What is ALS and What is the Philosophy of Care?

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Amyotrophic lateral sclerosis (ALS) is a rapidly degenerative disease involving upper motor neuron (UMN) and lower motor neuron (LMN) impairments, for which there is currently no cure. It is necessary for speech-language pathologists to understand the underlying neurological pathophysiology and philosophy of care for individuals with ALS in order to facilitate effective assessment and intervention in this population. The authors of this article review the characteristics of ALS, as well as the general philosophy of care for individuals with ALS. The article covers the topics of assessment and intervention within the context of a multidisciplinary ALS clinic; surgical and pharmacological interventions; and monitoring disease progression, including tracking respiratory status. Periodic multidisciplinary assessment aids in appropriate medical and therapeutic interventions, symptom management, and quality of life. Periodic assessment of respiratory function is especially important to aid in timely delivery of appropriate interventions.

Amyotrophic lateral sclerosis (ALS) is a degenerative motor neuron disease for which there is currently no cure. The cause of ALS is not known, and survival is, on average, approximately 3 years following symptom onset (Corcia & Gordon, 2012). The purpose of this article is to summarize the characteristics of ALS and related types of motor neuron disease (MND), as well as the current philosophy of care for individuals with ALS.

Characteristics of ALS

ALS is a rapidly degenerative disease involving motor neurons in the brain, brainstem, and spinal cord. It is commonly known as Lou Gehrig’s disease in the United States, motor neuron disease (MND) in Great Britain, and Charcot’s disease in France. Typically, the first symptom is weakness; about two-thirds of patients first experience spinal muscle weakness in either the upper or lower extremities, while one-quarter of patients have bulbar weakness and initial dysarthria and dysphagia. Respiratory symptoms are rarely a first sign of disease (Gautier et al., 2010), yet breathing difficulties are a serious complication for almost all patients with ALS, regardless of whether primary weakness is spinal or bulbar.
Though the etiology of ALS is unknown, about 10% of patients have a familial form of the disease (fALS), suggesting the mutation of a particular gene. All other patients are considered to have sporadic ALS, for which no specific genetic abnormality has been identified. The symptoms of fALS and sporadic forms are the same; however, age of onset tends to be about 10 years earlier in fALS and for some progression may be quite rapid.

ALS is characterized by degeneration of both upper motor neurons (UMNs; corticobulbar and/or corticospinal tracts) and lower motor neurons (LMNs). UMN degeneration occurs in pyramidal neurons (Betz cells, 5th layer of cerebral cortex) of the primary motor cortex. LMN degeneration involves alpha motor neurons with cell bodies found in the gray matter of the brainstem and spinal cord (Rocha, Reis, Simoes, Fonseca, & Mendes Ribeiro, 2005).

Diagnosis of ALS requires evidence for degeneration of both UMs and LMNs. Clinically, this is manifested by the neuromuscular impairments of spasticity—the UMN sign—and flaccidity—the LMN sign. Spasticity is a form of muscle hypertonia and is characterized by hyperactive reflexes, positive Babinski signs, stiffness in muscles with slowness in voluntary movements, and resistance to passive range of motion. The presence of muscle atrophy, often accompanied by visible fasciculations, is evidence for flaccidity due to LMN pathology. The neuromuscular conditions of spasticity and flaccidity may be detected in different muscle groups or even in the same muscle. For example, in the bulbar musculature, early signs of spastic lingual muscles contribute to the slow, imprecise articulation characteristic of spastic dysarthria, but progress to a spastic–flaccid dysarthria as the tongue begins to atrophy.

Another form of MND that must be distinguished from ALS is primary lateral sclerosis (PLS). The differentiating characteristic for PLS is that it involves only degeneration of UMs. Thus, patients experience progressive spasticity without significant muscle atrophy. Clinically “pure” PLS is defined as UMN degeneration without evidence of LMN involvement for a period of at least four years (Gordon et al., 2006). Most patients with PLS experience initial spinal symptoms (arm and leg spasticity), but initial bulbar symptoms can occur as well (Gordon, Cheng, Katz, Mitsumoto, & Rowland, 2009). Patients with diagnosed PLS survive and maintain independence much longer than patients with ALS, but most of the principles of assessment and management described for ALS will apply to PLS as well.

ALS with bulbar-onset is often referred to as progressive bulbar palsy (PBP). For patients who survive the initial symptoms of bulbar degeneration, the disease almost always progresses to classic ALS, eventually spreading to the limbs and thorax (Karam, Scelsa, & Macgowan, 2010). PBP presentations can vary: there are patients with only LMN symptoms; those with pure UMN disease, which is referred to as progressive pseudobulbar palsy; or those with the more common combination of LMN and UMN disease (Francis, Bach, & DeLisa, 1999; Rocha et al., 2005; Strong & Rosenfeld, 2003). Symptoms of LMN bulbar involvement include lingual atrophy, usually with visible fasciculations, loss of gag reflexes, and flaccid dysarthria/dysphagia. Symptoms associated with UMN pathology include hyperactive gag reflexes, pathologic release reflexes like the jaw jerk, spastic dysarthria/dysphagia, and disinhibition of more complex motor behaviors like laughter and crying (Caroscio, Mulvihill, Sterling, & Abrams, 1987). Asymmetry in findings is relatively common and reflects the asymmetrical degeneration of motor neurons.

