# Diagnostic Odyssey in ALS

# Patient #2: Bulbar Onset ALS

59-year-old-man

**Case contributor and commentary:**

**Dr. Martina Wiedau, MD**
University of California Medical Center |
Los Angelas, California

## Initial Symptoms

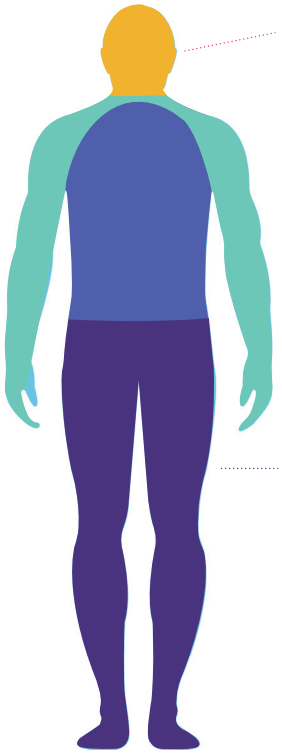
**Medical History:**

» No significant past medical history

» Emotional outbursts — mainly laughing, but also crying more easily (3 months)

**Figure Key**

- Bulbar region
- Cervical region
- Thoracic region
- Lumbar region
- Site of key/defining symptoms



- Increasingly slurred and slow speech (8 months)
- Difficulty swallowing fluids and crunchy foods (4 months)
- Mild difficulty with breathing when supine, uses 3 pillows
- Stiffness and mild imbalance when walking (2 months)
- No extremity fasciculations or cramps

**Commentary:** The patient in this case experienced bulbar onset symptoms, accompanied by emotional lability (known as pseudobulbar affect or PBA), and subsequent spreading to respiratory muscles and extremities. PBA is often associated with bulbar onset, rapid disease progression, and a lower score on a functional rating scale.[1] Pharmacological treatment is available to help manage emotional symptoms.[2]

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## Physical Exam

### A physical examination was performed:

- Cranial nerves exam: tongue fasciculations, spastic dysarthria/slowed speech due to increased muscle tone, slow tongue movements, no tongue atrophy
- Motor exam: normal bulk all limbs
- Increased tone both legs
- Fasciculations, all limbs
- Weakness left finger extensors 5-/5, left first dorsal interosseus 4/5,
- Deep tendon reflexes: 3+/4 bilateral upper extremities and lower extremities, (normal reflex exam: 2/4)
- Positive Babinski reflex, palmomental reflex, jaw jerk, glabellar reflex
- Gait analysis: spastic gait

## Laboratory Tests

### After the physical exam, follow-up tests were ordered:

**Electromyography (EMG)/nerve conduction studies (NCS)**

- Positive sharp waves and large polyphasic units of long duration firing at high rates with a reduced recruitment pattern in multiple muscles of the left arm only
- Fasciculations in the tongue
- Normal motor and sensory nerve conductions

**Blood work :**

- Screening for neurological disease appeared negative

**Imaging:**

- Normal brain MRI and mild stenosis C4/5 on C-spine MRI, no myelopathy

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## Diagnosis

### Differential Diagnosis:

The symptoms of progressive weakness of bulbar region, dysarthria, and dysphagia with hyperreflexia resulted in consideration of the following differential diagnoses:

» Brainstem disorder

» Myasthenia gravis

### Criteria Used for Diagnosis:

» UMN features in bulbar and lumbar region

» LMN features determined by EMG in bulbar, cervical, and lumbar regions

### Diagnosis:

Probably ALS per Airlie House and Awaji criteria

**Commentary:** When patients have bulbar onset, brain imaging is typically used to check for differential diagnosis of stroke or other intracranial process.

## Disease Progression

**The diagnosis was made 9 months after the patient first noticed symptoms. The patient experienced the following course of disease, passing away from respiratory failure at 29 months after symptom onset:**

**Diagnosis at 9 months following symptom onset**

| Symptom Onset | 6 mos | 18 mos | 21 mos | 23 mos | 24 mos | 29 mos |
|---|---|---|---|---|---|---|
| Progressive dysarthria and dysphagia with weight loss | | | | | | |
| | Started using walker | | | | | |
| | | Started using speech generating device and had PEG tube inserted | | | | |
| | | | Difficulty with sleeping led to sleeping in sitting position | | | |
| | | | | Started using BiPaP | | |
| | | | | | Became unable to ambulate and bedridden | |
| | | | | | | Passed away from respiratory failure |

References

1. Tortelli R, Copetti M, Arcuti S, et al. Pseudobulbar affect (PBA) in an incident ALS cohort: results from the Apulia registry (SLAP). Journal of Neurology. 2016;263(2):316-321. doi:10.1007/s00415-015-7981-3

2. Jackson CE, McVey AL, Rudnicki S, Dimachkie MM, Barohn RJ. Symptom Management and End-of-Life Care in Amyotrophic Lateral Sclerosis. Neurologic Clinics. 2015;33(4):889-908. doi:10.1016/j.ncl.2015.07.010

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