

AMYOTROPHIC LATERAL SCLEROSIS (ALS)

What are Motor Neuron Diseases?

Motor neuron diseases are neurodegenerative diseases that cause selective loss of the nerve cells that directly connect the brain to muscles.

What is ALS?

Amyotrophic lateral sclerosis (ALS, commonly called “Lou Gehrig's disease” in the US) is the most common motor neuron disease in adults. ALS affects both upper and lower motor neurons throughout the brain and spinal cord. There are other forms of motor neuron disease that affect only the upper motor neurons, affect only the lower motor neurons or have a restricted distribution.

Demographics

Based on U.S. population studies, a little over 5,600 people in the U.S. are diagnosed with ALS each year (approximately 15 new cases a day.) It is estimated that as many as 30,000 Americans have the disease at any given time.

Most people who develop ALS are between the ages of 40 and 70, with an average age of 55 at the time of diagnosis. However, cases of the disease do occur in persons in their twenties and thirties. ALS is 20% more common in men than in women. However with increasing age, the incidence of ALS is more equal between men and women (www.ALSA.org).

Symptoms/Signs of Lower Motor Neuron Disease

The primary symptom of lower motor neuron disease is weakness. The weakness usually begins in one hand, one foot, or the tongue. As weakness becomes more severe, involved muscles become smaller, and weakness spreads to other muscles. Cramping and twitching of muscles are common; however, these symptoms are also common in healthy people. Electromyography (EMG) is a test that is very sensitive in detecting lower motor neuron disease.

Symptoms/Signs of Upper Motor Neuron Disease

The primary symptoms of upper motor neuron disease are stiffness, slowness, and clumsiness of movement. The symptoms usually begin in one limb or in the mouth or throat, later spreading to other parts of the body. There are no standard laboratory tests for upper motor neuron disease, but spasticity (a specific type of stiffness), abnormally brisk tendon reflexes, Babinski's sign, and diminished fine motor coordination are seen as diagnostic signs on examination.

Physiology

CLINICALLY DEFINITE ALS

1. Upper motor neuron involvement in 3 of 4

- Bulbar
- Cervical
- Thoracic
- Lumbosacral

2. Lower motor neuron involvement in 3 of 4

- Bulbar
- Cervical
- Thoracic
- Lumbosacral

3. Spread of symptoms over time

4. Absence of any other explanation

OTHER MOTOR NEURON DISEASES

Only lower motor neuron involvement

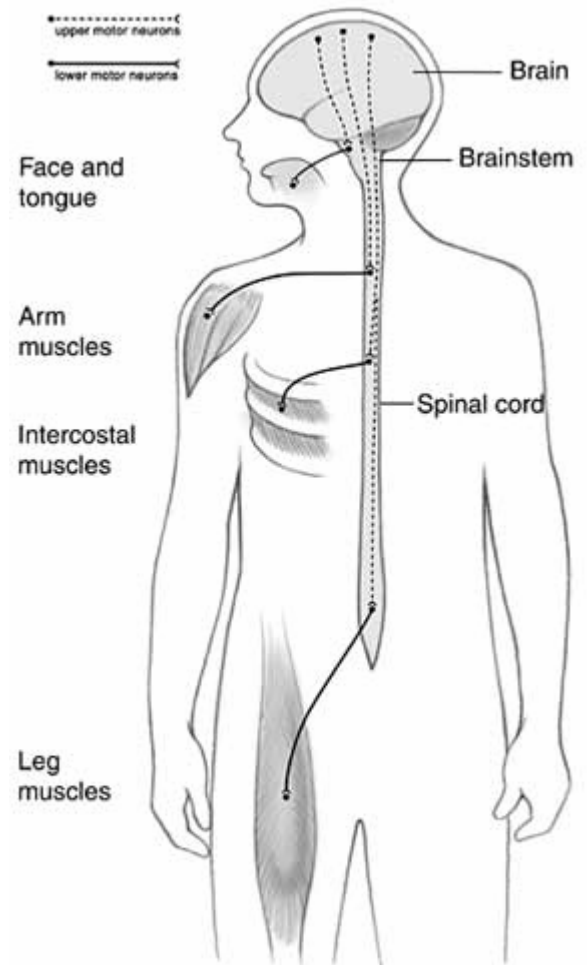
- Kennedy's disease
- Spinal muscular atrophy
- Progressive muscular atrophy variant of ALS
- Segmental motor neuron disease of limbs
- Progressive bulbar palsy

Only upper motor neuron involvement

- Primary lateral sclerosis
- Progressive bulbar palsy

Genetics of ALS

The genetic mechanisms that cause ALS are only partially understood. The only known cause of ALS is a mutation of a specific gene, called the SOD1 gene. This mutation is believed to make a defective protein that is toxic to motor nerve cells. The SOD1 mutation, however, accounts for only 1 or 2 percent of ALS cases, or 20 percent of the familial (inherited) cases.



Relationship between FTD and ALS

While estimates vary, it is now believed that approximately 20% of ALS patients also have the signs and symptoms of frontotemporal dementia. As a result, patients may lack the ability to fully understand the meaning of their illness, they may make poor decisions about their clinical care, or they may become agitated and difficult for caregivers who are trying to help them. Other patients may have trouble understanding language or communicating, which is a separate problem from the articulation problems that come with ALS.

Some ALS patients have had changes in their behavior or psychiatric problems for years before they developed ALS, and only until a full neuropsychological exam is completed does the family understand that an FTD disease had begun several years before. Researchers have found that in addition to those patients who have a diagnosis of FTLT, upwards of 50% of ALS patients have mild changes in their ability to concentrate, focus on multiple ideas or actions at one time, or make complex judgments, called 'executive functioning'.

Treatment

Physicians have limited choices for treating ALS, and the options that do exist have come into use within the last 10 years. Studies suggest that patients' length of survival and quality of life are enhanced by night-time breathing assistance early in the course of the disease and by aggressive application of alternate feeding options to assure good nutrition once swallowing becomes difficult. At this time, Riluzole® is the only drug that has been approved by the FDA for treatment of ALS. In clinical trials, Riluzole® has shown a slight benefit in modestly increasing survival time (www.ALSA.org.)

Patients with co-morbid FTLT and ALS more often have difficulty following treatment recommendations, perhaps because agitation, poor insight and cognitive changes result in a refusal to comply with physician's recommendations. As a result, life span is sometimes shortened in patients with both diseases. Specific medicines have been found to be helpful to treat the agitation, apathy and depression commonly seen in FTLT.

Caregivers

Caregiving for any patient with a debilitating illness can be both fulfilling and exhausting, and caring for a loved-one with ALS is particularly challenging. Patients' needs change rapidly as the disease progresses, and impaired communication can make caregiving more difficult. Caregivers for ALS patients are also at increased risk for depression and stress because of the emotional and economic stress of coping with an illness that requires expensive equipment and will result in losing their loved one to this terminal disease.

When patients have both FTD and ALS caregiving becomes more difficult for a variety of reasons. Patients often have poor insight into their ALS symptoms, and as a result may disagree with family about clinical changes, treatment recommendations and the need for necessary equipment. They sometimes have trouble using a communication device or making decisions regarding PEG and BiPAP, which are significant life-sustaining measures. All of these problems confound an already very difficult disease for families and health care providers.

Website Resources for Caregivers

[Family Caregiver Alliance](#)

[National Alliance for Caregiving](#)

[National Family Caregivers Association](#)

[The ALS Association](#)