Respiratory impairments eventually occur for most with ALS, contributing to the risk of aspiration and pulmonary complications, and ultimately leading to death (Benditt & Boitano, 2008; Heffernan et al., 2006). Respiratory impairments can occur in any aspect of the mechanical respiratory system: impairments in the respiratory centers located in the medulla, weakness of the respiratory musculature for inspiration and forced expiration (Braun, 1987), and/or dysfunction of the bulbar muscles that help to maintain upper airway patency. Incoordination of swallowing with breathing may also occur (Hadjikoutis & Wiles, 2001; Nozaki et al., 2008). When severe bulbar paresis is combined with severe weakness of the inspiratory
and expiratory respiratory muscles—a “lethal combination”—respiratory failure becomes imminent (Benditt, 2002). See Figure 1 below.

**Figure 1. Functional Effects of MND on the Respiratory System.** From Management of Speech and Swallowing Disorders in Degenerative Diseases, Third Edition (p. 13), by K. Yorkston, R. Miller, E. Strand and D. Britton, 2013, Austin, TX: PROED. Copyright 2013 by PRO-ED, Inc. Reprinted with permission.

**Philosophy of Care**

Offering therapeutic intervention to individuals with a rapidly progressive degenerative condition challenges the standard model of intervention, where improvements are expected following a relatively short duration of intensive therapy. The therapeutic needs of individuals with ALS continually change as the disease progresses. In addition, many individuals with ALS have difficulty with attending intensive multidisciplinary appointments due to issues such as traveling distance or difficulty with transportation when mobility impairments progress (Blackhall, 2012). For these and other reasons, periodic follow-up every few months with reassessment is typically recommended for ALS service delivery, with more intensive bursts of appointments as needed—for example, for augmentative and alternative communication (AAC) assessments or dysphagia intervention. Researchers have demonstrated some therapeutic improvements in the form of maintaining function and lengthening survival in the ALS population (e.g., due to interventions such as non-invasive ventilation, nutrition via gastrostomy tube, and Riluzole). Speech-language pathology intervention can target goals such as maintaining the ability to eat for as long as possible, minimizing risk for aspiration, managing sialorrhea and/or thick mucus, and maintaining the ability to communicate. Clinicians should also educate and counsel patients regarding the nature of dysphagia and dysarthria associated with ALS. Speech-language pathology assessment and interventions are best offered in the context of a multidisciplinary team approach, as described below.

**ALS Clinics/Multidisciplinary Care Centers**

Multidisciplinary care clinics, as advocated by organizations such as the Muscular Dystrophy Association (MDA) and the ALS Association (ALSA), have become a standard of care for individuals with ALS. The Quality Standards Subcommittee of the American Academy of Neurology (AAN) and the European Federation of Neurological Societies (EFNS) recommend that individuals with ALS be referred to specialized multidisciplinary clinics in order to optimize health-care delivery, as participation in such clinics may prolong survival and enhance quality of life (Andersen et al., 2012; Miller et al., 2009b). Participation with a multidisciplinary ALS clinic benefits the patient by ensuring that evidence-based interventions are offered, which promote lengthening survival time, management of symptoms, and end of life planning (e.g., Riluzole, non-invasive ventilation, nutritional management, timely referral to palliative care services; Andersen, et al., 2012). AAN and EFNS periodically publish ALS Clinical Guidelines.
that cover topics such as diagnosis, medicinal intervention, multidisciplinary care, respiratory assessment and intervention, nutrition, communication, and palliative care. However, save for discussing nutrition and timing for percutaneous endoscopic gastrostomy (PEG) placement and generally recommending the involvement of a speech-language pathologist on the multidisciplinary team, no specific guidelines have been offered for assessment, management and treatment of dysphagia. This lack of clear guidelines for dysphagia management in ALS reflects the current paucity of literature regarding dysphagia intervention for individuals with ALS.

