In its classic form, ALS affects motor neurons at 2 or more levels supplying multiple regions of the body.

A 42-year-old man presented to the pulmonary clinic with gradually worsening shortness of breath. He had received a diagnosis of amyotrophic lateral sclerosis (ALS) 6 months earlier by a neurologist and now had a variety of symptoms, including gradually worsening slurred speech and fasciculations of the arms (Videos 1a, 1b) hands, (Video 2) and legs. (Videos on next page.)

The ALS diagnosis had been based on results of a muscle biopsy after MRI of the brain and electromyography showed no pathology. His muscle tone was now 4/5 in both upper and lower limbs and sensation was intact overall. The patient was taking riluzone.

In the pulmonary clinic, the patient’s vital signs were as follows: temperature 98.7°F; blood pressure, 130/80 mm Hg; respiratory rate, 17 breaths/min; pulse rate, 91 beats/min. CPAP therapy at night was initiated.

Discussion
ALS, first described by Charcot in the 19th century, is a relentlessly progressive, presently incurable, neurodegenerative disorder that causes muscle weakness, disability, and eventually death. ALS is also known as Lou Gehrig’s disease, after the famous New York Yankee baseball player who was affected with the disorder.¹⁻³

Research suggests that a variety of factors may underlie the disease process, including genetic, environmental, and occupational factors; toxins; high-level physical activity; electrical injury; physical trauma; medical illness; and exposure to high magnetic field activity. All have been posited by different investigators at different times but none have yet been proved with certainty.⁴ It has been speculated that there is slow ongoing process of selective motor neuron destruction by a complex chain of injurious events after certain exposures that ultimately involves excitotoxins, oxidative stress, altered calcium homeostasis, and mitochondrial dysfunction.⁵

In its classic form, ALS affects motor neurons at 2 or more levels supplying multiple regions of the body. It affects lower motor neurons in the anterior horn of the spinal cord and in the brain stem; corticospinal upper motor neurons in the precentral gyrus; and, frequently, prefrontal motor neurons that are involved in planning or orchestrating the work of the upper and lower motor neurons. Loss of lower motor neurons leads to progressive muscle weakness and wasting (atrophy). Loss of corticospinal upper motor neurons may produce stiffness (spasticity), abnormally active reflexes, and pathologic reflexes.

When only upper motor neurons are involved, the disease is called primary lateral sclerosis (PLS). The course of PLS differs from that of ALS and is usually measured in decades. Rarely, the disease is restricted to bulbar muscles, in which case it is called progressive bulbar palsy (PBP). In most patients who present with initial involvement of bulbar muscles, the disease progresses to classic ALS.

Diagnosis
The diagnosis of ALS is primarily clinical. Patients with bulbar involvement experience slurred speech, hoarseness, and choking while feeding. Patients with lower limb involvement may trip, stumble, and fall while walking. A “slipping” gait has been reported by some patients. Initial cramping, stiffness, and weakness of intrinsic muscles are common with upper limb involvement. Diagnosis is made more easily when multiple organs have been affected. The hallmark of the disease is involvement of both upper and motor neurons while sensory functions are spared.
Nerve conduction studies and needle electromyography are useful to confirm the diagnosis of ALS and to exclude peripheral conditions that resemble ALS. Laboratory test results generally are normal and are performed primarily to rule out other disease processes. Biochemical markers in blood are used to identify diseases with similar presenting symptoms. Examination of cerebrospinal fluid usually is not necessary. Genetic testing may be performed to identify gene defects in some familial types of ALS and to detect other inherited motor neuron diseases.

Motor unit number estimation (MUNE), a nerve conduction study that can quantify the numbers of motor units innervating an individual muscle, may also be useful.  

**Treatment**

Treatment of ALS is divided broadly into patient education, mechanism-specific treatment, and adaptive or supportive treatment. Patient education can be enhanced by referral to multidisciplinary clinics staffed by specialists with an ALS focus, with educational material for both patients and families, and by participation in support groups.

The glutamate pathway antagonist riluzole (Rilutek) is the only medication that has shown efficacy in extending life in ALS. In 2 double-blind, randomized placebo-controlled trials, riluzole was shown to prolong median tracheostomy-free survival by 2 to 3 months in patients younger than 75 years with definite or probable ALS of less than 5 years’ duration.  

**References:**


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http://www.consultantlive.com/nervous-system-diseases/amyotrophic-lateral-sclerosis-0

**Links:**
