Amyotrophic lateral sclerosis (ALS), also known as Lou Gehrig's disease and motor neuron disease (MND), is a progressive condition caused by the deterioration of the motor neurons in the spinal cord and brain, resulting in paralysis.\(^1,2\) *A*-myo-trophic means "no muscle nourishment" in Greek.\(^1\) The lack of nourishment leads to atrophy, or muscle wasting.\(^1\) "Lateral" denotes the areas where the nerves that signal muscles are located in the spinal cord.\(^1\) "Sclerosis" indicates the scarring or hardening of this region.\(^1\)

The father of neurology, French physician Dr Jean-Martin Charcot, is credited with discovering ALS by correlating a series of case studies occurring from 1865 to 1869.\(^4,5\) Several pioneers in neurology, such as Sir Charles Bell, François-Amilcar Aran, and Charcot's colleague Jean Cruveilhier, described the symptoms of ALS, but did not give it a name.\(^6\) In 1874, Charcot named this disease, "amyotrophic lateral sclerosis," which is still known today as Charcot disease in many parts of the world.\(^4,5,7\) He stated, "The diagnosis as well as the anatomy and physiology of the condition amyotrophic lateral sclerosis is one of the most completely understood conditions in the realm of clinical neurology."\(^6\) Unfortunately, this was an underestimation of the complex nature of ALS.\(^6\)

**Pathogenesis**

The motor neurons are grouped into lower and upper populations.\(^1\) The lower population is located in the spinal cord and brain stem and the upper population is located in the motor cortex.\(^1\) Failure of the upper motor neurons (UMNs) results in brisk reflexes and slowed coordination of the limbs with spasticity and stiffness of the muscles.\(^1\) Disruption of the lower motor neurons (LMNs) is first exhibited by spontaneous muscle twitching, or fasciculations, and then progressively atrophies when the synapses connecting the muscles are lost.\(^1\) This tends to begin in the limbs and progresses to the eye and sphincter muscle neurons in the late stages.\(^7\) When the motor neurons of the spinal cord and brain stem perish, the ventral roots thin and the limb, tongue, and oropharynx muscles become amyotrophic.\(^1\) However, one-third of patients experience bulbar disease, manifesting as challenges with speaking, chewing,
or swallowing.1 Signs of bulbar disease include dysarthria, facial weakness, weakness pushing the tongue outward (pulsion), poor palate elevation, difficulty chewing, impaired swallowing, and more typically noted, tongue fasciculations and atrophy.4

In the spinal processes, the degeneration of the corticospinal neurons causes the spinal cord’s lateral tracts to scar.5,9 Inclusions, or aggregated proteins consisting of round or threadlike shrunken spinal motor neurons, deposit as ALS progresses.9,11 These cytoplasmic inclusions become ubiquitinated in ALS.9,10 Cytoplasmic proteins are commonly present in the motor neurons of sporadic and patients with familial ALS.1 Because ALS has many variants, certain aggregates are only present in particular ALS subtypes.1

### ALS Quality-of-Life Impact

The prognosis for survival in patients with ALS is 2 to 5 years.1,11,12 Despite this grim prognosis, 20% of patients live for 5 years, 10% for 10 years, and 5% for 20 years or longer.13 Patients with an older age at symptom onset, bulbar-onset ALS, lower Amyotrophic Lateral Sclerosis Functional Rating Scale (ALSFRS or ALSFRS-R) score, and early dysfunction of the respiratory muscles have a lower survival risk. However, patients with a younger age at ALS onset, limb-onset ALS, and longer time to diagnosis have independent factors associated with longer survival.14-16

### Etiology

#### Prevalence

Age increases the prevalence and incidence of ALS.1 In the United States, 6000 new patients are diagnosed with ALS each year.14 The prevalence of ALS is 5.0 per 100,000 of the US population, with approximately 16,000 to 20,000 individuals being identified with definite ALS.13,15 The highest prevalence of ALS is in whites, males, and people 60 years or older.17 Globally, the mean age of ALS onset is 62 years.18 In the United States, the Midwest and Northeast have the highest prevalence of ALS, possibly due to higher proportion of whites compared with the South and West.17,19 Potential risk factors for the development of ALS include family history, occupations associated with heavy manual labor, agricultural work, military service (particularly in the Gulf War), football, pesticides, chemicals, smoking, heavy metals, geography, and electric shock.20-26 Studies suggest an association with ALS and the following: cyanotoxins, football, geography, smoking, heavy metals, electric shock, pesticides and chemicals, and military service, but there is no clear evidence to prove causation.24,25

