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Case Report

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Bulbar Onset Amyotrophic Lateral Sclerosis in A 50 Years Old Female Patient: Case Report

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ABSTRACT

Amyotrophic lateral sclerosis (ALS) is a rare, fatal, and progressive neurodegenerative disorder disease characterized by varied clinical presentations, with early motor symptoms being the most prevalent. However, the early phases of the disease may present primarily with bulbar symptoms. Speech and swallowing problems are hallmark features of bulbar onset amyotrophic lateral sclerosis (B-ALS). Patient with B-ALS tend to have a poorer prognosis and show more pronounced neuroimaging changes. This case illustrates the clinical presentation, diagnostic approach, and complications in a 50-year-old female patient presenting with isolated lower motor neuron disease features across multiple body regions, ultimately diagnosed with bulbar-onset amyotrophic lateral sclerosis (ALS) based on the Gold Coast Criteria. Case presentation: A 50 years old female who presented with a progressively worsening difficulty of swallowing, cough with choking and slow speech that had nasal quality with decreased tone for a duration of two years. After structural and local causes were ruled out, a diagnosis of ALS was made with the Gold Coast diagnostic criteria. Conclusion: ALS with bulbar onset can have a grave prognosis and hence requires early diagnosis and a multidisciplinary approach toward effective treatment. Clinicians should be aware of the possibility of ALS presenting as an isolated lower motor neuron disease when it involves at least two body regions in the absence of alternative explanation.

Keywords: Amyotrophic Lateral Sclerosis, Bulbar Onset, Dysphagia

INTRODUCTION

Amyotrophic lateral sclerosis (ALS) is a lethal and progressive neurodegenerative disorder that manifests with several clinical features, of which limb weakness is the predominant initial symptom [1]. In the initial stages, the disease may exhibit symptoms localized to the bulbar region without affecting the extremities [2]. Speech and swallowing

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problems are hallmark features of bulbar-onset amyotrophic lateral sclerosis (ALS), and they are linked to worse outcomes due to their higher degree of neurodegenerative burden, as evidenced by more pronounced abnormalities in grey and white matter [3,4]. This sub-type also shows distinct patterns of brain atrophy, with more extensive atrophy correlated with greater severity and shorter disease duration (5). The clinical features of ALS vary greatly, with 22.1% having bulbar onset [1]. The only established risk factors identified so far are advanced age, male sex, and familial predisposition to ALS. The median duration of survival from onset to death is often 3 years after the initial manifestation of symptoms. Older age and bulbar onset are consistently reported to have poorer outcomes [3].

CASE PRESENTATION

A fifty-year-old female patient initially presented to the surgical outpatient department (OPD) with a complaint of swallowing difficulties lasting three months. Initially, she experienced difficulties with solid foods, which later progressed to problems with swallowing liquids. Her attempts to feed were often disrupted by episodes of coughing and choking. Further inquiry revealed that she had been struggling with communication over the past two years. Her speech was characterized as slow and nasal, with diminished tone, and this condition had progressively worsened. During the same period, she experienced significant yet unquantified weight loss, loss of appetite, and fatigue, but she was still able to perform self-care. Additionally, there was no family history of similar diseases, nor was there a history of neck trauma, abnormal body movements, behavioral changes, headaches, bowel or bladder incontinence, sensory disturbances, or known chronic medical conditions.

After her evaluation at the surgical OPD, the need to rule out esophageal cancer prompted a chest CT scan, which did not reveal any structural cause for her dysphagia. Instead, the scan showed a left basal lung ground-glass opacity with focal bronchial wall thickening, suggestive of aspiration pneumonia. Furthermore, a Flexible Fiber Optic Laryngoscopy was performed, revealing pooling of saliva in the glottis region with no visible mass; there was no documentation of a vocal cord adduction gap.

Once structural causes of dysphagia and voice changes were ruled out, the patient was referred for a neurological evaluation, which yielded the following findings: her Glasgow Coma Scale (GCS) was 15/15, and her Mini Mental Status Examination score was 27/30. Her speech remained slow and nasal in quality with decreased tone, fitting the profile of lingual-type dysarthria. Cranial nerve examination revealed decreased tone in the masseter and temporalis muscles, diminished gag reflex, and decreased tongue power with spontaneous fasciculation's. There was no difficulty moving or closing her eyes. Motor examination showed induced fasciculation's in the lower extremities, bilateral hypotonic in both upper and lower limbs with reduced reflexes, and muscle power graded at 4/5 in all extremities. Hoffman's sign, triple flexion, and the extension plantar reflex were absent. Sensory examination was normal across all modalities.