Organizations such as the MDA and ALSA provide funding for ALS/MND clinics that offer care meeting their criteria. See below for an example of ALS Clinic Criteria for an ALSA-certified clinic. In addition, ALSA offers certification for ALSA Centers that provide a national standard of care, and are recognized as among of the best in the field (The ALS Association, 2010). Similarly, the MDA also requires MDA/ALS Centers to have multidisciplinary care teams. These Centers allow patients to meet with multidisciplinary professionals on the same dates for routine follow-up, thus reducing the burden of needing to schedule multiple visits. A multidisciplinary clinic provides access to specialists with expertise in a variety of areas: neurology, pulmonology, gastroenterology, rehabilitation medicine, nursing, palliative care, speech-language pathology, nutrition, occupational therapy, physical therapy, as well as social and psychological services. These specialists work together in order to provide coordinated care for individuals with ALS. The EFNS recommends that patients be re-assessed by a multidisciplinary team every 2–3 months—sooner if concerns arise, and less frequently if their disease is progressing slowly (Andersen et al., 2012). ALS clinics are also advised to maintain contact with the patient, caregivers, and family members in the interim. Providers must effectively communicate and coordinate services both within the multidisciplinary team and with the patient’s community services (e.g., primary and palliative care, as well as other home-based services) as well.

ALSA Clinic Criteria (Becky Moore, Executive Director of The ALS Association Evergreen Chapter, personal communication, November 11, 2012), include the following:

- Specialized ALS expertise/knowledge by physician and team
- Attitude of hope, support, encouragement, and aggressive management of symptoms and strategies to maximize function, independence, and patient autonomy
- Multidisciplinary team with an interdisciplinary approach to assessment and treatment
- Routinely organized ALS Clinic days with coordinated multidisciplinary service to ALS patients
- Participation or referral for clinical drug trials
- Chapter and clinic involvement in support groups and educational programs

Patients with ALS face many decisions regarding in which interventions to engage and whether or not to accept life-prolonging interventions. Decisions include which medications or medication trials to accept, whether or not to participate with other therapies such as PEG placement, deciding between non-invasive and/or invasive mechanical ventilation, and considering options related to hospice care. In addition to counseling offered by the multidisciplinary team, palliative care can be helpful with counseling and medical management of symptoms for patients with ALS. Palliative care physicians specialize with symptom management and medical decision-making for individuals facing terminal illnesses (Blackhall, 2012). While most patients with ALS seek palliative care during the disease’s terminal phase, it is optimal to include palliative care earlier in the process along with other multidisciplinary interventions (Blackhall, 2012).
Medical Intervention and Pharmacologic Treatment

Currently, both the AAN and EFNS recommend that Riluzole be initiated as early as possible (Andersen et al., 2005; Miller et al., 2009a). Riluzole is the only FDA-approved medication that has been demonstrated to have a modest effect on survival in ALS (i.e., extended survival for approximately 3 additional months). Riluzole does not affect motor function. It was initially marketed for its ability to inhibit presynaptic glutamate release, as glutamate toxicity has been thought to be related to ALS. However, currently the effect of Riluzole is thought to be more related to blockage of voltage-dependent ion channels (Corcia & Gordon, 2012).

Researchers are currently investigating a variety of medications as a means to slow or treat progression of ALS, including high-dose creatine, tamoxifen, arimoclomol, NP001 (Novartis), CK-2017357 (Cytokinetics), fetal and mesenchymal stem cells, ceftiraxone, mexiletine, and dexpramipexole (Michael Weiss, Director of the Division of Neuromuscular Diseases, Department of Neurology, University of Washington Medical Center (UWMC), personal communication, August 20, 2012). Nuedexta is FDA-approved for pseudobulbar affect but is also being studied for possible reversal of bulbar symptoms (Weiss, 2012). One of the most promising new drugs being studied to treat ALS is dexpramipexole. A phase II trial revealed survival and neuroprotective function benefits (Corcia & Gordon 2012). A phase III trial with this medication is underway.

Often physicians will prescribe other medications to manage specific symptoms such as sialorrhea, mood, spasticity, and reflux.

Monitoring Disease Progression

ALS clinics use the ALS Functional Rating Scale–Revised (ALSFRS-R; Cedarbaum et al., 1999) as a means to track progression of symptoms in individuals with ALS. The ALSFRS-R is an efficient and easily administered instrument that aids in documenting current disease symptoms and progression. The ALSFRS-R is divided into subscales encompassing the following functional areas: bulbar, fine motor (upper extremity), gross motor (lower extremity), and respiratory. The ALSFRS-R is currently the most widely accepted functional outcome scale in research and clinical practice (Shefner, 2008).

Monitoring and managing nutritional status is important, as malnutrition results in faster disease progression (Limousin et al., 2010). For this reason, it is important for clinicians to consult with a nutritionist in conjunction with swallowing assessment and management. Please refer to the article on nutritional and metabolic support in this issue of Perspectives.