Roberts et al examined the association between socioeconomic status (SES) and race/ethnicity in patients with ALS by using the National Longitudinal Mortality Study (NMLS) records.27 Because ALS is not a reportable condition in the United States, the incidence of ALS was determined by using mortality as a surrogate marker. Mortality information was obtained by correlating the National Death Index (1979-2011) to NMLS records. The study includes information from more than 2 million people: 1,011,172 men and 1,145,368 women. Of these, 1299 deaths were attributed to ALS. The data were collected prospectively. Race was self-reported as black, white, or other races, and ethnicity was reported as non-Hispanic or Hispanic. Non-Hispanic whites had a substantially higher risk of ALS mortality despite adjusting models for SES, health insurance type, or birthplace. Additionally, ALS rates did not differ by gender. Thus, the authors concluded that this finding might be attributed to varying genetic risk factors.22

### Types

There are 2 primary classifications of ALS: sporadic (idiopathic) and familial. Familial ALS occurs in about 5% to 10% of patients with ALS, usually due to a dominant trait.1 Sporadic ALS encompasses all other patients with ALS. The affected population of sporadic ALS comprises approximately 67% males.1 In familial ALS, an almost 1:1 ratio of males to females is noted.1 Sporadic ALS usually occurs in patients in their mid-to-late fifties.1 On the other hand, familial ALS occurs in patients in their late teens or early adulthood.1 Sporadic ALS has no known cause, but it may possibly be caused by immune system abnormalities, toxic exposure, mitochondrial dysfunction, or glutamate toxicity.28

ALS has 4 main presentations: (1) Primary lateral sclerosis (PLS) with pure UMN involvement, (2) limb-onset ALS with a combination of UMN and LMN involvement, (3) progressive muscular atrophy with pure LMN involvement, and (4) bulbar-onset ALS with swallowing and speech difficulties initially and limb features later in the disease course.14,29 PLS involves the corticopontine and corticospinal motor neurons with slight dysfunction of the LMN.15-17 It is characterized by the gradual progression of severe muscle spasticity and stiffness, and modest muscle atrophy.1 PLS cannot be diagnosed as ALS until there is evidence of LMN dysfunction of at least 1 limb or region and it progresses to ALS in most patients.30 Limb-onset ALS is also known as flail leg or flail arm (Vulpian Bernhardt) variants and cannot be diagnosed as ALS until a minimum of 2 body regions are involved.30 Similar to limb-onset syndrome, progressive muscular atrophy (PMA) cannot be diagnosed as ALS until a minimum of 2 body regions are affected, but relevant genetic testing to rule out other motor neuron diseases should also be completed.30 PMA is considered a subform of ALS, based on postmortem findings of UMN dysfunction in a majority of patients, and several having an ALS-causing genetic mutation.30 Bulbar-onset ALS or progressive bulbar palsy (PBP) can be diagnosed as ALS when both UMN and LMN dysfunction is determined.30

### Diagnosis

#### Symptoms

Most patients develop ALS from age 40 to 70 years, with 55 years being the average in the United States.14 There are patients who have developed the disease as early as their twenties.19 Diagnosis is
primarily determined by clinical examination coupled with nerve conduction studies (NCSS), electromyography (EMG), and laboratory testing. NCSs tend to be normal or slightly abnormal with an absence of motor conduction block in patients with ALS. The presence of motor conduction block excludes an ALS diagnosis. Moreover, sensory nerve action potentials (SNAPs) tend to be normal in patients with ALS. Significantly abnormal SNAPs would not suggest ALS and would require further investigation into other potential causes. The use of needle EMG tests for acute and chronic denervation may lead to a diagnosis even before evident clinical onset. Needle EMG is not specific for ALS and must be coupled with extensive denervation in multiple muscle segments of distal and proximal muscles. Furthermore, EMG is used to determine the presence of muscle denervation to differentiate ALS from other conditions.

