SPASTIC PARAPLEGIA WITH PERIPHERAL POLYNEUROPATHY: A REPORT OF THREE CASES

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Spastic paraplegia is manifested by progressive spasticity and weakness of the legs and is one of the presenting signs of upper motor neuron disorders. Several diseases present solely with spastic gait disorders, but spastic paraplegia with severe polyneuropathy is an uncommon condition. We report three cases of spastic paraplegia from childhood with severe distal atrophy due to profound polyneuropathy; two of them had a hereditary and one sporadic (nonhereditary) background. In the present report, positive clinical and paraclinical findings with the possible differential diagnosis have been discussed.

Keywords • hereditary complicated • neurophysiology • polyneuropathy • spastic paraplegia • sporadic

Introduction

Spastic paraplegia was first reported by Seeligmüller; it is commonly manifested by corticospinal tracts involvement beginning in the lower extremities which causes spastic gait difficulty and may progress to involve the upper limbs as well as cranial nerves. Many different conditions have been reported regarding the full range of clinical manifestations of the patients with spastic paraplegia including nystagmus, optic atrophy, ocular palsies, ataxia, ichthyosis, kyphoscoliosis, pes cavus, and cardiac conduction defects. Most cases seem to have an inherited pattern.1

Harding divided hereditary spastic paraplegia syndrome into pure and complicated types depending on the clinical manifestations.5 Although “pure” spastic paraplegia is typically a relatively benign disorder without significant progression,3–6 but the “complicated” type may include parkinsonism, amyotrophy, blindness, deafness, epilepsy, and dementia.2,5

However, the association of spastic paraplegia and peripheral neuropathy is relatively rare.3,4,7,8

We report three cases of spastic paraplegia associated with peripheral polyneuropathy in either hereditary and/or sporadic (nonhereditary) patterns.

Case Report

Case 1

An 18-year-old male was referred to the Electrodiagnosis Department of Azzahra Hospital, Isfahan, Iran in November 2000 with a six-year history of progressive gait problems. He was born to healthy consanguineous parents and was thought to be normal at birth with no significant complaints until the age of 12 when he gradually developed progressive limb weakness and clumsy gait.

On physical examination, he was a normally-developed male with nasal speech, having normal mentation, and cranial nerves. He had steppage gait which was also spastic with a mild ataxic pattern. Marked weakness and atrophy of the hands and feet muscles with pes cavus were noted, however, the bulk and strength of the proximal muscles were normal. Hypertonicity of the limb muscles was present with bilateral extensor plantar responses. Deep tendon reflexes were normal in the arms, but abnormally brisk in the legs with unsustained ankle clonus. There were also clumsy...
hand movements, impaired tandem gait and ataxic finger to nose test. Romberg’s sign was negative. There were no bowel or bladder problems. No evidence of sensory abnormalities or involuntary movements could be found. Fasciculation was not observed.

Electrodiagnostic investigations were performed in this patient with the following results:
1. Nerve conduction studies revealed markedly prolonged distal motor latencies (e.g. median nerve = 8 m/s) and slowed velocity (e.g. ulnar nerve = 26 m/s), and decreased amplitude in all limbs. No sensory responses could be detected in upper or lower limbs. H-reflexes were unobtainable. F-waves showed prolonged latencies (e.g. ulnar nerve = 44 m/s) with decreased repetition rates.
2. Needle electromyography revealed 3+ spontaneous activities (i.e. fibrillations and positive sharp waves) in the foot, 2+ in the hand and leg, and 1+ in the quadriceps muscles with increased polyphasic and long-duration motor unit action potentials and partial interference pattern.

The overall electrodiagnostic impression was mixed sensory and motor peripheral neuropathy (mostly of demyelinating type) with more prominent involvement of the distal segments especially in the lower limbs.

All other paraclinical investigations including blood chemistries and radiological studies including magnetic resonance imaging (MRI) and computerized tomography (CT) scanning were normal.

Case 2
A 16-year-old male (the first case’s brother who had been referred to the same department and at the same time as mentioned for case one) was affected with the same problems as his brother, but appeared mentally retarded and had more severe cavus deformity of the feet, with an earlier onset of symptoms (starting at about 8 years of age). The electrodiagnostic result was the same as the first case. Other paraclinical investigations were also normal.

