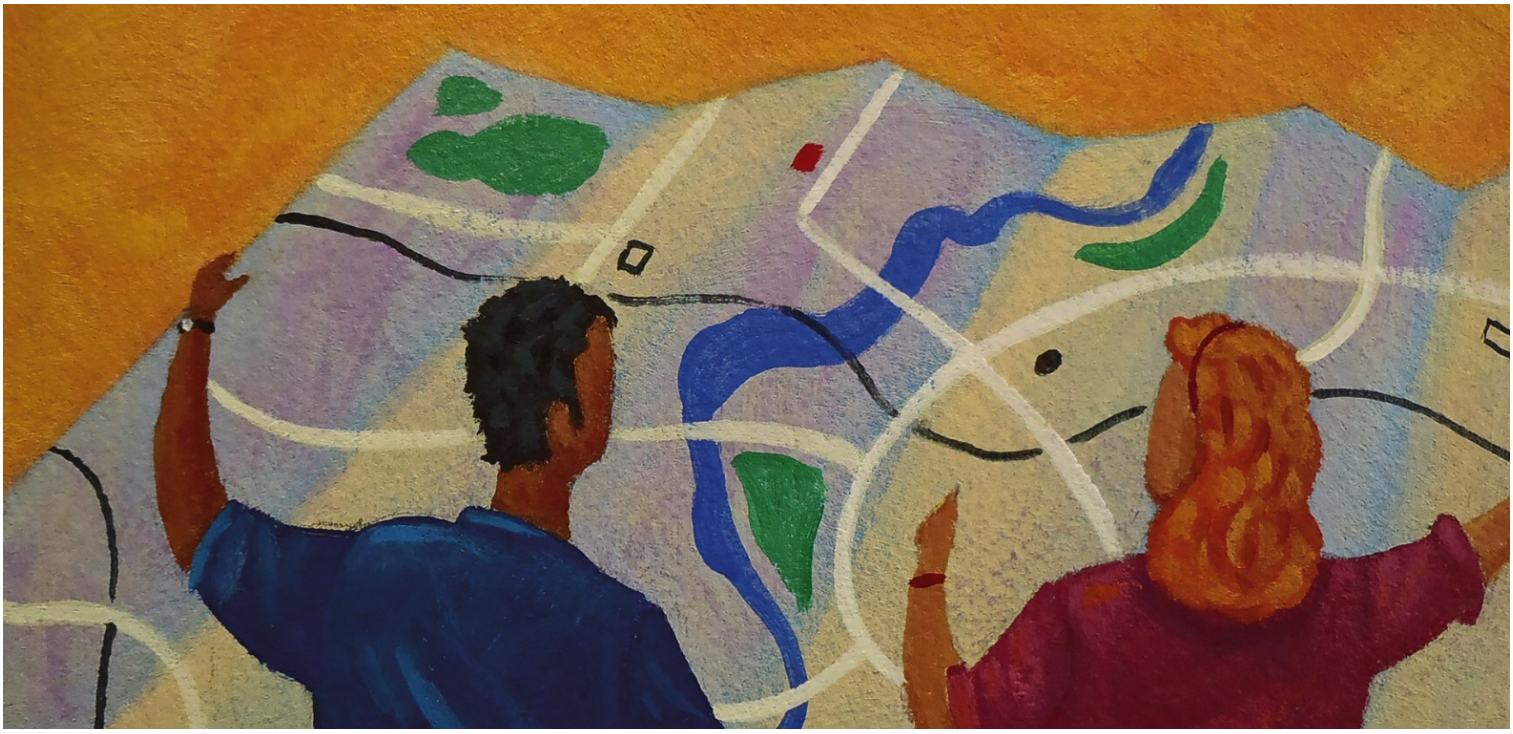
THE BASICS OF ALS FOR MENTAL HEALTH CLINICIANS

From the Massachusetts General Hospital
ALS Parenting At a Challenging Time (ALS PACT Program)



Mental health clinicians frequently work with people who have medical challenges in addition to mental health vulnerabilities. Sometimes an individual's adjustment to a new or changing medical condition is the primary focus of therapy. Sometimes medical concerns remain in the background, or only emerge as treatment for other issues is already well underway. It can be time consuming to find pertinent and reliable information on the range of medical conditions we may encounter as therapists, but in many cases, understanding the day-to-day experience of living with a particular illness is vital to our ability to provide a patient or client with what they need.