The primary initial symptom of ALS is progressive, unilateral weakness in the distal legs and arms without remission or relapse. Atypical presentation includes emotional lability, frontal lobe-type cognitive dysfunction, weight loss, and fasciculations and cramps without muscle weakness. Signs of UMN disease include muscle tone increase, slow movement, and hyperreflexia. The presence of the Babinski sign, or upward response of the plantar reflex, is also evidence of UMN dysfunction and is segments of distal and proximal muscles. Symptoms may be described as “limb onset” and “bulbar onset.” With limb onset, patients may experience difficulty with simple actions, such as holding a cup or buttoning a shirt, stumbling more easily, and experiencing changes in their running or walking gaits. Patients with bulbar onset may experience challenges with chewing, swallowing, and speaking, such as nasal or slurred speech. Patients may also experience other symptoms, such as muscle cramps or twitches. Unfortunately, patients with ALS are aware of their gradually declining ability to function. They usually retain higher mental functions, such as problem solving, reasoning, understanding, and remembering. Additionally, patients do not always progress on a linear path. Weeks to months may pass where there is little to no function loss. A rare patient will have considerable improvements, such as recovering lost function. These ALS “reversals” and “arrests” are regrettable fleeting with fewer than 1% of patients maintaining marked improvement for at least 12 months.

The ALSFRS was developed in 1996 to assess activities of daily living (ADLs) in patients with ALS in an easily administered format. In 1999, the ALSFRS was revised and named the Revised Amyotrophic Lateral Sclerosis Functional Rating Scale (ALSFRS-R). The ALSFRS-R is a 12-item scale with each item scored from 0 (unable) to 4 (normal ability) with a possible total score range of 0 to 48. The higher the score, the better the patient is physically functioning. The items evaluate speech, salivation, swallowing, handwriting, cutting food and handling utensils (with or without gastrostomy), dressing and hygiene, turning in bed and adjusting bed clothes, walking, climbing stairs, dyspnea, orthopnea, and respiratory insufficiency. The major difference between the ALSFRS and ALSFRS-R is that the ALSFRS-R weighs respiratory function similarly to bulbar and limb function, possibly increasing the scale’s sensitivity.

Laboratory tests are done in patients with ALS to exclude other conditions. These tests include complete blood count, electrolytes, liver and thyroid function tests, creatine kinase, erythrocyte sedimentation rate, antinuclear antibody, rheumatoid factor, vitamin B₁₂, anti-GM1 ganglioside antibody, serum protein electrophoresis with immunofixation, and 24-hour urine protein electrophoresis with immunofixation. Differential diagnoses include Lyme disease, metal toxicity, peripheral neuropathy, thyroid disease, multifocal motor neuropathy, myasthenia gravis, Guillain-Barré syndrome, and vitamin B₁₂ deficiency. Additionally, a clinical similarity exists between PLS and hereditary spastic paraplegias, with the exception of the pronounced bulbar association found with PLS. In patients experiencing dementia with Pick disease, 15% to 20% have progressive cognitive abnormalities.

Other tests are available, some for research purposes, to aid in the diagnosis of ALS. Examples of testing used for research include magnetic resonance imaging of all regions rostral to symptoms, magnetic resonance spectroscopy, diffusion tensor imaging, and transcranial magnetic stimulation. Cerebrospinal fluid (CSF) analysis should be considered in patients with malignancy, Lyme disease, chronic inflammatory demyelinating polyneuropathy, and HIV, and possibly to obtain a CSF protein profile for future use. Additionally, muscle biopsies may be performed in patients with possible myopathy.

The El Escorial criteria were developed to create a consistent diagnostic process for ALS, guarantee its certainty, and clarify that the varying clinical qualities are complex. These criteria also assisted with determining clinical trial inclusion and exclusion indicators. Despite its original purpose, the staging system of possible, probable, and definite ALS, as determined by the number of affected body areas, has given rise to the ability of researchers to determine if the initial diagnosis has any effect on a patient’s prognosis. Conditions such as PMA, PBP, limb-onset ALS, and PLS have similar pathologic processes to ALS and should therefore be considered for neuroprotective therapies as well.

