Hirayama disease with juvenile myoclonic epilepsy: A case report

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Abstract

Hirayama disease (HD) is rare, but benign anterior horn cell disease, predominantly affecting young men. One of the symptoms, besides weakness, is abnormal movement in the hand. Juvenile myoclonic epilepsy (JME) is one of the most common types of generalized epilepsies and can be recognized by a myoclonic jerk and electroencephalography (EEG) features. We report the case of a 19-year-old male who had HD, with unilateral abnormal movement in the hand, which was diagnosed as JME. We should consider performing an EEG in patients with HD, who present with atypical hand movements, in order to differentiate it from seizure.

Key Words

Electroencephalography, Hirayama disease, juvenile myoclonic epilepsy, myoclonus

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What was known?

Hirayama disease is a rare, but benign anterior horn cell disease, predominantly affecting young men. It can be accompanied by abnormal movement in the hand.

Introduction

Hirayama disease (HD) is a rare disease that primarily affects young men in the second to third decades and it is characterized by weakness and atrophies in the hand and forearm, without sensory symptoms or pyramidal signs. The course of HD is initially progressive, with an insidious onset, followed by spontaneous stabilization within several years.[1] Hirayama disease differs from other types of motor neuron diseases, because of its nonprogressive course and pathological findings that show focal ischemic changes in the anterior horns of the lower cervical cord.[2] The common symptoms include distal hand weakness, wasting, cold paresis, and dysesthesia in the affected muscles.[2-5] One of the findings, besides weakness, in HD, is abnormal movement in the hand.[1]

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Juvenile myoclonic epilepsy is a common idiopathic epilepsy syndrome affecting up to 10% of all epilepsies with a high genetic predisposition that typically presents with generalized tonic–clonic, myoclonic, or absence seizures, or a combination of these.[6] In typical cases of JME, the seizures are usually bilateral and symmetric. However, focal and asymmetric clinical features are also reported in some cases.[7]

We present the first case of a 19-year-old male, who was diagnosed as having HD, with unilateral abnormal movement in his right hand, which was later proved to be JME.

Case Report

The patient is a 19-year-old male who visited our Neurology Clinic in November of 2013, due to persistent right distal hand weakness and involuntary movement in his right hand. His distal hand weakness started three years ago, but the weakness did not progress, and was only aggravated during the cold seasons. He complained of involuntary movement in his right hand that developed two months prior to admission. He did not have a history of seizures or other medical disease.

His initial vital signs were stable and his mentality was alert. The neurological examination at admission did not show any abnormalities in his cranial nerves. There was no sensory deficit, but on his motor examination, he had right distal hand and forearm weakness, which was graded as a medical research council grade 4+. His interossei and first dorsal interosseous muscles were atrophic, without fasciculation and his deep tendon reflexes were hyperactive without Hoffmann

or Babinski signs. However, he showed intermittent, irregular, involuntary, brief jerky abnormal movements of his right hand, which had a frequency of 1 to 2 Hz.

Routine laboratory tests including a complete blood count, electrolytes, blood sugar, liver, renal function tests, and thyroid function tests were within normal limits. Other autoimmune laboratory findings like antinuclear antibody, antineutrophil cytoplasmic antibody, rheumatoid factor, anti- Ro/La, and cardiolipin antibodies were normal. We performed a nerve conduction study and electromyography that demonstrated denervation potentials, with chronic denervation, and a reinnervation process in his right C7, C8, and T1 innervated muscles. Magnetic resonance imaging (MRI) of his cervical spine with gadolium enhancement illustrated a significant loss of cervical lordosis, but no significant cord compression or localized cord atrophy was seen in the supine position, however, anterior displacement of the spinal cord with the compression of the dural sac was observed on the flexion cervical MRI of the patient [Figure 1]. On account of the focal myoclonus of his right hand, we performed electroencephalography (EEG) and it illustrated a periodic 4-Hz generalized spike and wave complexes [Figure 2], which was suggestive of JME. The brain MRI did not demonstrate any abnormality that could cause asymptomatic epilepsy. Based on the above clinical, imaging and electrophysiological findings, he was diagnosed as HD with JME. His focal myoclonus improved after taking 600 mg of valproic acid.