ALS (Amyotrophic Lateral Sclerosis) is one such case. Because it is relatively rare, most people do not have direct experience with it- in contrast to an illness like cancer, that is often all too familiar in our own lives. However, people affected by ALS tell us frequently how difficult it is to really "connect" with a therapist who seems to have no idea what their lives are like.

This document is written specifically with mental health clinicians in mind. It aims to provide an accessible overview of some key physical and medical aspects of living with ALS. It represents a collaboration among clinicians of varied professional backgrounds from the Massachusetts General Hospital ALS Multi-disciplinary Clinic. We hope that it provides information that will be useful to you in any work you do with individuals or families affected by ALS.

"The Basics of ALS for Mental Health Clinicians" is part of a bigger project funded by the ALS Finding a Cure organization (ALS-FAC). This project, "Parenting with ALS: Guidance for Supporting your Children" comprises a series of handouts for adults with ALS and their partners offering parent guidance for supporting children's adjustment to the parent's illness, a guide for members of an ALS Multi-disciplinary Clinic, and this guide for mental health clinicians. These parent materials may also be useful to you. They discuss, for example, how parents can help children prepare for and adjust to the range of functional changes associated with ALS that are described below. They also address many of the emotional aspects of living with ALS. Some of these will be familiar to most mental health clinicians, but others may be less so. For example:

- Emotional impact: ALS can have significant emotional and psychological effects on patients and their families. Mental health clinicians can acknowledge these challenges, such as grief, anxiety, depression, and difficulties adjusting, and provide a supportive and compassionate environment for processing and problem solving.
- Loss and grief: Living with a progressive disease like ALS entails ongoing loss and anticipatory grief. Patients will benefit from support to help them adapt to their constantly changing circumstances.
- Caregiver support: Living with ALS often requires extensive caregiving, which places significant stress on family members—children as well as adults. Mental health clinicians can help address the emotional burden, the risk of caregiver burnout, and the need for respite care.
- Parenting and co-parenting: ALS affects the parenting experience in a variety of ways. Changes may occur in the ways a parent is able to engage physically and to communicate with their children due to speech and motor function symptom progression. Parents and co-parents may also experience worry, sadness, and/or guilt. All of this may contribute to lower parenting self-efficacy and enjoyment. For co-parents, the balancing act of caring for children as well as for the partner can significantly change the parenting experience. Both parents can benefit from monitoring for mood disorders, opportunities to process a range of emotions and to re-imagine their identities as parents.

The parent guidance materials in this project can be freely accessed here:
https://www.massgeneral.org/als-pact

Key Concepts:

This document provides information about:

1. Basics of ALS—symptoms, prognosis
2. Genetic forms of ALS
3. Clinical course
4. Diagnostic workup and delays in diagnosis
5. Treatment: multi-disciplinary care and medications
6. Mobility challenges, interventions and equipment
7. Speech challenges and interventions
8. Swallowing and eating challenges and interventions
9. Respiration (breathing) challenges and interventions
10. Palliative care and hospice

Overview

Amyotrophic Lateral Sclerosis (ALS) is a fatal, neuromuscular disease that causes progressive muscle weakness throughout the body, eventually leading to death within 2-5 years of diagnosis on average. The rate of progression can vary considerably: people with slower progressing forms may survive for years, but others with rapid progressing forms face quick decline and may die within a year of diagnosis. ALS causes motor neurons (nerve cells in the brain and spinal cord that connect to and control muscles) to die, causing primary symptoms of:

- progressive weakness in the limbs (typically starting distally, e.g., hands or feet)
- weakness in muscles that control speech and swallowing (bulbar symptoms)
- weakness of the diaphragm, leading to respiratory failure and death.

