ALS for the non-expert: Review of the Guidelines for treatment

Every Tuesday as I make my way into work I think of the book, “Tuesdays with Morrie.” Tuesday is the day when our hospital holds the ALS Multidisciplinary Clinic, and the patients and families I see in clinic remind me of the courage portrayed in Morrie’s story. In my head I change “Morrie” to Phil or Sandy, or whomever I know is coming to the clinic that day. This is a little ritual I have developed to get into my mind the incredible individuals I will be seeing over the next 7 hours. Those of us who are familiar with the signs and symptoms of motor neuron disease (MND) are undoubtedly also familiar with the inspirational spirit that so many of these patients and families exhibit. But it is likely the case that many clinicians rarely have the opportunity to work with this population, to be inspired by them, and to learn the signs, symptoms, and treatments of MND.

As a speech-language pathologist you are likely familiar and comfortable with diagnosing aphasia or memory deficits, but would you know it if a patient in the early stages of bulbar Amyotrophic Lateral Sclerosis (ALS) was sitting in front of you? If you are a speech-language pathologist who sees adults, you should have the skills needed to perform a thorough and diagnostic motor speech examination, and should be confident enough in what you see to formulate a strong suspicion and thereby refer a patient for appropriate medical intervention. Based on U.S. population studies, over 5,600 people in the U.S. are diagnosed with ALS each year, thus it is estimated that as many as 30,000 Americans have the disease at any given time. If you work with adults, you will likely see a patient with ALS at some point, regardless of where you practice.

“Amyotrophic” is from the Greek and means roughly, no muscle nourishment. "Lateral" refers to the spinal cord, which houses portions of the nerve cells that signal and control the muscles. In this disease, these nerve cells degenerate, resulting in scarring or hardening, known as “sclerosis.” ALS is a progressive neurodegenerative disease that affects nerve cells in the brain and the spinal cord, and the progressive degeneration of the motor neurons in ALS eventually leads to neuron death. When motor neurons die, they can no longer send impulses to the muscle fibers that should result in muscle movement, resulting in the loss of control over muscle movement. As voluntary muscles are affected, patients have less and less motor control and in the later stages of ALS, they may become totally paralyzed.

Early symptoms found in ALS often include increasing muscle weakness, especially involving the arms and legs, but may also include weakness in the muscles used for speech, swallowing or breathing. The early symptoms may be so subtle that they are overlooked or ignored. The symptoms seen in early bulbar ALS may be twitching (fasciculation) of the tongue, nasal regurgitation, “thick speech” or a strained/strangled vocal quality, but this is not an exhaustive list. To be diagnosed with ALS (versus other motor neuron diseases such as Primary Lateral Sclerosis or Bulbar Palsy) patients must exhibit both upper and lower motor neuron signs.

If a patient’s first symptoms of ALS or other motor neuron diseases are bulbar, the patient may present to a speech-language pathologist due to difficulties with speaking, swallowing, or with other oral motor functions. For example, the patient may report to
their primary care physician (PCP) complaining of subtle heaviness in the tongue at night when he is tired, or of feeling that with sticky foods like peanut butter sandwiches, he always needs a few extra sips of water to get the bite down. She may complain that when she brushes her teeth at night she consistently gets water up her nose while bending over the sink to spit. The PCP may refer to speech-language pathology after hearing these symptoms. If you are a keen observer and clinician, you may be the first professional to initiate the necessary extensive neurological work-up. If you do not recognize subtle signs and symptoms, the patient may continue down a long road of visits before an accurate diagnosis is reached.

Dysarthria assessment is one of the few areas in the field of speech-language pathology where our diagnostic skills can be invaluable to other health care providers. (We certainly are invaluable in many other ways as well, but most patients come to us with a diagnosis already). During this evaluation, diagnostic information about upper and lower motor neuron signs may become apparent to us. While we can not diagnose a patient with ALS, we can identify when it is time for an urgent referral to a neurologist specializing in neuromuscular disorders, thereby expediting the diagnostic process. But our role does not stop with the diagnostic phase.

Once a patient has received this devastating diagnosis, we should be part of a multidisciplinary team that cares for the patient in the difficult journey ahead. As a member of this team we constantly assess the patient’s speech and swallowing, offering strategies for minimizing difficulty and maximizing function. The team ideally consists of the neurologist, nurse practitioner, respiratory therapist, physical therapist, occupational therapist, dietician, social worker and speech pathologist. In addition, most patients attend a clinic only once every two to three months, therefore it is critical to be available via e-mail or phone to respond to between visit questions.

