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Case Report

Bulbar-Onset Amyotrophic Lateral Sclerosis Presenting as Acute Respiratory Distress

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Abstract

Introduction: Amyotrophic Lateral Sclerosis (ALS) is a rare, progressive, and fatal neurodegenerative disorder characterized by degeneration of upper and lower motor neurons. Early diagnosis remains challenging, particularly in atypical or bulbar-onset forms, often leading to delayed management and poorer outcomes.

Case Presentation: We report the case of a 52-year-old woman with a one-year history of generalized fatigability, head drop, dysphagia, and exertional dyspnea, who was admitted to the intensive care unit following emergency tracheostomy for acute respiratory distress due to bilateral vocal cord paralysis. Initial investigations, including cervical CT scan, brain MRI, cerebrospinal fluid analysis, and electroneuromyography (ENMG), were inconclusive. Differential diagnoses included myasthenia gravis and cerebellar pathology. After multidisciplinary reassessment and repeat ENMG, a diagnosis of bulbar-onset ALS was established according to the Gold Coast criteria. Despite supportive care, the patient died three days after transfer to the neurology department due to acute respiratory failure.

Discussion: This case illustrates the diagnostic difficulty of atypical ALS presentations, particularly when respiratory or bulbar symptoms predominate. Delayed neurological evaluation contributed to late diagnosis. Although emerging biomarkers such as neurofilament light chain show promise for earlier detection and prognostic assessment, they remain unavailable in many settings. Early recognition of clinical warning signs and adherence to updated diagnostic criteria are essential to reduce delays and optimize patient care.

Conclusion: Bulbar-onset ALS may initially mimic other neuromuscular or otolaryngological conditions, leading to significant diagnostic delay. Increased awareness of atypical presentations, appropriate use of ENMG, and integration of updated diagnostic criteria are critical to improving timely diagnosis and management.

Keywords: Amyotrophic Lateral Sclerosis; Bulbar onset; Diagnostic delay; Electromyography

Introduction

Amyotrophic Lateral Sclerosis (ALS) is a rare, progressive, and incurable neurodegenerative disease that affects motor neurons, leading to progressive muscle weakness, disability, and eventually death [1]. The symptoms of ALS may mimic those of compressive neuropathies or other motor neuron diseases, particularly at the early stages of a patient's clinical course. The main warning signs warranting further investigation include weakness without sensory symptoms, atrophy in multiple nerve distributions, progressive bilateral and global symptoms, the presence of bulbar signs (such as tongue fasciculations and speech/swallowing difficulties), and, when surgery is performed, a lack of improvement.

The aim of this presentation is to highlight the diagnostic challenges of ALS in its atypical form, which may delay patient management.

Case Presentation

We report the case of a 52-year-old female patient with a past medical history of arterial hypertension and hypertensive cardiopathy under treatment. She was admitted to the intensive care unit following a tracheostomy complicated by a moderate pneumomediastinum, performed on an emergency basis due to acute respiratory distress.

History of illness: The patient had experienced generalized

fatigability for one year, with a tendency toward head drop. A few months later, dysphagia and exertional dyspnea developed, prompting consultation with a pulmonologist. Two days prior to her presentation at the ENT emergency department, she developed acute laryngeal dyspnea and dysphonia. Flexible nasofibroscopy revealed bilateral vocal cord immobility in the closed position, with no patent airway, necessitating emergent tracheostomy.

ICU admission findings: On admission, the patient was fully conscious (GCS 15/15) and hemodynamically stable. Respiratory assessment showed SpO_2 at 92% on room air without signs of respiratory distress. Thoracic imaging demonstrated bilateral pneumonia of infectious appearance, for which empirical antibiotic therapy was initiated. Neurological examination revealed no sensorimotor deficit but axial hypotonia with cervical weakness, brisk tendon reflexes in the lower limbs, abolished reflexes in the upper limbs, induced fasciculations in the lower limbs, atrophy of the legs and hands, bradykinesia, and ocular paresis.

Etiological work-up: Cervical CT scan was unremarkable, brain MRI showed no abnormalities, and cerebrospinal fluid analysis did not reveal albuminocytological dissociation. The initial electroneuromyography (ENMG) showed no evidence of anterior horn cell involvement, and immunological testing was negative.
The patient was re-evaluated in collaboration with the neurology team and a repeat ENMG was performed. The final diagnosis was bulbar-onset Amyotrophic Lateral Sclerosis (ALS).

