Amyotrophic Lateral Sclerosis, forgotten symptoms: case report

Esclerosis Lateral Amiotrófica, síntomas olvidados: reporte de caso

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Resumen

La esclerosis lateral amiotrófica (ELA) se caracteriza por ser una patología neurológica con compromiso neurodegenerativo progresivo de las primeras y segundas neuronas motoras en la vía piramidal, produciendo de manera marcada debilidad muscular y discapacidad motora. Puede presentarse un compromiso no motor hasta en el 50 % de los pacientes al momento del diagnóstico, como cambios en el comportamiento, alteraciones cognitivas, síntomas autonómicos, alteraciones en la deglución, la voz y parkinsonismo. Presentamos el caso clínico de una paciente femenina de setenta años en quien se sospechó ELA durante una consulta ambulatoria en el servicio de neurología, con posterior confirmación del diagnóstico mediante estudios electrodiagnósticos.

Palabras clave

Esclerosis Lateral Amiotrófica, Síndrome de la neurona motora superior, Síndrome de la neurona motora inferior.

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Abstract

Amyotrophic lateral sclerosis (ALS) is characterized by being a neurological pathology with progressive neurodegenerative compromise in first and second motor neurons at the pyramidal pathway, markedly producing muscle weakness and motor disability. It can present a non-motor compromise in up to 50 % of patients at the time of diagnosis, such as changes in behavior, cognitive alteration, autonomic symptoms, alterations in swallowing, voice and parkinsonism. We present the clinical case of female 70-year-old patient in whom a suspicion of ALS is made during an outpatient

Palabras clave

Amyotrophic lateral sclerosis, upper motor neuron syndrome, lower motor neuron syndrome.

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Introduction

ALS is defined as a progressive neurodegenerative disease that compromises both upper and lower motor neurons, generating progressive muscle weakness leading to death (1,2). According to recent reports, ALS has an incidence of 1.7–3 cases per 100.000 people per year and a prevalence of 10–20 cases per 100.000 people per year, rendering it a rare pathology and, consequently, difficult to diagnose, often delaying detection until advanced stages (3-5). The disease can follow different courses, with the most common site of onset being the spinal region, representing 58–82 % of cases, in contrast to bulbar onset, which accounts for approximately 27–28 % of cases (1,6,7).

Bulbar-onset ALS is characterized by an average survival of approximately 2 years from symptom onset, is more frequent in women than men, and patients typically present with spastic dysarthria, facial spasticity, lingual atrophy, fasciculations, dysphagia, and later progression to other spinal regions (4,5,8).

Case Report

A case is presented of a 70-year-old female patient from Cartagena, Colombia, with no significant personal or family pathological history. The patient attended the neurology outpatient clinic with a clinical picture of approximately two years' evolution, initially characterized by episodes of coughing during meals, changes in behavior, and a nasal tone of voice. One year later, these symptoms were accompanied by limitations in upper limb mobility, difficulty holding objects, and loss of strength in both upper extremities.

During the neurological examination, the patient was oriented to time, place, and person. Facial asymmetry was evident, with no ophthalmoparesis; fasciculations were observed in the left periorbital region, and the labial commissure was deviated to the right. Additionally, lingual fasciculations at rest, absence of the gag reflex, and a muscle strength of 3/5 in both upper extremities—accompanied by muscular fasciculations during contraction and stimulation—were identified. Furthermore, "monkey hands" were observed, characterized by atrophy of the thenar and hypothenar muscles as well as the hand flexors, with no compromise of the lower extremities nor alterations in deep tendon reflexes.

Electrodiagnostic studies, including electromyography and nerve conduction studies of the upper and lower extremities and the tongue, were indicated. The results revealed fibrillation potentials and positive sharp waves in the tongue muscles, as well as motor unit potentials with increased amplitude and giant potentials in the upper limb muscles with polyphasic characteristics, establishing signs of pure motor neuropathy with evidence of chronicity. Additionally, signs of neuronal dropout strongly suggesting amyotrophic lateral sclerosis were observed.