In October 2009, the American Academy of Neurology (AAN) issued guidelines for the care of patients with ALS. Though there is no cure for ALS at this time, Dr. Robert Miller of the California Pacific Medical Center, who was the lead author of the new guidelines, has looked at all of the data collected to formulate the list. He reports that “While we are waiting for a cure, people need to know that a lot can be done to make life easier and longer for people with ALS.” The new guidelines stipulate the following:

- **Patients should be offered Riluzole.** While there is not a cure or treatment to date that halts or reverses ALS, riluzole, an FDA approved drug, has been found to modestly slow the progression of ALS.
- **If patients have trouble swallowing and chewing, they should be offered a feeding tube.** Use of the tube for feeding can help stabilize a patient’s weight or assist with small weight gain. This should be done while patients still have good forced vital capacity (FVC) (50% or greater).
- **Patients should be offered noninvasive ventilation (such as BiPAP) at the earliest signs of respiratory insufficiency.** This can slow the rate at which FVC declines, decrease overall fatigue, help with maximizing cognition and prolong life.
• Patients with a weak, non-productive cough should be offered an in-exsufflator (cough-assist device). If secretions can’t be cleared due to progressive weakness of respiratory muscles, a device is needed to help with clearance to increase comfort and reduce risk of infection.

• Patients should attend a multidisciplinary ALS Clinic. The specialists can work together to coordinate care for the patient. Studies have shown that patients who attend the multidisciplinary clinics have better symptom management, maintain as much independence as possible, have an improved quality of life, and a prolonged survival. The participation in a multidisciplinary ALS Clinic also allows for ongoing, individualized treatment as everyone with the disease has a slightly different experience. These patients are more likely to take Riluzole, get a feeding tube early, and use non-invasive ventilatory support.

• If patients have excess saliva that is not responding to medications, they should be offered botulinum toxin. Botox causes the salivary glands to produce less saliva.

• Patients should be screened for cognitive and behavioral changes. A small number of patients with ALS do develop fronto-temporal dementia, and require a health care proxy when it comes to making medical decisions (to get a feeding tube, etc). A much larger number develop some degree of cognitive and/or behavioral impairment. These deficits can lead to a lack of understanding about long-term care choices, etc. so need to be detected to be sure information is delivered in a manner commensurate with the patient’s level of understanding.

Whether a patient is seen for a speech and swallowing evaluation in a multidisciplinary clinic or in your office, it is critical that the speech-language pathologist delivers information in an honest yet sensitive way so as not to impart a feeling of hopelessness or gloom and doom. There are many things we have to offer that significantly improve quality of life. As the weakening and paralysis spreads to the muscles affecting speech, swallowing, chewing and breathing (which may be early with bulbar onset and later in spinal or limb) the services of the speech-language pathologist are critical. Many patients may come to you seeking exercises or therapy routines to “fix” the problems. It is critical that we are candid with patients. Oral motor exercise does not improve speech and in fact is contraindicated as it may hasten fatigue and have a negative impact on intelligibility. Instead we need to focus on modifying the environment and teaching simple compensations to help facilitate verbal communication or to transition to non-verbal means as needed. We also need to point out the importance of energy conservation so that patients have the energy they need for what they deem most important. If they want nothing more than to read a story aloud to their grandchild, they should plan on doing it early in the day, and be sure to have rested before hand, and not chewed a bagel for breakfast. But if the bagel is all they want, skip social hour and have it! They make the choices, but you can support them by giving them the tools that make their desire a reality. If they are having a lot of choking and their aspiration risk is high, explain what that means and the options that are available to them (thicken liquids, G tube) but once they have made an informed decision, support their choice and teach them the best and safest way to proceed without passing judgment. Since ALS attacks only motor neurons, the sense of taste and smell are not affected, and many patients still love to eat even when it seems unbelievably effortful or dangerous to us.
Finally, as most patients (between 60% and 75%) with motor neuron disease will eventually need some sort of alternative communication device, you should be comfortable facilitating a referral to aid your patient in getting an appropriate device. If you are not familiar with these tools and the complex assessment that is needed to select the appropriate device, find a speech pathologist or clinic in your area that can do a comprehensive assessment, but be sure they are familiar with ALS! There are many factors that have to be considered (e.g. What will the patient’s hand function be as the disease progresses? How is their vision, hearing, etc? Are they having any cognitive impairment that will preclude them from learning a complex communication system?). And patients should be given several options and allowed trial periods with various devices before one is ordered. This process should start as early as possible, well before they are reliant on it to communicate, as delivery can take months. Further, many families will need help with the funding issues that arise as dedicated AAC devices are quite costly (typically $2000 to $15,000).

What I find so remarkable about patients with ALS, and their families, is that despite the grave nature of the diagnosis, they have an unbelievable sense of hope, resilience, and tenacity. They know what the future likely holds, but are determined to go forward on their own terms. We, as speech-language pathologists, are in a position to help them along on this journey by providing the necessary tools, strategies, education, and support to make the best of each valuable day.

Resources for professionals and patients:

www.alsa.org
www.mda.org
www.ninds.nih.gov/disorders/amyotrophiclateral sclerosis.htm

Bibliography: