Hirayama's Disease

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Hirayama's disease, also known as juvenile non-progressive cervical amyotrophy or monomelic amyotrophy is a rare focal motor neuron disease that primarily affects young Asian males (15-25 years old). The first case was reported by Hirayama in 1959. The patient often has insidious and slowly progressive weakness, followed by a spontaneous arrest within several years. Even though its etiology has not been clearly identified, the hypothesis of dynamically chronic compression of cervical spinal cord was proposed. This is demonstrated by magnetic resonance imaging (MRI) of the cervical spine which shows forward displacement of the posterior wall of the lower cervical dural canal. It leads to marked, often asymmetric, flattening of the lower cervical cord. Early diagnosis is key to successful management. Surgical decompression in some cases and physical therapy are the treatment options.[1-3] This is a report of a young Thai adult, presenting as a classic case of Hirayama disease.

Case Report

A 20-year-old man presented with two years' history of weakness and tremor in both hands (more severe on the right side). The symptoms progressed slowly over the years and seemed to accelerate for a period of six months. He also noticed mild tremor of both hands while holding objects or using his hand in fine motor tasks. There was no sensory or bulbar symptoms, accident or other preceding illnesses. Neurological examination showed mild atrophy and weakness of the right triceps, both forearm and hand muscles which were more affected on the right side (Figure 1). The hand grip was also mildly weak on both sides. Rare fasciculations were found at the right forearm. Irregular and coarse postural tremors, especially with finger extension, were present on both sides. Cranial nerve, sensations and reflexes were normal.

Nerve conduction studies revealed normal median, ulnar and radial nerves, in both motor and sensory components. Electromyography (EMG) showed few positive sharp waves and fibrillation potentials at the right triceps, extensor digitorum communis and first dorsal interosseous muscles. Several very large and polyphasic motor units with reduced interference pattern (more severe at distal muscles on the right side) were found at the right biceps, both EDC, right FCR, both FDI, both triceps. The abnormalities were more severe on the right side. Magnetic resonance imaging (MRI) of the cervical spine, in an initial neutral position revealed a focal atrophic change at the spinal cord at C4/5 to C6/7 levels, without an abnormal signal at the spinal cord. On the flexion sagittal view, the study showed enlargement and a crescent of high signal intensity of T2WI of the posterior epidural space at C3-C6/7 levels, with an epidural flow void in this space (Figure 2).

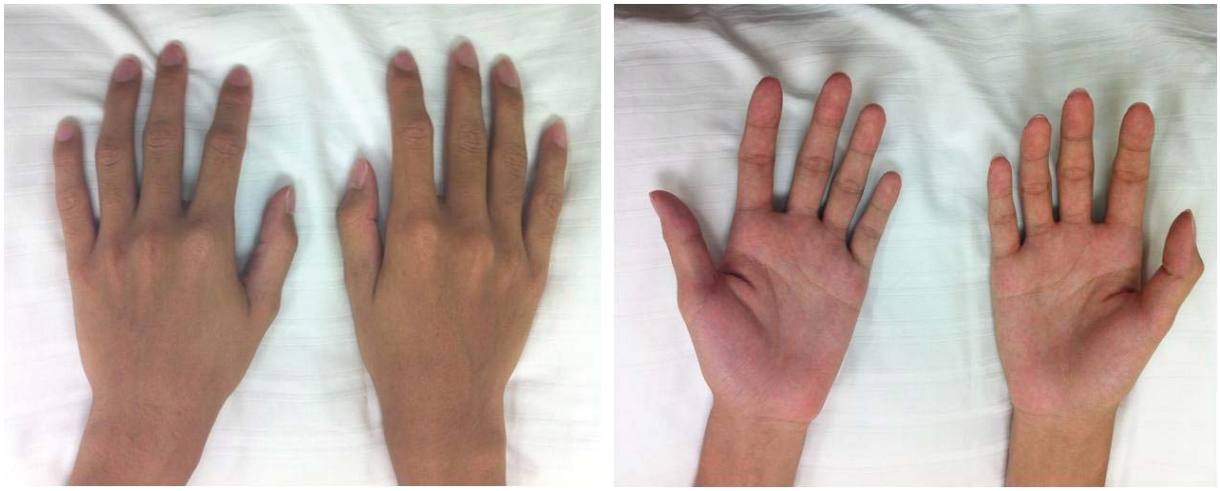

Figure 1: *Atrophy of bilateral intrinsic hand muscles. It was more prominent on the ulnar side and more severe on the right side.*