The onset of muscle weakness can be subtle and is often first noticed only when it begins to impair function – e.g., trouble fastening buttons or turning keys due to hand/finger weakness, altered gait, choking/problems swallowing, or changes in the quality of speech.

Genetics

According to the CDC, the prevalence of ALS is about 4-6 people per 100,000. About 5-10% of ALS is caused by an inherited genetic mutation (Familial ALS); the remainder are considered Sporadic ALS as there is no family history or identifiable gene. As genetic testing becomes more widespread in clinical practice, more ALS-causative genes are being identified. There are currently over 30 known genetic mutations that cause or increase the risk of developing ALS.

ALS genes have an autosomal dominant inheritance pattern, i.e., only one mutated copy of the gene is required to cause ALS, and a person affected with familial ALS has a 50% chance of passing the gene on to each offspring. The most common genetic mutations that cause ALS are:

- C9orf72 (“C9”) mutation. Currently the most common genetic cause of ALS worldwide. Humans have two copies of the C9 gene, one from each parent. In individuals with C9 ALS, one of the copies of C9 gene has hundreds to thousands of repeated sequences of a specific six-digit string of DNA base pairs (hexanucleotide repeat expansion), while normal C9 genes typically just have a few (<10) repeats of this sequence.

The C9orf72 mutation is associated with both ALS as well as Frontotemporal Dementia (FTD). Individuals can develop just ALS, just FTD, or both ALS and FTD.

- SOD1 mutation. SOD1 is the first causative ALS gene discovered. The SOD1 gene comes in many different variants that affect the likelihood of expressing disease and disease progression. The most common variant, SOD1 A5V, causes the most rapid onset and shortest survival time, typically within one year of diagnosis.

Family Members of Familial ALS Patients

- Predictive genetic testing can help family members learn if they have an increased chance of developing ALS. It can also impact family planning decisions, e.g., Preimplantation Genetic Testing (PGT) can allow for the selection of embryos without a genetic abnormality, when undergoing In-Vitro Fertilization (IVF).
- Genetic testing for ALS is not available for children.

Clinical Course

ALS presents heterogeneously; while progressive muscle weakness is the hallmark of the disease, the age and location of symptom onset and rate of progression can vary from person-to-person and cannot be predicted. Certain genetic variants have been found to cause faster progression than others.

Limb symptoms:

- Weakness in a distal limb (e.g., hands or feet), (typically on one side at first) which progresses proximally (e.g., arm or leg) and spreads to other limbs.
- Spasticity, an abnormal tightness of muscles, can occur in ALS causing rigidity and stiffness that prevents smooth movement.

Bulbar symptoms: Weakness and spasticity in the muscles of the tongue and throat causes:

- Dysarthria of speech (slurred words, slowed speech, increased nasal quality, reduced volume, hoarseness), eventually leading to anarthria (inability to speak).
- Swallowing difficulties, drooling (sialorrhea) and choking.
- Early recognition of bulbar symptoms is important to initiate interventions like Augmentative Communication Devices and Gastrostomy Tubes.

Respiratory symptoms: Weakness in the diaphragm and other respiratory muscles that progresses until respiratory failure and death.

Cognitive symptoms: 50% of people with ALS are affected cognitively.