Discussion

Hirayama disease was first reported in 1959, by Hirayama, and most of the cases were reported in Asian countries, such as, Japan, India, China, and Korea, showing an ethnic difference.[1-5] It is a benign, self-limited disease, characterized by asymmetric or unilateral weakness in the distal upper extremities. The pathomechanism of HD is generally explained by aggravation of the lower cord compression by the posterior dural sac, during neck flexion. This eventually leads to microcirculatory changes in the anterior spinal artery causing ischemia in the anterior horn cell.[1] This is supported electrophysiologically, where a somatosensory and motor-evoked potential study on HD demonstrated a reversible compromise in the central conduction pathways.[8] One of the common findings, besides weakness, in this disease, is abnormal movement in the hand. A previous study showed that hand abnormal movement was noted in 9.6% of the HD patients.[8] However, the published articles did not illustrate the findings in detail and described them simply by various terminologies, like tremor, contraction, trembling of fingers, and polyminimyoclonus.[1-5]

Janz described JME for the first time in 1957.[9] In JME, the myoclonic jerks are a distinctive feature and the EEG typically shows a 4 to 6 Hz spike and wake discharge. In general, JME demonstrates a bilaterally symmetrical motor manifestation, as well as interictal and ictal EEG patterns. Furthermore, myoclonic jerks frequently involved the distal upper limbs, including subtle extension of the fingers or hands.[10] Although JME is classified as a generalized epileptic syndrome, it can also show focal semiological patterns, and a recent study showed a focal semiological feature in 46% of JME patients.[7]

In our patient, the interesting abnormality was the presence of an irregular involuntary jerky abnormal movement of his right hand, which developed later following distal hand atrophy and weakness. It was initially confused with an HD-related symptom. However, he was diagnosed as JME after the completion of the EEG study. Further investigation is needed to clarify the association of timing between the myoclonus and epileptiform discharges. However, this is a limitation of our study. We did not perform a further study because the patient refused it.

The main pathogenesis of HD is explained by an ischemic change in the anterior horn cell without brain involvement. However, a recent clinical study reported on the possibility of more widespread involvement within the spinal cord as well as in the somatosensory cortex of the brain.[11] Gallo *et al*., proved this by using multiparametric magnetic resonance imaging in a HD patient and illustrated the cortical activation during simple motor tasks. They explained the cortical function changes by an

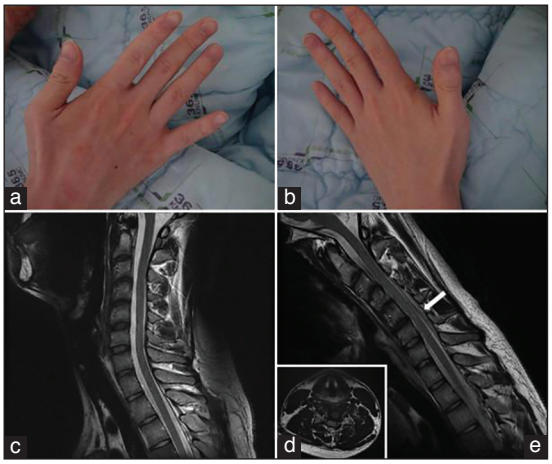

Figure 1: The right hand of the patient shows first dorsal interosseous and intrinsic muscle atrophy (a) Compared to the left hand. (b) The cervical magnetic resonance imaging (MRI) demonstrates the neutral (c) and flexion (d and e) MRI of the patient. The anterior displacement of the spinal cord with compression of the dural sac (arrow) is illustrated in the flexion MRI of the patient

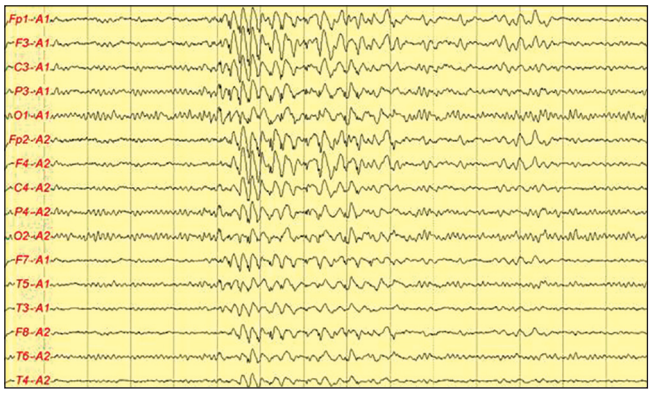

Figure 2: Electroencephalogram shows 4-Hz generalized epileptiform discharges

adaptive role in limiting the clinical consequences of structural cord damage.[11] Therefore, the underlying JME triggered in our patient could be partly explained by the result of 'hyper' cortical activation, secondary to reorganization of the somatosensory motor cortex, which coincided with the late onset of myoclonus that appeared three years after the weakness of the right hand.

To our knowledge, this is the first report on concurrent HD with JME. We should consider performing an EEG in patients with HD, who present with an atypical hand movement, in order to differentiate it from seizure.

What is new?

This is the first report on the concurrent Hirayama disease with juvenile myoclonic epilepsy.

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