

Monomelic Amyotrophy: Case Report

Monomelik Amyotrofi: Olgu Sunumu

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Summary

Monomelic amyotrophy (MMA) is a benign motor neuron disorder. MMA is usually restricted to the upper limb, lower limb involvement is rare. MMA should be considered in differential diagnosis in patients presenting with amyotrophy involving lower and upper extremity behind other diagnosis. A rehabilitative approach can be useful for these patients to compensate for the atrophy and decreased muscle strength. In this report, we discussed the clinical and electrophysiological features of two cases with MMA and the literature is reviewed. *Türk J Phys Med Rehab* 2008;54:116-8.

Key Words: Monomelic amyotrophy, lower extremity, upper extremity, motor neuropathy

Özet

Monomelik amyotrofi (MMA) iyi seyirli bir motor nöron hastalığıdır. Genellikle üst ekstremitayı tutar, alt ekstremita tutulumu nadirdir. MMA alt ve üst ekstremitede amyotrofi yapan nedenler arasında diğer tanılar yanında mutlaka düşünülmelidir. Rehabilitatif yaklaşım bu olgularda atrofi ve azalmış kas kuvvetini yerine koyma açısından önemlidir. Bu çalışmada, MMA'li iki olgunun klinik ve elektrofizyolojik bulgular tartışıldı ve literatür gözden geçirildi. *Türk Fiz Tıp Rehab Derg* 2008;54:116-8.

Anahtar Kelimeler: Monomelik amyotrofi, alt ekstremita, üst ekstremita, motor nöropati

Introduction

Monomelic amyotrophy (MMA) is a benign motor neuron disorder (MND) in the young with male predominance (1,2). The disease is characterized by insidious onset of weakness and wasting restricted to a single limb which usually progresses for a few years followed by spontaneous arrest and lack of sensory, bulbar, and pyramidal signs (2). MMA has been reported chiefly from Asian countries, although there has been scattered cases reported from other regions (3,4). But as yet there is no convincing evidence to explain the unique geographic distribution and predilection to Asian. The precise cause for this disorder is yet unknown (2). Laboratory testing is frequently normal or nonspecific except for electrophysiologic studies which typically demonstrated reduced compound muscle action potential (CMAP) amplitudes, and features consistent with acute and chronic denervation in affected muscle (5-8).

MMA is commonly misdiagnosed and should be considered in differential diagnosis in patients presenting with amyotrophy involving lower and upper extremity behind other diagnosis.

In this report, we described the clinical and electrophysiologic features of a 22-year-old man with MMA and of the lower limb, a 53-year-old man with MMA of the upper limb.

Case Reports

Patient 1

A 24 years old man presented with relatively stable weakness and wasting of the right calf since the age of 19. He related the problem in his right leg to a heavy lifting during a home moving event. The following months he developed insidious and progressive atrophy of the right calf and pes cavus deformity (Figure 1). Then the symptoms appeared to be arrested. He had no history of previous poliomyelitis or a febrile illness, exposure to toxins, heavy

metals. He did not report significant pain in low back and legs, but complained of weakness in right leg during running or walking long distances. He was not aware of significant muscle cramps or fasciculation. There was no family history of neuromuscular disease.

His examination demonstrated severe wasting and mild weakness of his right calf muscle (maximum circumference at 15 cm below the lower pole of patella, right 34 cm, left 40 cm), and 2 cm shortening of the right lower extremity (Figure 1). Other extremities were normal. He could walk on his toes and heels. Muscle stretch reflexes were absent in the right lower limb, 1+ at the left patellae and 2+ at the left Achilles tendon. Babinski signs were absent bilaterally. Sensory examination and mental status were entirely normal. Routine laboratory tests, serum electrolytes, creatinine kinase, urea, creatinine, liver and thyroid function tests were normal. Nerve conduction studies (NCS) were normal. Only right peroneal nerve CMAP amplitude was decreased compared the other side. Needle EMG showed abnormal spontaneous activity at rest (positive sharp waves and fibrillation potentials) together with large amplitude motor unit potentials (MUPs) and reduced recruitment in right tibialis anterior, gastrocnemius, soleus, peroneus longus, and vastus lateralis. Lumbosacral and cervical magnetic resonance imaging (MRI) were performed together with thin slice lumbar and dorsal computed axial tomography (CT), which all were normal. Based on previous clinical, electrophysiological, and radiological descriptions of this disorder, a diagnosis of benign MMA of the right lower limb was made.

As the patient had 2 cm shortness in right leg and pes cavus deformity, we recommended proper foot orthotics for preventing possible complications and helping him in activities of daily living. Exercise program was given to strengthen lower extremity muscles.

Patient 2

A 53 years old man was admitted with insidious onset, gradually progressive weakness and wasting of right intrinsic



Figure 1. Posterior view showing amyotrophy of the right lower limb.

hand and forearm muscles since the age of 13. Initial symptom was difficulty in grasping and in performing fine work. The weakness in his hand slowly worsened over several months. Gradual progression was followed by stabilization of symptoms, cold paresis, and finger tremor. The right hand weakness was stable until this time. He denied symptoms of pain, numbness, paresthesia, or dysesthesia in the right upper limb. There was no history of poliomyelitis, cervical trauma, fracture and surgery of the right upper limb. There was no exposure to toxins or heavy metals and allergic phenomena or family history.