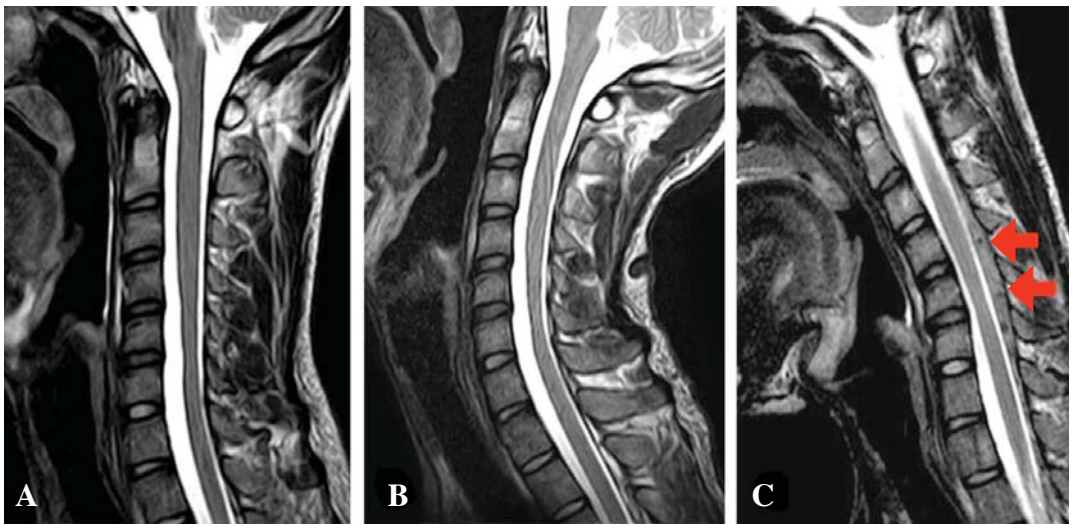

Figure 2: *A, B: Neutral position the sagittal T2-weighted MR image shows spinal cord atrophy at the C4/5 to C6/7 vertebral body, without enlargement of the posterior epidural space. C: The flexion MR image sagittal T2-weighted image, the study shows anterior widening of the posterior epidural space about C3-C6/7 level (red arrows) with a few flow void signals inside.*

The diagnosis of Hirayama's disease was made at that time, based on typical clinical characteristics and classic MRI findings on the flexion position. Propranolol was prescribed for relieving tremors. At the three-month follow up, the tremors had significantly improved. The atrophy and weakness of his forearm and hands had not progressed. A repeated MRI study revealed an unchanged widening of the posterior epidural space in the flexion position, with increased intensity enhanced after a Gadolinium injection (Figure 3).

Discussion

This case demonstrated the typical clinical findings of Hirayama disease. His clinical course was slowly progressive and predominantly involved in the right arm. The symptoms in many reported patients started on one side and spread to the other over several years, hence the term **'monomelic amyotrophy'.** The disease is often benign and does not cause severe disability. The weakness and atrophy are mainly found in muscles which are innervated by the eighth cervical and the first thoracic nerve roots, and less so by the seventh cervical roots. EMG almost always shows chronic denervation change in the affected limbs and sometimes in the asymptomatic limb. Acute or ongoing denervation is often found in active cases. This corresponds to the slow progressive degeneration of the anterior horn cells which leads to functional denervation and loss of motor neuron function. The pathology is limited to the relevant arm segments