Basic laboratory investigations and EMG/NCS studies produced the following results: Table 1 summarizes the patient's investigations: Brain and neck CT scan with contrast: Unremarkable

Table 1. Basic laboratory investigations of the patient

Investigation	Parameter	Result	Reference Range
Complete Blood Count (CBC)	WBC	10,500 /µL	4,000–11,000 /µL
	Neutrophils (%)	63	40–70%
	Lymphocytes (%)	31.9	20–40%
	Hemoglobin	12.9 g/dL	12.0–16.0 g/dL (female)
	Hematocrit	36.80%	36–46% (female)
Renal Function	Creatinine	0.51 mg/dL	0.5–1.1 mg/dL (female)
	Urea	14.4 mg/dL	7–20 mg/dL
Serum Electrolytes	Sodium	143 mEq/L	135–145 mEq/L
	Potassium	3.67 mEq/L	3.5–5.0 mEq/L
	Chloride	98.1 mEq/L	98–107 mEq/L
Liver Enzymes	AST	27 U/L	5–40 U/L
	ALT	17 U/L	7–56 U/L
Thyroid Function Test	TSH	0.73 mIU/L	0.4–4.0 mIU/L

EMG/NCS results showed that motor conduction tests were normal in all 8 tested nerves across four muscle groups in both upper and lower extremities. Sensory conduction tests in 8 nerves were also normal. F-wave and H-reflex studies were normal. However, the needle EMG was abnormal in all 16 tested muscles, showing abnormal spontaneous insertional activity in the left first dorsal interosseous, bilateral thoracic paraspinals, bilateral vastus medialis, bilateral tibialis anterior, and bilateral gastrocnemius medial head. Neurogenic motor unit potential (MUP) waveform abnormalities and abnormal interference patterns were observed in the tongue, bilateral thoracic paraspinals, and both upper and lower extremities.

Three months after her initial presentation, the patient returned to the Emergency Department with a productive cough of white sputum lasting two weeks, associated with low-grade fever and shortness of breath. Her initial symptoms of swallowing difficulty and communication issues had further worsened.

Physical examination revealed a pulse rate of 102, respiratory rate of 21, and oxygen saturation of 84% in room air. Chest examination highlighted decreased air entry over the left basal lung area, with a chest X-ray suggestive of aspiration pneumonia. She was started on IV antibiotics, oxygen support, and subsequently transferred to the ICU due to hypotension, suspected to be related to sepsis from a chest focus.

Following ICU admission, her hemodynamic condition improved, and the aforementioned care continued. On the sixth day of her ICU stay, a feeding gastrostomy tube was inserted due to ongoing feeding difficulties.

Ten days later, she developed bradypnea, with breathing rates ranging from six to ten breaths per minute and a change in mentation. Arterial blood gas (ABG) analysis revealed a pH of 7.16, PCO2 of 92, and HCO3– of 31. She was subsequently placed on a mechanical ventilator in AC/VC mode, and her condition stabilized. Eight days later, a tracheostomy was performed due to anticipated prolonged intubation. Attempts to wean her from the mechanical ventilator were unsuccessful, as she repeatedly developed bradypnea and required continuous pressure support. Her pulse rate fluctuated persistently, ranging from bradycardia at 48 to tachycardia at 118, as depicted in an ECG during one of the bradycardic episodes shown in Figure 1. After fifty days in the ICU, she was transferred to another institution, where similar care was continued. Disease-modifying treatment was not initiated, as the patient could not afford the medication, and its efficacy was limited once the disease had progressed to the stage necessitating mechanical ventilation.

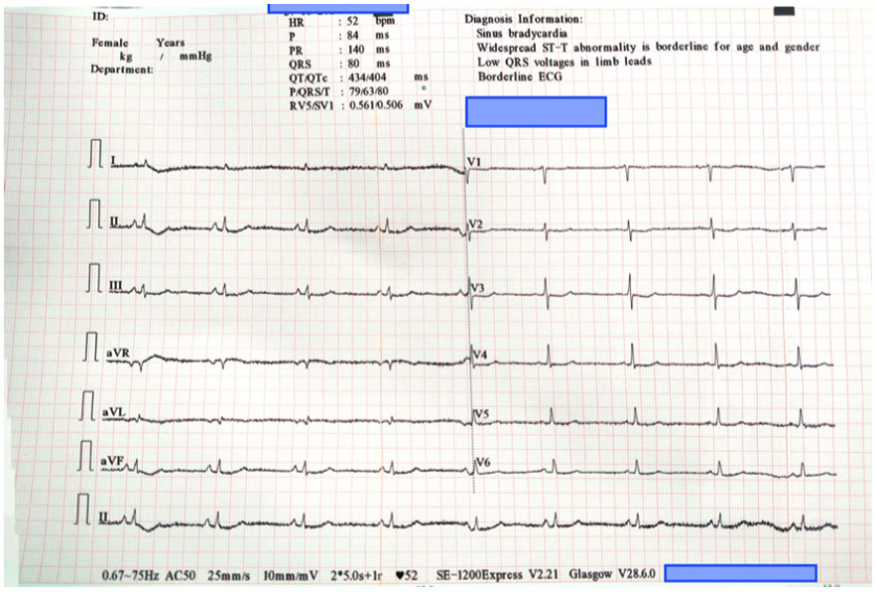

Figure 1. ECG of 50 years old female patient with bulbar Onset amyotrophic lateral sclerosis.

