopathy. A study comparing cervical magnetic resonance images with those of normal controls. *Surg Neurol* 1998;50:421–30.
- 8 Konno S, Goto S, Murakami M, *et al.* Juvenile amyotrophy of the distal upper extremity: pathologic findings of the dura mater and surgical management. *Spine (Phila Pa 1976)* 1997;22:486–92.
- 9 Tokumaru Y, Hirayama K. [Cervical collar therapy for juvenile muscular atrophy of distal upper extremity (Hirayama disease): results from 38 cases]. *Rinsho Shinkeigaku* 2001;41:173–8.

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