Discussion

At our institution, the 20 August 1953 Hospital in Casablanca, the majority of acute respiratory distress cases are of obstructive otolaryngological origin, most commonly related to neoplastic diseases such as laryngeal carcinoma, hypopharyngeal carcinoma, or locally advanced anaplastic thyroid carcinoma, as well as infectious causes such as cervicofacial cellulitis leading to airway compression. Consequently, before conducting a thorough interview with the patient and her family, an upper airway pathology of ENT origin was initially suspected. However, the cervical CT scan revealed no abnormalities. Following nasofibroscopy and a detailed history, a neurological etiology appeared more likely.

It is noteworthy that the patient's symptoms had been evolving for approximately 12 months prior to hospitalization. She had previously consulted a pulmonologist due to exertional dyspnea, which prompted thoracic imaging. Nonetheless, at no point was a neurological evaluation sought, despite the presence of axial hypotonia and cervical weakness, which were key clinical features pointing toward a neuromuscular disorder.

Atypical forms of ALS account for a significant proportion of diagnostic delays. Clinicians must be able to recognize the hallmark features of ALS while carefully excluding other potentially treatable conditions. Throughout this process, it is essential to remain vigilant for the onset of respiratory or bulbar symptoms that require urgent management, to take into consideration treatment delays, access to clinical trials, as well as the psychological impact of diagnostic uncertainty on patients and their families. The diagnostic work-up should be targeted, pragmatic, and tailored to each clinical context in order to ensure both reliability and timeliness.

In our patient, the diagnosis was only confirmed after a 15-day stay in the intensive care unit. The initial ENMG was inconclusive, and myasthenia gravis was considered due to the presence of fatigability, dysarthria, and dysphagia, although immunological testing was negative. Cerebellar involvement was also discussed, as certain clinical manifestations may initially mimic a neuromuscular disorder; however, brain MRI findings were normal.

The positive diagnosis of ALS is based on clinical assessment and electromyography (ENMG). The new diagnostic criteria (the "Gold Coast" criteria, proposed in 2020) [2] include:
- Progressive motor impairment, documented by history or serial clinical examinations, following a period of normal motor function, and:
• Either evidence of both upper and lower motor neuron involvement in at least one body region (if limited to one region, both central and peripheral signs must be present),
• Or evidence of lower motor neuron involvement in at least two body regions, and;
- Exclusion of alternative pathological processes through appropriate paraclinical investigations, depending on the clinical context.

In our case, after 15 days of hospitalization, a repeat ENMG was performed and reviewed by neurologists specialized in ALS, who confirmed the diagnosis of bulbar-onset ALS. The patient was subsequently transferred to the neurology department, tracheostomized and breathing spontaneously on room air, for further management. Unfortunately, she died three days later due to acute respiratory failure.

A key question remains whether reliable biomarkers could help distinguish ALS from other clinically similar disorders at an earlier, even preclinical stage. Among the most promising candidates are neurofilaments, which are cytoskeletal proteins of Neurons Composed of Light (NFL), medium, and heavy chains [3].

In ALS, plasma and cerebrospinal fluid NFL concentrations are significantly elevated compared with healthy controls and patients with other neurological conditions. Although no threshold value is specific to ALS, NFL levels are correlated with disease prognosis [4], [5]. In addition, serum creatinine has been shown to inversely correlate with survival [6], while elevated creatine kinase (CPK) levels are associated with longer survival [7]. Several other molecular biomarkers are currently under investigation [8].

Unfortunately, NFL measurement is not available in our setting, and other laboratory parameters in our patient were within normal limits.

It is clear that ALS is an incurable and fatal neurodegenerative disease, however, it would likely have been possible to slow the progression of the disease if the diagnosis had been made earlier and improve the quality of life of this patient, who was diagnosed after a year of progression and at the stage of acute respiratory distress. For this reason, it is crucial to expand the clinical and communication skills of healthcare professionals, starting as early as medical training [9]. Furthermore, diagnostic criteria must emphasize the persistent challenge of ALS recognition, which remains a common problem in clinical practice [9].

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Conclusion

Our case highlights that the diagnosis of ALS remains a frequent clinical challenge. Familiarity with rare phenotypes and differential diagnoses, appropriate use of ENMG and emerging biomarkers, as well as the adoption of the Gold Coast criteria, represent key strategies to reduce diagnostic delays and optimize patient management.

Author Declaration

We, the authors, confirm the following:

- This manuscript complies fully with the Instructions to Authors of the journal.
- Individual author contributions are detailed as follows: [DAMAAN and ABIDI designed the study and drafted the manuscript; BENHAMZA et LAZRAQ collected data; MILOUDI et BENSAID contributed to critical revision; all authors approved the final version].
- Authorship requirements have been met, and all authors have reviewed and approved the final manuscript prior to submission.
- This manuscript has not been published previously, and is not currently under consideration by any other journal.
- This observation was conducted in accordance with ethical standards.

Conflict of Interest: The authors declare that they have no conflicts of interest.

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