- **Pseudobulbar Affect (PBA)**: PBA affects the brain's ability to regulate emotions, causing unpredictable, atypical emotional responses that are exaggerated, or don't match the situation or how someone is feeling, e.g., a sustained outburst of laughter at a mildly amusing joke, or crying at a festive parade.
- 15 to 45 percent of all ALS patients experience PBA. This condition can affect multiple aspects of life including occupation, relationships, sociability, and self-image. People with PBA may isolate themselves due to anxiety or to avoid embarrassment. PBA is thought to be caused by a disruption in the brain's neural circuits specifically where emotions are regulated.
    - Neudexta (see **Medications** below) has been approved to treat PBA. Tricyclic antidepressants can also be used.

Frontotemporal Dementia (FTD): Several ALS genes (most commonly, C9orf72) also cause FTD. People with a C9orf72 mutation may develop ALS, FTD, or ALS with FTD. FTD is a group of disorders caused by the degeneration of the frontal lobe that can manifest in several ways:

- Behavior: Personality changes marked by apathy, disinhibition and impulsivity, difficulty planning and executing activities, socially inappropriate behavior, hypersexuality, and/or social withdrawal
- Speech: Effortful speech, Impaired word retrieval, difficulty with longer phrases and sentences
- A hallmark of FTD is that patients lack insight and do not believe that they have any symptoms.

Neuropsychological tests and brain imaging can help diagnose FTD, in conjunction with a thorough clinical history.

There are currently no medications approved to treat FTD.

Diagnostic Workup

There is currently no single definitive test or biomarker for diagnosing ALS. Diagnosis is complex and requires a combination of progressive muscle weakness and signs of pathology in upper motor neurons (neurons that connect the brain and spinal cord) and lower motor neurons (motor neurons that originate from the spinal cord and send signals to muscles throughout the body). These signs include muscle weakness, abnormal (hypo or hyperactive) reflexes, muscle wasting (atrophy), and involuntary muscle twitches (fasciculations). Neurological exam findings suspicious for ALS are often evaluated in conjunction with Electromyography (EMG), a test to find abnormalities in the electrical activity of muscles.

The “Revised El Escorial Criteria” is a commonly used framework for diagnosing ALS. It is a complex algorithm that considers factors such as the presence of lower or upper motor neuron signs, the number of body regions affected by these signs, EMG results, as well as genetic testing results. Depending on the combination of factors, the El Escorial criteria categorizes ALS diagnoses as “suspected”, “possible”, “probable”, or “definite” ALS. While these categorizations were created to better define eligibility for clinical research trials, the diagnostic labels are confusing and can easily be misinterpreted by patients.

Delays in Diagnosis

The average time between symptom onset and an ALS diagnosis is about one year. Diagnostic delay is common in ALS and is caused by multiple factors, including patients delaying medical attention until symptoms become more noticeable or affect function, as well as the fact that ALS symptoms can mimic many other conditions.

Patients may spend months or even years being evaluated and misdiagnosed by providers of different specialties before a diagnosis of ALS may even be considered. They may undergo unnecessary and painful diagnostic tests, or even invasive procedures (e.g., spinal surgery). Besides being stressful on patients and family members, the delay in diagnosis also delays initiation of treatments for ALS (which tend to be more effective the earlier they’re started). A delay in diagnosis can also limit eligibility for clinical trials, as many research studies have strict diagnosis date cutoffs in order to participate.

Treatment

Medications

As of April 2023, there are four approved disease-modifying medications for ALS:

Brand Name	Generic Name	Route of Administration	Mechanism	Most common side effects
Rilutek	riluzole	Oral or G-tube (pill, liquid)	Slows progression. Reduces signals that overstress and damage nerve cells.	Nausea, fatigue, elevated liver enzymes
Radicava	edaravone	Oral or G-tube (tablet, suspension, oral film), IV infusion	Slows progression. Free radical scavenger that is thought to reduce oxidative stress.	Headache, fatigue, walking problems, bruising at injection site (IV only), sulfite allergic reaction (if susceptible)
Relyvrio	sodium phenylbutyrate + TUDCA	Oral or G-tube (powder)	Slows progression. Reduces cellular stress pathways.	Gastrointestinal upset, upper respiratory tract infection, nausea, abdominal pain, diarrhea; severe: increased bile acid levels, salt retention
Qalsody (*Only for SOD1 familial ALS)	tofersen	Intrathecal Injection (i.e., administered via lumbar puncture)	Blocks production of toxic SOD1 protein. Found to reduce ALS biomarker Neurofilament Light Chain (NFL).	Pain including back pain and pain in arms or legs. Fatigue. Muscle and joint pain. Increased white blood cell count.