Case 3
A 19-year-old male was referred in May 2001 for electrodiagnostic studies due to the clumsy gait. No abnormalities were noted at birth or during the first year of life. He walked at 20 months but was clumsy. The pattern of his gait deteriorated and could only walk about 250 feet with assistance. The gait was wide-based and stiff, however, he was able to take care of his personal needs. He had 4 brothers and 4 sisters; all of them were normal. The patient seemed to have normal mentation, but his unrelated parents thought that although he had received formal schooling, he was a little lower than his siblings in mental activities.

On physical examination, the patient had a slow-pitched and slurred speech. Bilateral mild pes cavus and weakness of distal musculature in the hands and feet were noted with marked atrophy of the thenar and dorsal interosseous muscles. The bulk and strength of the proximal muscles were mildly decreased. There were increased muscle tone and deep tendon reflexes with bilateral ankle clonus and extensor plantar responses. No fasciculations or sensory deficits were noted.

Nerve conduction studies revealed very low amplitude motor responses of median, ulnar, and tibial nerves with prolonged distal latencies and mildly decreased velocity. No responses could be elicited in the study of peroneal, sural, and sensory fibers of the median and ulnar nerves. F-waves were unobtainable too.

Needle electromyography showed 4+ spontaneous activity in the leg and hand muscles and 2+ in the proximal muscles of the upper and lower limbs. There were also high amplitude, long duration, and polyphasic motor unit action potentials with decreased interference pattern in all tested muscles.

The final electrodiagnostic impression was mixed sensory and motor peripheral polyneuropathy (mostly of axonal type). All other paraclinical investigators were normal.

Discussion
Hereditary spastic paraplegia (HSP) presents with a relatively uniform clinical picture in the majority of patients. In the “uncomplicated” form, there is insidiously progressive lower limb spasticity with moderate weakness and no lower motor neuron features. The pattern of inheritance is usually autosomal dominant and less often recessive. In some families, the disease may be “pure”, but the existence of “pure plus” families is suggested in others.

Although some authors have reported impaired sensation and variable degrees of peripheral neuropathy in some patients with otherwise
uncomplicated HSP (especially those at older age), reports of HSP with prominent distal muscle wasting and peripheral neuropathy as a consistent early feature are uncommon. It has been mentioned that some degrees of generalized muscle atrophy is not uncommon in spastic paraplegia, but this is usually of late onset.

Some authors have reported variable disorders in which spastic paraplegia could be associated with peripheral neuropathy such as Werner’s syndrome, adrenomyeloneuropathy, Troyer or Troyer-like syndrome, tropical myeloneuropathies due to malnutrition, cobalamin, and other micronutrient deficiencies.

In this paper, we reported three cases of spastic paraplegia in association with a profound mixed sensory-motor neuropathy. Cases 1 and 2 appeared to have an inherited pattern while case 3 seemed to be sporadic. Although similar cases have been reported before, the sporadic occurrence in unrelated families has obscured the possibility that noninherited spastic paraplegia with peripheral neuropathy may constitute a distinct syndrome. Therefore, additional studies are necessary to determine the nature of this condition.

Individuals with hereditary sensory-motor neuropathy type V (with spastic paraplegia) usually present in the latter half of the first or second decade with a gait difficulty. Regarding the clinical and electrodiagnostic pictures, we think that the cases 1 and 2 (the two brothers) could fall in this category.

Histological abnormalities found in the patients with pure HSP consist of axon degeneration mainly involving the longest, large-diameter ascending and descending spinal cord pathways. Spinal roots and peripheral nerves, however, have been reported to be normal in both morphological and electrophysiological studies, but abnormalities in motor and somatosensory evoked potentials have been reported. Cases 1 and 2 (the two brothers) showed a more demyelinating type of peripheral neuropathy while case 3 demonstrated an axonal type.

In conclusion, it should be borne in mind that, though rare, some patients with spastic paraplegia could manifest clinical and electrophysiological evidences of profound peripheral polyneuropathy either in an inherited background or sporadically.

References