Based on these findings, pharmacological management with riluzole at a dose of 50 mg every 12 hours was initiated, a treatment that was well tolerated by the patient.

Image 1. Monkey hands with thenar and hypothenar atrophy.

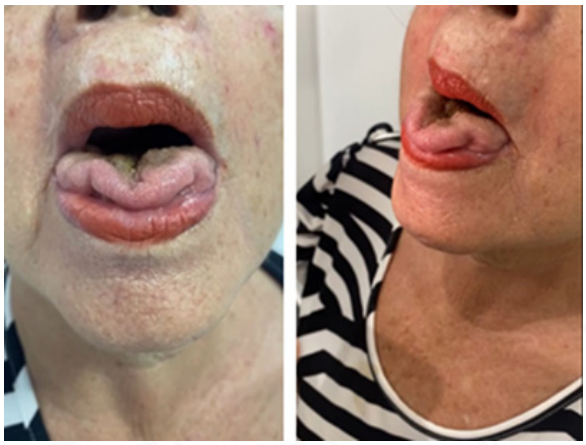

Image 2. Lingual fasciculations and atrophy

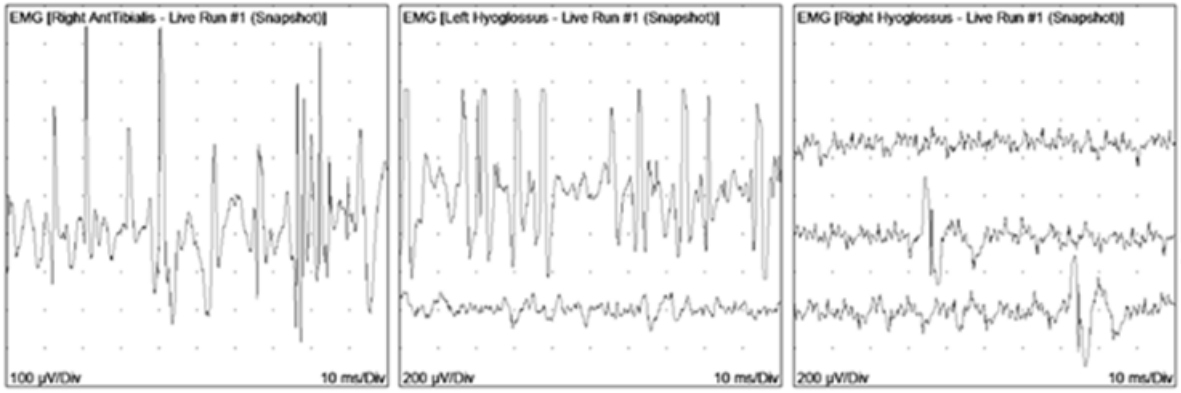

Image 3. Electromyography and nerve conduction study of the tongue.