Medications for symptom management:

Symptom	Brand Name	Generic Name	Route of Administration	Most common side effects
Pseudobulbar Affect	Neudexta	Quinidine + Dextromethorphan	Oral	Diarrhea, dizziness, cough, vomiting
Muscle cramps	Mexitil, Namuscla	Mexiletine	Oral	Dizziness, ataxia, abnormal gait, numbness, paresthesia, confusion, headache, and tremors
Muscle cramps		Baclofen	Oral	Fatigue, diarrhea, nausea, vomiting
Sialorrhea (excess saliva)		Atropine	Sublingual drops	Blurred vision, chest pain, dizziness
Sialorrhea (excess saliva)	Transderm Scop®	Scopolamine	Transdermal patch, oral	Lightheadedness, itching (transdermal patch)
Sialorrhea (excess saliva)	Levsin® , Hyosyn	Hyoscyamine	Oral, sublingual drops	Dizziness, drowsiness, blurred vision

Multidisciplinary Care

Since there is no cure for ALS, and disease-modifying therapies for ALS are limited, the palliation of symptoms is a key component of treatment. Patients receiving Multidisciplinary ALS care live longer and have a higher quality of life. Multidisciplinary ALS clinics combine periodic visits with a Neurologist with the services of Physical and Occupational Therapists, Speech Language Pathologists (to assist with difficulties with speaking, chewing and swallowing), and/or Respiratory Therapists when breathing is affected.

Interventions and Equipment

Mobility

Progressive muscle weakness in the feet, legs and trunk muscles impair mobility in ALS. Physical and occupational therapists are critical to assessing and prescribing interventions to sustain a person's mobility and independence for as long as possible.

Walking and transferring:

- Ankle-Foot Orthosis (AFO) is an external brace commonly worn to stabilize and support weakness of muscles in the foot and ankle.
- Power chairs can help preserve mobility once leg muscle weakness makes walking unsafe or impossible.
- A Hoyer lift is a tool that assists caregivers with safely transferring patients with limited mobility due to muscle weakness.

Driving:

- Occupational therapists can recommend vehicle modifications to allow patients with ALS to continue to drive safely
- Modifications include enhanced steering wheels, hand controls, and wheelchair access vehicles

Parent guidance materials related to motor skills and mobility can be found here: https://www.massgeneral.org/als-pact.

Speech

While hearing and sight are not affected by ALS, communication challenges are common in ALS due to the gradual loss of speech. Muscles used to produce speech become weak or rigid, causing difficulty speaking (dysarthria).

Dysarthria typically begins with slurred speech, reduced volume of speech, increased nasality, until disease progression fully renders patients incapable of producing any comprehensible speech (anarthria).

Speech Language Pathologists are an important part of Multidisciplinary ALS Care and can provide assistance with augmentative and assistive communication technologies such as:

- Voice banking: Early in the disease course while a patient's speech is still preserved, patients work with SLPs to methodically digitize different samples (sounds, words, even personal catch phrases) of their own voice. When patients become anarthric, their characteristic vocal identity remains preserved if they use a computerized text-to-speech program.
- Eye gaze devices: When muscle weakness progresses to the point where writing or typing become impossible, devices exist that can sense a patient's eye gaze, which can be used to "type" by selecting letters, words or phrases.

Corresponding parent guidance materials related to changes in speech can be found at https://www.massgeneral.org/als-pact.