The Lambert criteria used to diagnose ALS by using EMG was incorporated into both the original and revised El Escorial criteria. The Lambert criteria have since been deemed incomplete and insensitive. Additionally, the studied muscle had to have exhibited both ongoing denervation and motor unit remodeling, decreasing the chances of early ALS diagnosis. In an effort to increase the potential of earlier diagnoses, the Awaji Island criteria were created. The Awaji consensus criteria reformulated the EMG measures to allow for diagnosis based on the presence of either fasciculations in the presence of chronic neuropahtic muscle potentials or fibrillation muscle potentials alone to be markers of acute denervation in patients with clinically suspected ALS.
After all other diagnoses have been excluded, the revised El Escorial criteria–2015 requires at least one of the following: the progression of UMN and LMN dysfunction in at least 1 limb or body region or LMN dysfunction in 1 region identified by clinical examination and/or by EMG in 2 regions (ie, lumbosacral, bulbar, thoracic, cervical). The EMG findings consist of sharp waves and/or fibrillation and neurogenic potentials.

Genetic testing is recommended if the patient has a family history suggestive of ALS. This should be contemplated if a minimum of 1 first- or second-degree relative has ALS and/or frontotemporal ALS, ubiquitinated inclusions resulting from gene mutations are reading frame 72 (C9orf72), prioration and loss of functional activity. Chromosome 9 open reading frame 72 (C9orf72) is the most recent gene discovered. The ALS On-line Database (ALSoD) is a central genetic repository of various gene mutations linked to ALS. It also gives information about patients’ family history, gender, phenotype, geographical data, and age of onset, divided into phenotypic group, gene, or mutation.

The genes are loosely categorized by function: disturbance of distal terminal and motor neuron axon cytoskeletal dynamics, alteration of protein quality control and proteinostasis, and disruption of RNA metabolism, stability, and function. Most mutations are missense substitutions, but C9orf72 is caused by a 6-nucleotide repeat.

C9orf72
C9orf72 is the most common genetic variant detected in patients with ALS. Approximately 25% to 40% of patients with familial ALS and a few with sporadic ALS have a defect in this gene, most especially in patients with FTD. Patients with behavioral variant FTD experience disinhibition, inappropriate social behavior, perseveration, abnormal eating patterns, apathy, loss of empathy, and obsessive-compulsive behaviors.

FUS
Fused in sarcoma (FUS) is a nucleoprotein responsible for the regulation of gene expression, DNA and RNA binding, and mRNA gene splicing. It is found in approximately 5% of patients with familial ALS. FUS shares mechanisms of pathogenesis with TDP-43 and co-localizes with it, resulting in motor neuron granules. FUS mutations are correlated with a rapid progression, young onset, and prominent bulbar manifestations.

SOD1
SOD1, or copper zinc superoxide dismutase, is found in roughly 1% of sporadic ALS and 12% to 20% of familial ALS. The mutations are believed to be caused by autosomal dominant inheritance. SOD1 is responsible for the conversion of the superoxide anion to a less-damaging hydrogen peroxide. However, loss of this function due to mutation does not result in significant motor neuron loss. It is believed that neuron loss is caused by the damage to mitochondrial function and axonal transport. Depending on the variant, SOD1 mutations are linked with rapidly progressing or slow course by mutation of Ala4Val and Asp90Ala, respectively.

TARDBP
Transactive response (TAR) DNA-binding protein 43 (TARDBP) is the most recent gene discovered. NEK-1 is the most recent gene discovered. The ALS On-line Database (ALSoD) is a central genetic repository of various gene mutations linked to ALS. It also gives information about patients’ family history, gender, phenotype, geographical data, and age of onset, divided into phenotypic group, gene, or mutation.

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ubiquitinated, cytoplasmic aggregates or inclusions seen in ALS and a subgroup of FTD. These mutations are found in about 5% of patients with familial ALS and are autosomal dominant. TDP-43, a DNA- and RNA-binding protein, is responsible for the regulation of transport, transcription, stability, and mRNA splicing. It is relocated from its usual position in the nucleus, causing it to accumulate in the cytoplasm of affected spinal cord motor neurons.