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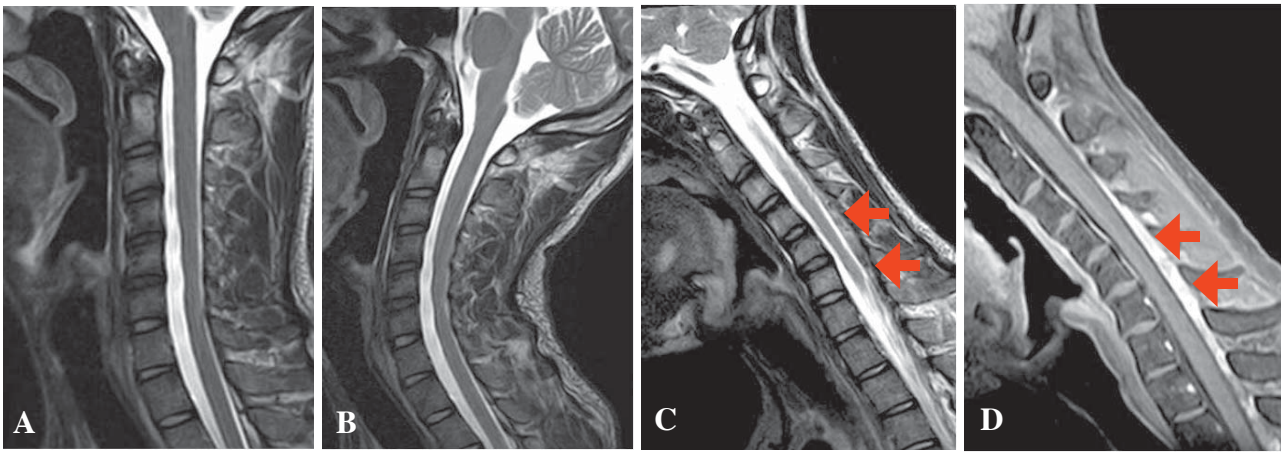

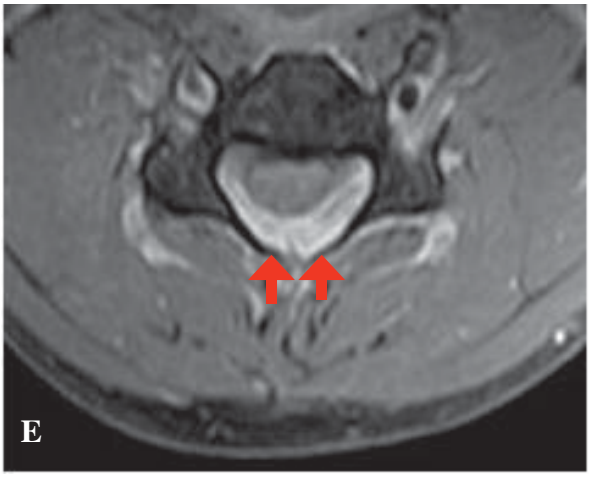

Figure 3: *A, B: Neutral position the sagittal T2-weighted MR image shows an unchanged spinal cord atrophy at the C4/5 to C6/7 vertebral body, without enlargement of the posterior epidural space.*
C- E: Unchanged anterior widening of the posterior epidural space about C3-C6/7 level (red arrows) with a few flow void signals inside on flexion MR image sagittal T2-weighted image and contrast enhancement after supportive treatment after about 3 months.

and both sides are involved. Interestingly, this disease is more prevalent in Asian populations and genetic association studies in Korean patients identifies KIAA1377 and C5orf42 as susceptibility genes for monomelic amyotrophy.[4,5]

The MRI and CT may show muscle atrophy.[1] Spinal cord flattening is an important finding in routine non-flexion MR images and should arouse suspicion. Detection of the focal spinal cord atrophy is another supporting diagnosis found in later stages of the disease.[1,6] Dynamic spinal cord compression at the neck flexion with a forward displacement of the posterior dura is an unequivocal finding in the progressive stage of this disease. The MRI imaging with extension and flexion of the cervical spine showed the relationship of posterior dura mater with the spinal cord and also the reduction in the anteroposterior diameter of the spinal cord.[7] A tight dural canal during flexion of the neck due to the disproportional length between the vertebral and the dural canal is the cause of Hirayama's disease.[6]

These findings are important for the diagnosis of this disease.[3,6-8] Morphologic changes on MR images should be correlated correctly with clinical and electromyographic data. Early diagnosis and supportive treatment is the gold standard to prevent progressive muscular weakness and atrophy.[7,8] However, the clinical course in most cases is benign. The progression of weakness is often stopped and becomes static. Therefore, conservative treatment is preferable. In some cases, a cervical collar may prevent neck flexion and stop disease progression.[9] Anterior cervical decompression and fusion is an option to prevent progressive neurological deficit in rapidly progressive cases and helps the patient to regain a better quality of life.[10]

Conclusion

Hirayama's disease is a juvenile non-progressive segmental spinal muscular atrophy. The correct diagnosis and physical therapy is the effective treatment in case focal atrophy of the spinal cord due to compression. Decompression and fusion is the treatment of choice.

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