Examination was remarkable for significant atrophy of forearm and intrinsic hand muscles including the interossei and right thenar eminence (Figure 2). Right brachioradialis reflex was reduced, but other reflexes were symmetrically normal. Muscle power were grade 5/5 in the shoulder girdle, 4/5 in the extensor digitorum communis, flexor carpi ulnaris (FCU), flexor carpi radialis (FCR) and 3/5 in the abductor pollicis brevis (APB), opponens pollicis (OP), dorsal and palmar interossei (IO), abductor digiti minimi (ADM). Sensory examination and mental status were entirely normal. Investigations revealed normal blood counts, serum electrolytes, creatine kinase, urea, creatinine, liver and thyroid functions were normal. Motor NCSs revealed reduced amplitudes of the CMAP recorded from right ADM and absent recorded from right APB. Other motor NCSs were normal. Sensory NCVs were normal in all extremities. Needle EMG showed abnormal spontaneous activity at rest (positive sharp waves and fibrillation potentials) together with large amplitude MUPs and reduced recruitment in the right upper extremity muscles (FCU, first dorsal IO, FCR, APB, ADM). Right biceps muscle showed only chronic neurogenic changes in MUP morphology. Cervical paraspinal muscles were normal. MRI of the cervical spine showed median disc protrusion at C5-6 and there was not root compression.

He went to several doctors in the last 15 years. Surgery was suggested by some physicians for the atrophies of forearm and hand muscles. They blamed median and ulnar nerve entrapment and cervical disc herniation for this condition. The patient was informed about his illness. No surgery was recommended. Exercise program was given to strengthen right upper extremity and hand muscles and proper devices were recommended for helping him in activities of daily living.



Figure 2. Amyotrophy of the right upper limb and hand.

Discussion

The condition of our patients shares many features with focal amyotrophy first described by Hirayama in 1959 (5). The disease manifests as the insidious onset of weakness and atrophy in one limb, usually involving the intrinsic muscles of the hand and flexor and the extensors of the forearm (5). There are rare cases in which the weakness and atrophy occurs in the lower limb, or only in the calf muscle (4,6-8). One of our patients was presented with unilateral amyotrophy of distal lower limb (calf muscles), the other was presented with unilateral amyotrophy of distal upper extremity.

In the majority of patients, reflexes are normal or reduced (5,9,10). These findings are very important also in differential diagnosis. Our findings were similar with literature. We excluded upper neuron involvement with this findings.

EMG features are suggestive of anterior horn disease (8). Positive sharp waves, fibrillations, and mainly large MUPs are seen, recruitment is reduced in a pattern corresponding to the areas of weakness and atrophy (4,8,9). Similar abnormalities can be found in uninvolved or less affected limbs (10). NCSs are usually normal except where prominent axon loss exist in motor nerves, resulting in reduced CMAPs, prolonged motor distal and slight reduction in motor NCVs (4,8,11).

The differential diagnosis of MMA includes illnesses which present with painless weakness and atrophy in limbs (1-6). Those disorders include intraaxial processes of the spinal cord such as syrinx, intramedullary neoplasms, cord infarctions, and early presentation of more diffuse motor neuron diseases. When pain and sensory loss is absent or mild compared to motor symptoms, polyradiculopathy, brachial plexopathy, mononeuropathy, inherited neuropathies, and rare forms of distal myopathies can resemble MMA (1,12,13). Detailed EMG testing and physical examination and history taking can usually differentiate between those entities in situations in which the clinical examination is unclear.

In our cases sensory NCSs were entirely normal. We did not find any conduction block, temporal dispersion, conduction velocity slowing or F-wave prolongation in these cases. We excluded these pathologies by NCSs.

Imaging studies of the spinal cord have shown variable results. Metcalf and colleagues (12) identified cervical segmental cord atrophy in one patient using myelography and postmyelographic CT. Biondi and colleagues (13) reported MR studies of the cervical cord in seven patients. In their series, five MR studies showed atrophy of the cervical spine. In two other cases showed normal findings. MRI and CT exclude spinal, root and plexus pathologies. In these cases we performed MRI and CT of the spinal cord. In second patient we observed median disc protrusion at C5-6 but there was not root compression in MRI evaluation and needle EMG did not indicate C5-6 root pathologies. In first patient, MRI evaluation was entirely normal did not indicate root pathology.

Second patient's medical history was very important about the course of these conditions. Some physicians related the atrophy and weakness blamed to ulnar and median nerve entrapment and cervical disk herniation. Surgery was suggested for the treatment.

One of the diseases that should be considered is post-polio syndrome, which can be differentiated from MMA by a definite history of poliomyelitis (14). MMA is a self-limiting disease with a spontaneous arrest, unlike as in progressive muscular weakness in post-polio syndrome (14).

The relationship to other MNDs like ALS is unclear. MNDs involving the upper and lower motor neurons are heterogeneous group of syndromes with various clinical presentations, most with poor prognosis. Absent bulbar and pyramidal signs, and lack of spreading to the other limbs after 4 years, distinguish MMA from a monomelic onset of ALS (6).

One can assume that MMA may become generalized and progress to ALS. Most of the follow-up studies demonstrate that MMA does not progress to ALS (4,8,11,15). However, Rowin et al. (1) reported a case with MMA exhibiting late progression after a prolonged period of disease stability.

There are no distinctive clinical or laboratory findings with other motor neuron diseases, and the diagnosis of MMA can be made only retrospectively after a prolonged observation time (1).

Conclusion

MMA should be considered in differential diagnosis in patients presenting with amyotrophy involving lower and upper extremities. Studies to exclude other causes of amyotrophy and careful follow-up examinations to document disease stabilization are necessary to diagnose this uncommon disorder. Also, our approach should be to ease the daily activities and to prevent long term complications.

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