**Benefits of Early Diagnosis**

**Clinical Outcomes and Progression**

The time of symptom onset to ALS diagnosis is reportedly about 9 to 12 months. This delay in diagnosis is a challenge to providing patients with early treatment. It is estimated that by the time patients experience ALS signs and symptoms, approximately 50% to 70% of the motor neurons are nonfunctional.

A case-control study by Harrison et al described 36 patients with ALS classified as “ALS reversals” who were compared with 10,723 patients from the Pooled Resource Open-Access ALS Clinical Trials (PRO-ACT) database. Of the control group, 6352 patients had available family history and demographic data available for comparison. Twenty-three patients were diagnosed with clinically probable lab-supported or clinically probable ALS, 7 with PMA, 4 with definite ALS, and 2 with clinically possible ALS. Twenty patients’ diagnoses were verified by chart review and 16 patients through review of the literature. Reversals were measured by ALSFRS-R gains of at least 4 points, denervation resolution as determined by EMG, and/or improved manual muscle testing strength. When compared with the control group, patients with ALS reversal were more likely to take curcumin, copper, azathioprine, fish oil, vitamin D, and glutathione. However, this finding should not be interpreted as a definite correlation, but as a possible hypothesis for future studies. The authors concluded that although all the patients with ALS reversal did not have El Escorial clinically definite ALS diagnoses, the possibility of ALS was high. They additionally suggested that the patients with ALS reversal may have genetic mutations, leading to disease “reversal” via reinnervation, and whole genome sequencing will be performed to further explore this theory. Studying these patients with reversal may lead to a better understanding of ALS and treatments that delay and ultimately cure the disease.

**Best Practices**

A patient with ALS wrote, “ALS patients can have a zeal for life rare among patients with other diseases. Shorter life expectancy often spurs patients with ALS to make life experiences and relationships deeper. It is helpful to understand the concept that ‘everyone has a wheelchair,’ and that no one avoids life’s crises forever.” This quote is a silver lining in the struggles experienced by patients with ALS. It illustrates that despite patients having to accept the challenges of ALS, they should maximize their potential and time. To assist patients with better management, guidance with best practice recommendations has been developed. The ALS Worldwide guidance includes a multidisciplinary approach to manage symptoms and provide patient support. Multidisciplinary team members may include neurologists; pulmonologists; respiratory, occupational, physical, massage, and speech therapists; social workers; nutritionists; support organizations; behavioral health specialists; and pharmacists. According to a review by the Quality Standards Subcommittee of the American Academy of Neurology (AAN), specialized multidisciplinary teams should be considered for patients with ALS to potentially decrease mortality (level B), increase quality of life (level C), and optimize the delivery of healthcare (level B). Additionally, the team may support the organization with achieving performance measures developed by the AAN. The measures include developing and updating a multidisciplinary care plan; cognitive and behavioral impairment screening; offering of therapies for ALS symptoms; inquiring about the patient’s respiratory status and referring the patient for pulmonary function testing; screening for impaired nutrition, weight loss, and dysphagia; offering nutritional support; communicating support referral; reviewing disease-modifying pharmacotherapy; discussing noninvasive ventilation treatment with respiratory-insufficient patients; assisting with end-of-life planning; and assessing the patient for falls.

Neurologists assess, monitor, and treat patients. They are also involved in clinical trials and research that may be beneficial to the patient. The respiratory team, consisting of pulmonologists and respiratory therapists, provides patients with respiratory support because breathing issues are a key symptom in progressive ALS. Occupational therapists are essential to identifying a patient’s challenges with ADLs and assisting them with modifying their current practices or overcoming these challenges. Despite earlier beliefs that exercise damages muscles in patients with ALS, it has been proven to help muscles maintain their power and energy, and the lack of exercise can be harmful. However, patients are usually advised to maintain their current activity level if they are able to do so safely and comfortably. Physical therapists are best suited to assist patients with achieving their exercise capacity. Even though massage may be considered a luxury, it is beneficial to both patients and caregivers due to the physical strain of the disease. Caregivers often lift heavy items and regularly shift patients. Because patients are at risk from aspiration due to loss of muscle function, speech therapists not only assist with language, but also assess the patient’s capability to chew and swallow food.