Swallowing and eating

Gastrostomy Tube (G-Tube)

- ALS affects the muscles required for chewing and swallowing, eventually leading to an inability to swallow (dysphagia). A G-tube is a surgically placed port that allows direct access to the stomach for food, water and medication.
- Many patients view G-Tube as a 'last resort' due to the surgery required for insertion and the loss of autonomy when eating or drinking. However recent studies suggest G-Tubes should be inserted earlier in the course of ALS, i.e., before respiratory function declines past a threshold that would cause the surgery to be unsafe. This can cause patients anxiety as they lose control of bodily functions earlier than they typically anticipated.
- Living with a G-tube can be uncomfortable and sometimes painful. Besides completely changing how someone receives their nutrition, it may require patients to change sleeping position, adhere to strict maintenance and cleaning time, and can cause other complications.

As nutrition and weight maintenance is also an especially critical component for ALS patients with a G-Tube, they must be mindful of nutritional supplements, blended meals, and tracking their weight. However, lack of oral stimulation may cause difficulty perceiving hunger. Overall, this is a major change for patients and may bring up feelings of anxiety and sadness. Patients may feel concerned about these changes being burdensome to caregivers, and may grieve the loss of independence. Conversely, a G-tube can be a relief from the fear and stress of potentially choking when taking foods or liquids by mouth.

Corresponding parent guidance materials related to swallowing and eating can be found at https://www.massgeneral.org/als-pact.

Breathing Difficulties (Respiration)

ALS can affect all voluntary muscles of the body, including those that facilitate breathing. When those muscles are affected, breathing becomes labored which is exhausting and frightening for a person. As this weakness progresses, respiratory failure is a common cause of death in ALS. Breathing ability by way of diaphragm muscle strength is typically tested at every multidisciplinary ALS clinic visit. Patients exhale through a spirometer, yielding a measurement called Slow Vital Capacity (SVC), which is considered an important marker of disease progression.

Respiratory therapists can offer the following interventions to improve quality of life and survival in ALS patients:

- **Cough Assist devices**: As respiratory muscle weakness progresses, it becomes more difficult or impossible to cough, an important action to clear secretions from the lungs. A Cough Assist device provides positive and negative pressure to simulate a cough, allowing lung secretions to be cleared, reducing the likelihood of lung infections.

- **Non-Invasive Ventilation** (NIV) refers to mechanical ventilation that does not require intubation. A BIPAP (Bilateral Positive Airway Pressure) machine mechanically provides positive and negative pressure to facilitate inhalation and exhalation. Patients typically wear a facemask connected via a hose to the BIPAP machine. BIPAP may be worn only during sleep or certain strenuous activities.

as respiratory weakness progresses, patients may require the continuous use of BIPAP 24 hours a day.

Tracheostomy and Ventilation

When respiratory muscle weakness has advanced to the point that breathing is no longer possible with just NIV, patients may elect to have a surgical procedure called a tracheotomy. A surgeon creates a small opening (tracheostomy) in the trachea (windpipe), in which a tube connected to a mechanical ventilator can be placed (intubation). The mechanical ventilator does all the work of breathing, continuously moving air in and out of the lungs.

People with a tracheostomy on a ventilator must be cared for 24 hours a day by family members or hired staff. Rehabilitation facilities provide comprehensive training and education to family members and other caregivers on care and equipment management. A person who has a tracheostomy and is on a ventilator must have one trained person always present. Home health aides' wages may not be covered by insurance and may have to be paid out of pocket.

Given the sheer invasiveness and financial burden, the decision to have a tracheotomy/ventilator is very personal and is often an ongoing discussion between patients and their care teams, beginning early on in the course of illness as part of defining the patient's goals of care and end-of-life wishes.

Corresponding parent guidance materials related to respiration/breathing difficulties can be found at https://www.massgeneral.org/als-pact.