Monitoring and Management of Respiratory Symptoms

Management of respiratory symptoms is considered the most challenging aspect of care for patients with ALS (Miller et al., 2009a). To guide appropriate, effective interventions, specific measures of pulmonary function are recommended. Forced vital capacity (FVC) is the primary outcome measure used clinically and can be supplemented by sniff nasal pressure (SnP) and peak cough flow (PCF) measures. Forced Vital Capacity (FVC): FVC is the maximum volume of air that can be exhaled with forced effort following a maximal inspiration. FVC should be measured from both upright and supine positions (Lechtzin, Rothstein, Clawson, Diette, & Wiener, 2002), as supine measures may be more sensitive to changes associated with ALS (e.g., diaphragmatic weakness). FVC is routinely measured throughout the ALS disease process to anticipate and assess respiratory care needs. Despite differences in the rate of disease progression and site of initial presentation, all patients with ALS exhibit a linear decline in their FVC scores over time (i.e., average decline of 3.5% per month; Fallat, 2002). The decreasing slope of FVC is the primary clinical indicator of disease progression and predictor of survival (Lechtzin et al., 2002). Thus, all patients with ALS should have their FVC assessed every 3 months (Miller et al., 2009a). When FVC falls within a critical range (i.e., 50% of their predicted scores), it is used to determine when to initiate non-invasive ventilation and PEG tube feedings (Miller et al.,
An FVC of 20%–30% signals transition to end-of-life care, as it is associated with a significant risk of respiratory failure and death. Hence, FVC taken at regular intervals (every 3 months, minimally) is essential to anticipating care needs and the timing of interventions for symptom management (Miller et al., 2009a). Despite the importance of FVC measures in the care of patients with ALS, FVC should not be used as the sole measure of respiratory function. FVC is a global measure of lung capacity and has low specificity and sensitivity to mild respiratory weakness, a challenge common among measures obtained using spirometry (American Thoracic Society/European Respiratory Society, 2002). In patients with ALS, the volitional nature of the task and the physiological demands of performing the FVC maneuver can make it difficult to obtain necessary values. As such, it is often necessary to include other tests of pulmonary function to complement results of FVC testing.

Sniff Nasal Pressure (SnP): SnP is a non-invasive test that provides an indication of diaphragm function and intra-thoracic pressure (Lechtzin et al., 2002). The maneuver involves a short, sharp volitional inspiration. Pressure is measured with a transducer via a catheter inserted into one nostril. Patients with ALS, even in the later stages, are able to perform the sniff task. Normative data are available by age group (Uldry & Fitting, 1995). SnP may detect mild changes in inspiratory muscle strength over time (Fitting, Paillex, Hirt, Aebischer, & Schluep, 1999).

Peak Cough Flow (PCF): PCF is a measure of airway clearance ability. Maximal PCF depends on a coordinated sequence of inspiration, compression with laryngeal adduction, and expiration with active laryngeal abduction. It is measured using spirometry or a simple peak flow device. Effective PCF is needed to avoid pulmonary complications associated with secretion encumbrance and poor airway clearance (Bach, 1993). Over the past few years, clinicians have recognized the importance of cough augmentation and airway clearance support, and serial measures of PCF are now standard practice in neuromuscular respiratory care (see Cleary and Richman Eisenstat, this issue, for a review of respiratory interventions in ALS). In fact, therapeutic thresholds for PCF have been reported (see Table 1). These thresholds are important in the timing of cough and airway clearance interventions (Boitano, 2006). For example, in individuals with respiratory weakness and a concomitant respiratory infection, the mucociliary escalator becomes overwhelmed after repeated bouts of coughing. Fatigue then sets in, and individuals can lose the ability to muster even minimum levels of PCF (160 L/m) to clear the airway. In fact, the risk of airway encumbrance increases with PCF levels less than 270 L/min (Boitano, 2006). Therefore, it is recommended that preventative airway clearance strategies (such as lung volume recruitment, mechanical in-exsufflation, and/or manual assisted coughing) be initiated when patients with ALS present with a PCF of greater than 270 L/min (McKim et al., 2011).

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<tr>
<th>PCF Level</th>
<th>Clinical Significance</th>
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<tr>
<td>Greater than 500 L/min</td>
<td>Typical threshold for healthy adults; minimal risk of airway encumbrance</td>
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<tr>
<td>Less than 270 L/min</td>
<td>Increased risk for airway encumbrance</td>
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<tr>
<td>160 L/min</td>
<td>Minimum threshold to move mucous from lungs into the upper airway</td>
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(Bach & Saporito, 1996; Boitano, 2006; Toussaint, Boitano, Gatot, Steens, & Soudon, 2009)

Conclusions

ALS is a rapidly degenerative disease involving UMN and LMN impairments. Periodic multidisciplinary assessment facilitates appropriate medical and therapeutic interventions, symptom management, and quality of life. Periodic assessment of respiratory function is especially important to aid timely delivery of appropriate interventions.
Additional Resources

- ALS Association: www.alsa.org
- Muscular Dystrophy Association page on ALS: http://mda.org/disease/amyotrophic-lateral-sclerosis

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References