Social workers assist with direction on the navigation of the social services system, end-of-life-planning, such as advanced directives, and other available resources, such as transportation and support groups for both the patient and patient’s caregiver. Considering that maintaining adequate nutritional stores and caloric intake is essential to life, nutritionists are key members of...
the team. Support organizations are available to assist the patient, caregiver, and researcher. Such organizations include the ALS Association, Muscular Dystrophy Association (MDA), and Motor Neurone Disease Association. Psychosocial support, especially from professionals who understand patients with ALS, is beneficial for the patient, patients’ family, and caregiver. The patient may need support making decisions on matters such as making advanced directives. Additionally, the clinic coordinator may assist the patient with the navigation of the medical process, answering questions, collecting information to relay to the respective healthcare professionals, and addressing the patient’s needs and desires.57

The pharmacist is responsible for managing the medication-related aspect of the patient’s care, educating the patient, and assessing the patient’s medication regimen for potential errors, cost savings, adherence, and preference.57 In a study by Jefferies et al, the 2 major interventions performed by a clinical pharmacist participating on a multidisciplinary ALS team were optimizing medication regimens to manage the symptoms of ALS and medication monitoring.40 Additionally, the pharmacist’s interventions allowed more time for the neurologist to focus on neurological complaints. In general, the pharmacists may also be involved in deprescribing, determining the best medication formulations and delivery devices based on the patient’s current level of functioning and physical abilities, and assisting with medication alternatives based on the insurance formularies.

In addition to the items mentioned above, patients may need genetic counseling. Patients with familial ALS may have genetic testing performed. After taking a thorough medical and family history, a genetic counselor will walk the patient through risk evaluation and genetic testing impact.41 The Genetic Information Nondiscrimination Act of 2008 prohibits genetic discrimination from health insurance providers and employers, but not disability, life, and long-term care insurance.52

There are several diseases with symptoms similar to ALS, and most of these conditions are treatable. Because of this, the ALS Association recommends that a person diagnosed with ALS seek a second opinion from an ALS expert—someone who diagnoses and treats many patients with ALS and has training in this medical specialty.33 The ALS Association maintains a list of recognized experts in the field of ALS. Also, local ALS Association chapters or the national office may be contacted. Groups that provide support for patients and research include the ALS Association, the MDA, and the Les Turner ALS Foundation.12

Early diagnosis, participation in clinical trials, being able to identify signs and symptoms, and referral to a multidisciplinary specialty clinic are considered best practices.37 The Centers for Disease Control and Prevention (CDC)’s National Amyotrophic Lateral Sclerosis (ALS) Registry is the only population-based registry in the United States that gathers information for the purpose of studying ALS.46 Goals of the registry include estimating the incidence and prevalence of ALS, studying risk factors associated with ALS, and providing a database for research to improve the care of patients with ALS.44 The registry collects information, such as gender, age, physical activity, family history, military service information, work history, and environmental and occupational risk factors.44 In addition to collecting survey information, the CDC collects and stores the biological samples of the National ALS Registry patients in the National ALS Biorepository.65 The type of biological samples collected includes proteins, blood, DNA, urine, cells, and tissue.45 The collection also includes a post-mortem component involving the collection of bone, brain, skin, spinal cord, muscle, and cerebral spinal fluid.45 Informing patients about these registries and the impact of their involvement on the future of ALS may lead to discovery of more about various aspects of ALS, such as improved knowledge and treatments.

Conclusions

ALS is a devastating disease. It is difficult to diagnose, debilitating, and has a short survival and poor prognosis for most patients. Unfortunately, there is currently no cure. With the invention of DNA technology, several potential therapy targets have been identified. Through the advancements of medicine and voluntary enrollment of patients with ALS into registries, a better understanding of ALS and therapies will ensue. In the interim, patients should be referred to a multidisciplinary team who will assist them, their families, and caregivers with managing the disease. ■

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