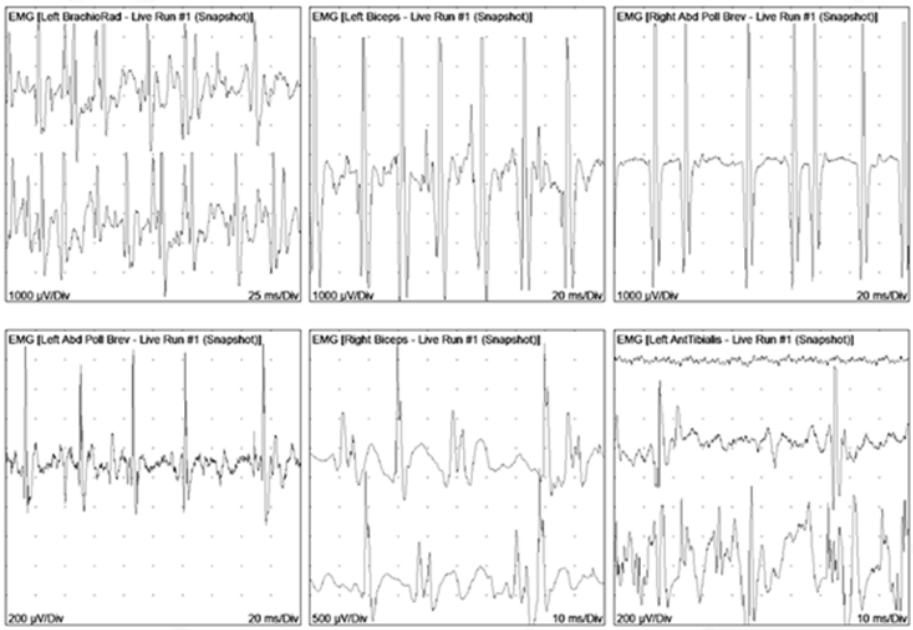

Image 4. Electromyography and nerve conduction study of the upper extremities.

Discussion

Amyotrophic lateral sclerosis (ALS) is defined as a progressive neurological disorder characterized by the degeneration of motor neurons in both the brain and spinal cord (3,4). Typically, it begins with focal muscle weakness that gradually progresses to involve the respiratory musculature, eventually leading to death (3,9). Statistically, the most common initial symptom, present in approximately 70 % of cases, is weakness of a limb (4,10). In 25 % of patients, the disease may debut with involvement of the muscles responsible for swallowing, chewing, or phonation, while less than 5 % begin with respiratory muscle compromise. Additionally, between 5–15 % of patients may develop personality changes, decreased intellectual capacities, and significant behavioral alterations (1,11,12).

In the presented clinical case, the patient initially exhibited symptoms without evident involvement of the upper or lower limbs. Instead, the initial symptoms included frequent coughing during meals, changes in voice quality, and behavioral alterations, with a progressive evolution over approximately 24 months. This initial presentation, which includes bulbar involvement, represents a smaller percentage of ALS cases. Subsequently, the patient developed upper limb weakness with no involvement of the lower extremities. The observed signs and symptoms evidenced compromises of both upper and lower motor neurons. Additionally, family members reported changes in the patient's personality and behavior, which further supported the clinical suspicion.

Regarding the physical examination, characteristic findings of the disease were identified, such as muscle weakness and atrophy, dysphagia with episodes of choking, coughing, sialorrhea, facial diparesis, lingual fasciculations, loss of muscle tone, and emotional lability (3,13). In this patient, specific findings included thenar and hypothenar atrophy in the upper extremities, manifesting as "monkey hands" (Image 1), as well as lingual fasciculations and atrophy (Image 2), decreased strength in the upper limbs, and facial asymmetry.

Diagnostic aids play an essential role in confirming this clinical entity. Electromyography and nerve conduction studies are fundamental tools, revealing findings such as fibrillation potential and positive sharp waves in the affected muscle groups (5,14,15). In this case, electromyography demonstrated fibrillation potentials and positive sharp waves in the tongue (Image 3), as well as signs of polyphasic potentials and pure motor neuropathy in the upper extremities. These parameters strongly suggest a highly probable diagnosis of ALS.

To further support the diagnosis, recognized clinical criteria—such as the El Escorial, Airlie House, and Awaji-Shima criteria—can be employed, which achieve a sensitivity exceeding 80 % and a specificity approaching 95 % (1,7,12). In this case, the scores obtained in the patient strongly supported the diagnosis.

Regarding treatment, the therapeutic options approved by the Food and Drug Administration include riluzole and edaravone. Riluzole acts by suppressing the excessive activation of motor neurons, while edaravone reduces oxidative stress (9,12). Although these therapies are not curative, they have been shown to delay the need for gastrostomy and reduce associated morbidity and mortality, moderately improving survival. In this case, pharmacological management with riluzole, administered at a dose of 50 mg every 12 hours, was initiated as the initial treatment, and it was well tolerated by the patient.

Conclusion

Amyotrophic lateral sclerosis remains a rare clinical entity with significant involvement of both upper and lower motor neuron groups. Its diagnosis requires a high index of clinical suspicion, and it is essential to remember that the clinical presentations of the disease can vary from one patient to another. Therefore, whenever there is the presence of progressive bulbar symptoms associated with behavioral changes and emotional involvement, ALS should be considered as a differential diagnosis.

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