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***Case Report* / Olgu Sunumu**

Motor Neuron Disease Presenting with Dropped Head

Baş Düşmesi ile Prezente Olan Motor Nöron Hastalığı

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Motor neuron diseases are a group of progressive neurological disorders that destroy motor neurons that control voluntary muscle activity such as swallowing, walking, speaking and breathing. The common form of motor neuron disease is amyotrophic lateral sclerosis. Neurological examination presents specific signs associated with upper and lower motor neuron degeneration. In the absence of any biological marker, the diagnosis of motor neuron disease is based on clinical features, combined with the results of electromyography. Some patients of motor neuron disease with atypical presentation have been reported in the literature. We presented here a case of motor neuron disease with atypical presentation who had progressive weakness of the neck muscles.

Key words: Motor neuron disease; dropped head.

Motor nöron hastalığı, yutma, yürüme, konuşma ve nefes alma gibi işlevlerden sorumlu olan istemli kasları innerve eden motor nöronların tutulumu ile karakterize progresif nörolojik bir hastalıktır. En sık görülen formu amyotrofik lateral sklerozdur. Nörolojik muayene bulguları üst ve alt motor nöron dejenerasyonunu gösterir. Hastalığın tanısı için spesifik bir biyolojik marker bulunmamaktadır. Klinik özellikler elektromiyografik bulgularla birleştirilerek tanı konur. Literatürde az sayıda atipik prezentasyonlu motor nöron hastalığı olgusu bulunmaktadır. Bu yazıda boyun kaslarında progresif güçsüzlükle başlayan atipik bir motor nöron olgusu sunulmuştur.

Anahtar sözcükler: Motor nöron hastalığı; baş düşüklüğü.

Motor neuron diseases (MND) are a group of progressive neurological disorders that destroy motor neurons that control voluntary muscle activity such as swallowing, walking, speaking and breathing.[1] The common form of motor neuron disease is amyotrophic lateral sclerosis (ALS).[1] Neurological examination presents specific signs associated with upper (spasticity, brisk reflexes, Babinski sign) and lower (muscle atrophy, weakness, fasciculations) motor neuron degeneration.[1,2] Patients may present with symptoms like wasting of the muscles in the hands, dragging foot or slurred speech.[1,2] In the absence of any biological marker, the diagnosis of motor neuron disease is based on clinical analysis, combined with the results of electromyography.[1] Some patients of motor neuron disease with atypical presentation have been reported in the literature.[2,3] The pattern of muscular atrophy in these patients differed from that of typical MND in that severe muscle involvement was confined to the upper limbs, predominantly the proximal portion and shoulder girdle.[2,3] We presented here a case of MND with atypical presentation.

CASE REPORT

A 53-year-old man presented with the progressive weakness of the neck muscles [Medical Research Council Score (MRC) was found 4/5 in neck flexors and extensors]. On admission, his neurological examination except neck

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weakness was normal and initial electroneuromyography demonstrated only neurogenic involvement of the muscles innervated by upper cervical segments. Eight months later, he noticed weakness of the proximal limb muscles, especially shoulder girdle (MRC was found 4/5 in deltoideus, biceps and triceps brachii, trapezius, brachioradialis but normal in distal limb muscles). In the following six months, dysarthria, dysphagia became appearent beside the weakness of the upper limbs. Proximal muscles (deltoideus, biceps brachii, trapezius, brachioradialis) were more severely affected than distal hand muscles, and examination revealed normal higher mental functions, mixed dysarthria, sluggish palatal reflex and wasting of the tongue. Motor system revealed fasciculations and severe wasting of the upper limb muscles. The involvement of respiratory muscles were also observed at this stage of the disease. There was mild diffuse weakness of the lower limbs but he could walk independently. Tendon reflexes were normoactive and plantar response was flexor at the early stage of the disease but the deep tendon reflexes were reduced in the upper extremities by the time with the wasting of the muscles. At this stage of the disease, motor and sensory nerve conduction studies were all normal. Electromyography showed fibrillations and fasciculation potentials, large motor units with long duration, a decrease in motor units, and an incomplete recruitment pattern, especially in the upper limb muscles and the tongue muscles. Routine blood investigations, thyroid function tests, collagen vascular work-up, protein electrophoresis were normal. There was no testicular atrophy, gynecomastia and sensory disturbances. Relevant investigations to look for evidence of malignancy, lymphoproliferative disorders, thyroid dysfunction, and collagen vascular disease were negative. The patient died 56 months of the disease because of aspiration pneumonia.

DISCUSSION

We described a case with atypical presentation of motor neuron disease. The initial symptoms of motor neuron diseases are usually localized in the limbs or bulbar muscles.[1] Over the past few years, there were only a few case reports about the atypical clinical forms of motor neuron disease especially the form which presented with the weakness of the neck muscles and/or the proximal muscles of the upper limbs.[2,3] Our case also presented only with dropped head. "Dropped head syndrome" caused by neck extensor weakness and has been reported in a variety of neuromuscular disorders. This syndrome can be seen in inflammatory myopathies, myasthenia gravis, amyotrophic lateral sclerosis, facioscapulohumeral muscular dystrophy, nemaline myopathy, carnitine deficiency and spinal muscular atrophy.[4] Katz et al.[5] reported that 12 of 790 patients evaluated in a neuromuscular clinic had neck extensor weakness with head drop, and only one of them was diagnosed with motor neuron disease. Gourie-Devi et al.[4] reported nine cases with neck extensor weakness observed as an early symptom or developing during the later stages of the motor neuron disease. They reported that the neck muscle weakness with head drop was seen at varying intervals (mean 28 months) after the onset of limb weakness or bulbar symptoms, and was not a presenting symptom. However, in our patient, the dropped head was the presenting symptom and the head drop could be caused by early involvement of anterior horn cells innervating the paraspinal muscles. Hu et al.[3] found 10% of proximal limb involvement in patients with motor neuron disease and they found the median survival of atypical forms of motor neuron disease longer than typical forms (57 months versus 39 months) and more common in males (male/female: 9/1). Our patient was male and lived 56 months after the diagnosis. Motor neuron disease should be taken into consideration in patients with atypical presentations such as dropped head.

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