CLINICAL VIGNETTE

Advanced Care Planning in a Patient with Amyotrophic Lateral Sclerosis

Balbir S. Brar, MD and Michael E. Lazarus, MD

An emaciated 60-year-old man was brought to the emergency department of a community hospital by his family for diffuse weakness. The patient was found to have a low sodium level of 128 mmol/L and is admitted to the UCLA hospitalist service. Further history revealed that he had been diagnosed with Amyotrophic Lateral Sclerosis (ALS) five years prior. He did not have any history of abdominal pain, dysuria, or loss of consciousness nor cough, dysphagia, odynophagia or difficulty breathing. The patient's family reported he had been more lethargic and tired over the preceding few days with loss of appetite and decreased oral intake. His presentation was similar to a previous admission with symptomatic hyponatremia with serum sodium of 126 mmol/L a few months earlier. His wife noted that he was having difficulty with movement as well as significant insomnia. Unfortunately, no comprehensive neurological evaluation had been performed for several years. He had never seen an ALS specialist and was not participating in any ALS clinical trials. His previous medical history was notable for hypertension, nephrolithiasis and benign prostatic hypertrophy. The patient was on ipratrpium two puffs every twelve hours, tamsulosin 0.4 mg nightly, metoprolol succinate 50 mg tablet once daily and duloxetine 60 mg daily. He was also on riluzole 50 mg twice a day and more recently had started on an edaravone infusion. He was a lifelong non-smoker and denied alcohol use. Patient lived with his son, who was the primary caretaker, as well as his wife. On physical exam, vital signs are normal, there is weakness and muscle wasting of his bilateral upper extremities, including the intrinsic muscles of his hands. Spontaneous muscle twitches are present in his bilateral thumbs, gastrocnemius and paraspinal muscles. Muscle weakness is noted in bilateral lower extremities with brisk deep tendon reflexes throughout. Sensory perception and cranial nerve exam are normal. During the hospitalization, the patient's sodium corrected with fluid restriction to 1500 cc per day. He was alert and oriented to person, place and time. He was diagnosed with Syndrome of Inappropriate Antidiuretic Hormone excretion (SIADH) secondary to duloxetine and this medication was changed to an alternative serotonin reuptake inhibitor (SSRI). He had previously seen a psychiatrist for anxiety and depression but it was not clear whether he was getting appropriate counseling and was not part of a comprehensive ALS treatment plan. Additionally, there was no advanced directive on file. A palliative care consultation was requested by his hospitalist to address these issues. Over the next several days the palliative team discussed all the alternatives and options available for life sustaining treatment. He affirmed that he wanted to have a gastrostomy-tube (PEG) as well as tracheostomy inserted if needed when his disease advanced to affect swallowing or breathing. Unfortunately, he did not discuss this option with his family prior to admission and he was not certain of his prognosis. There was strong sense that he was in denial regarding the gravity of his ALS diagnosis. He was also hesitant to talk about his condition in front of family members. One day before projected discharge, his condition deteriorated with increasing dyspnea requiring supplemental oxygen and positive pressure ventilation (BiPAP). The patient did not tolerate non-invasive ventilation and became progressively obtunded. He was diagnosed with acute hypercarbic respiratory failure after arterial blood gas analysis. He was offered intubation, which he declined. Later that day after intensivist consultation and discussion with the family and palliative team, he was placed on comfort measures and expired peacefully a short time later. The palliative team supported family members who were understandably distraught given his 'sudden' deterioration and lack of advanced planning.

Discussion

Amyotrophic lateral sclerosis (ALS) is a progressive neurodegenerative disorder that causes muscle weakness, disability, and eventual death, with a median survival of 3-5 years.[1] Symptomatic management is the mainstay of treatment of ALS. More importantly, management of the patient using a multidisciplinary approach as the disease progresses through its different stages is key. The heterogeneous clinical presentation and wide temporal range of disease progression make the diagnosis of ALS challenging especially for the non-neurologist. No single diagnostic test has been shown to definitively diagnose ALS. Suspicion should be high in patients presenting with progressive weakness with evidence of lower motor neuron and upper motor neuron involvement, and that importantly, no other disease processes adequately explain. Informing the patient and the family of the diagnosis of ALS is a daunting task for the clinician but once confirmed should be given with sensitivity and allow the patient and caregivers time to fully understand the condition and its implications.[2] Many patients have delayed diagnoses that can be distressing and time consuming. For these patients, timely discussion reduces the uncertainty and makes way for planning how symptoms can be treated, as well as the role of disease modifying treatments such as riluzole. This is the only drug known to offer a survival benefit, which is a disappointing three months at best.[3] Treatment offered in a multidisciplinary ALS clinic setting is the standard of care. Studies show that patients with ALS who are