DISCUSSION

This case highlights the clinical presentation, diagnostic process, and complications in a 50-year-old female patient with bulbar-onset amyotrophic lateral sclerosis (ALS). The patient initially presented with dysphagia that progressively worsened from difficulties swallowing solids to liquids, accompanied by coughing and choking during meals. These symptoms are characteristic features of bulbar dysfunction, in which the degeneration of motor neurons in the brainstem results in reduced coordination and muscular weakening of the oropharyngeal muscles. The progression from solid to liquid dysphagia indicates a deterioration in bulbar muscle function, characteristic features of this ALS subtype [3,4,6].

Additionally, the patient experienced dysarthria, manifesting as slow, nasal speech with reduced tone over two years. Dysarthria is a common bulbar presentation in ALS, often co-occurring with dysphagia [7]. These symptoms are nonspecific and characteristic features of bulbar ALS, which often creates diagnostic challenges.

The initial diagnostic approach involved the use of a CT scan and flexible fiberoptic laryngoscopy to exclude structural causes of dysphagia. Such diagnostic delays are common in bulbar-onset ALS, as patients often seek medical care from multiple specialists before a definitive diagnosis is established [8]. Neurological examination and electrophysiological findings of the patient fulfill updated Gold Coast diagnostic criteria for ALS. The Gold Coast criteria (GCC) have a greater sensitivity compared to prior diagnostic criteria like the revised El Escorial (rEEC) and Awaji criteria while maintaining high specificity [9-11]. It also has simplified the diagnosis of ALS and made it possible to diagnose ALS in those patients who have only a progressive lower motor neuron disease in at least two body regions in the absence of alternative explanation. In this case, electromyographic (EMG) findings such as abnormal insertional activity, abnormal neurogenic motor unit potential(MUP) waveforms, and an abnormal interference pattern indicated that the patient had lower motor neuron disease across four body regions: the bulbar, cervical, thoracic, and lumbosacral. Normal motor and sensory conduction studies rule out other neuromuscular disorders, solidfying the diagnosis of ALS.

The disease course in ALS might leads to multi-organ complications beyond the nervous system [12]. A chest CT of the patient demonstrate the presence of ground-glass opacity in the left basal lung and thickening of the bronchial wall, indicative of aspiration pneumonia. Aspiration is a significant concern in bulbar amyotrophic lateral sclerosis (ALS), leading to potentially life-threatening complications [13]. Aspiration arises from impaired swallowing mechanisms, a hallmark of bulbar dysfunction.

ALS also significantly impacts respiratory function, with complications often leading to ventilatory failure, the most common cause of death in ALS patients [14]. This patient also has respiratory problems such as an ineffective cough, difficulty clearing secretions, and gradual ventilatory failure that necessitates a treacheasotomy and ventilator support [15]. These complications result from neural, airway, pulmonary, and neuromuscular changes, affecting integrated respiratory functions like sleep, cough, swallowing, and breathing [16]. A range of respiratory therapies, such as non-invasive ventilation (NIV), lung volume recruitment, mechanical insufflation-exsufflation, and respiratory strength training, are used to address these challenges. NIV, in particular has demonstrated the ability to enhance both the quality of life and survival rates in individuals with ALS [16,17].

Amyotrophic lateral sclerosis (ALS) patients may also experience cardiovascular problems. A number of studies have documented different ranges of cardiovascular issues connected to heart rhythm and rate [18]. In this case, the patient exhibited sinus bradycardia and low QRS voltage in limb leads on ECG. Although bradycardia is not common, autonomic dysfunction, particularly affecting parasympathetic cardiac control, is observed in amyotrophic lateral sclerosis (ALS) patients, especially those with bulbar involvement which can manifest as reduced heart rate variability and depressed sinus arrhythmia [19,20].

This case underscores the complex interplay of neurological, respiratory, and cardiovascular complications in ALS, emphasizing the importance of a multidisciplinary approach to optimize patient outcomes.

CONCLUSION

Amyotrophic lateral sclerosis (ALS) with bulbar onset can have a grave prognosis, making early diagnosis and a multidisciplinary approach to treatment essential. This disease poses a significant economic burden for both the patient and the healthcare provider. Clinicians should be aware that ALS can present as an isolated lower motor neuron disease, particularly when it affects at least two body regions without an alternative explanation. In these cases, the Gold Coast criteria should be employed to aid in the diagnosis of this rare condition.

CONSENT FOR PUBLICATION

Written informed consent was obtained from the patient for publication and use of images. The written consent is available for review by the Editor-in-Chief of this journal upon inquiry.

ETHICAL CONSIDERATIONS

Ethical approval is deemed unnecessary by St. Paul's Hospital Millennium Medical College Institutional Review Board as this is a single rare case faced during clinical practice and it does not involve experiments on humans or animals.

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DISCLOSURE

The authors declare no conflicts of interest in this work.

ABBREVATIONS

ABG: Arterial Blood Gas; ALS: Amyotrophic Lateral Sclerosis; B-ALS: Bulbar Onset Amyotrophic Lateral Scelrosis; ECG: Electrocardiography; EMG: Electromyography; GCC: Gold Coast Criteria; MUP: Motor Unit Potential; NCS: Nerve Conduction Test; NIV: Non-Invasive Ventilation; rEEC: Revised El Escorial.

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