Participation in Research and Clinical Trials

Over the past ten years, there has been an immense increase in the availability of clinical research trials for ALS. Participating in clinical trials, besides accelerating a path to new treatments and possibly a cure, often benefits patients with ALS on an individual level in many ways as well. They may receive experimental treatments that could slow or stabilize their disease progression, they are likely monitored by clinicians from their research team on a more frequent basis than their clinical providers, and they may feel a sense of hope, purpose and empowerment that they are able to help out, while having a fatal disease that feels so helpless. Observational studies exist for family members of genetic ALS patients, as well, and can benefit them by offering free, confidential genetic testing (often with the support of a genetic counselor), as well as frequent symptom monitoring and rapid access to care should they develop ALS as well.

Palliative Care, Hospice & End of Life

Palliative Care medicine is a specialty focused on caring for people living with a serious illness, such as ALS, cancer or heart failure. Palliative care is focused on treating and managing symptoms that accompany serious illness, such as pain, shortness of breath, fatigue and emotional distress, to improve a person's overall quality of life. Patients may also receive treatment intended to cure or slow their serious illness at the same time, so palliative care can be helpful to people who may not have long to live as well as to people who expect to get better, people who expect to live for a long time with an illness, and people who have been just recently diagnosed with an illness.

Hospice care and palliative care medicine are sometimes confused. They overlap in some ways but there are important differences between them. Hospice care focuses on the care, comfort, and quality of life of a person with a serious illness who is approaching the end of life.

At some point, it may not be possible to cure or slow progression of a serious illness, or a patient may choose not to undergo additional treatment that has these aims. Hospice is designed for this situation. For ALS patients, hospice care is typically offered when a person has experienced significant and rapid functional decline like being bed-bound, requiring total assistance, having extremely impaired breathing (when they do not want to be placed on a ventilator), and severe nutrition issues (when they do not want a G-tube). Some patients may view hospice care as “giving up,” but it is important to know it is meant to improve comfort as one nears end-of-life.

It is often helpful for people to consider hospice care sooner rather than later to avoid additional stress during emergency situations- that way, the family is already familiar with the hospice care team and process for accessing help.

Patients and families may struggle to engage in end-of-life planning, such as deciding when and how to use hospice and palliative care services, and planning end-of-life rituals and traditions. But ideally, ALS patients should have frequent and ongoing conversations with their family and care team to ensure that the end-of-life care they choose is consistent with their goals and expectations, and will meet both their emotional and physical needs.

Corresponding parent guidance materials related to the end-of-life period can be found at https://www.massgeneral.org/als-pact.

Resource List:

ALS Association – ALSA is an organization that serves, advocates for, and empowers people living with ALS to live their lives to the fullest, and works to discover treatments and a cure for ALS. https://www.als.org

ALS ONE Research Partnership: https://alsone.org

Compassionate Care ALS – CCALS provides resources including equipment, educational opportunities, Medicare/Medicaid assistance, communications assistance, guidance and awareness with regards to living with ALS, caregiving, and exploring end-of-life when invited https://ccals.org

ALS Finding a Cure Foundation: https://www.alsfindingacure.org

ALS Information at the National Institute of Health (NIH): https://medlineplus.gov/amyotrophiclateralsclerosis.html

Everything ALS – A non-profit organization that offers people with ALS a platform for direct engagement with other patients, caregivers, researchers, and drug companies. Everything ALS hosts bi-monthly community meetings designed to provide support and information to patients & caregivers. https://www.everythingals.org

I Am ALS – An organization dedicated to reshaping the public understanding of ALS and connecting patients, family, and friends to community resources whose mission is to educate and motivate all about ALS. I Am ALS is a platform for patients living with ALS to take lead in the fight against ALS and search for a cure. https://iamals.org/

Your ALS Guide – An online guide to help those recently diagnosed with ALS walk through the most important things to know about your diagnosis to planning ahead. https://www.youralsguide.com/newly-diagnosed.html