followed by multidisciplinary or tertiary ALS centers have improved survival compared with those who were followed by general neurology clinics.[4] As the disease progresses, the focus of care shifts. An understanding of the disease trajectory enables the clinician to anticipate when care priorities are likely to alter and to pre-empt complications. In the three quarters of patients who present primarily with limb weakness, the initial therapy is weighted toward providing physical therapy, assistive devices and relief of symptoms, such as fasciculation's, muscle cramps and spasticity. Towards the latter stages, more complex interventions may be required depending upon the patients goals of care. These include noninvasive ventilation (NIV) and gastrostomy (PEG tube) feeding. In a quarter of cases, dreaded bulbar dysfunction leads to speech and swallowing disturbance as the hallmark signs. These individuals need early speech therapy and optimization of communication and significant nutritional support. Hyperalimentation is usually required earlier in the disease course than in those with pure limb-onset disease.[5] A primary care physician and different sub-specialists cared for our patient. Unfortunately, he lacked the benefits of a comprehensive and holistic care approach with modifications as his symptoms progressed. No counseling or social service discussions had taken place despite two hospitalizations. He had not planned for any financial consequences nor made plans for the living arrangements for his wife after death. Our patient's "unexpected' death caused significant financial and psychological distress for his survivors. Multidisciplinary ALS clinics provide care from neurologists, physical therapists, occupational therapists, speech therapists, respiratory therapists, dietitians, social workers, and nurses.[6,7] These specialized clinics guide the management of the complex issues of ALS, which include respiratory symptoms, anorexia, dysarthria, dysphagia, functional decline and psychosocial problems.[8] In addition, multidisciplinary care improves quality of life. Specialized multidisciplinary clinic referrals and early palliative care should be considered for all patients with ALS. By optimizing healthcare delivery, there is evidence of prolonged survival, and enhanced quality of life.[9] As modern healthcare keeps on making progressive and positive enhancements, it is imperative that all patients with ALS have access to this multidisciplinary approach. Similarly, European guidelines note that multidisciplinary care should be available for all people affected by ALS.[10]

ALS is a chronic progressive disease and early integration of care can provide an adequate layer of support to help address progressive disability, symptom burden, and family and caregiver stressors.[10] Involvement of palliative care does not have to be limited to end of life care. Comprehensive palliative support includes establishing goals of care respecting the patient's values and preferences; ensuring consistent and sustained communication between the patient and all caregivers; psychosocial, spiritual, and practical support to both patients and their family members; and coordination across all sites of care. Evidence suggests that specialty palliative care services can be successfully integrated into ALS care and are associated with improved quality of life and a reduction in intensive care unit use and length of stay in the hospital.[11]

Amyotrophic lateral sclerosis (ALS) has traditionally been considered a neuromuscular disease, despite the degeneration affecting both upper motor neurons and lower motor neurons. Recent clinical, imaging, and neuropathological data have demonstrated more extensive involvement of the central nervous system than was previously recognized. Population-based data show that up to half of all patients with ALS develop cognitive and behavioral impairment, and about 13% of patients have concomitant frontotemporal dementia.[12] Recognizing cognitive impairment in ALS patients is significant for several reasons. It may be associated with genetic mutations of relevance to offspring, portend a more aggressive disease course, non-compliance with treatment and increased caregiver burden. These issues are vital to address as early as possible as capacity and power of attorney issues become paramount.

In retrospect, had our patient been offered early multidisciplinary and palliative services we may have mitigated much of the distress that occurred during the advanced stage of this disease and in the last days of this patient's life.

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