

## An Evolving Understanding of ALS with Frontotemporal Degeneration

**A**myotrophic lateral sclerosis (ALS), also called “Lou Gehrig’s disease,” is caused by the death of motor neurons, nerve cells that control voluntary muscles. Doctors and researchers are increasingly recognizing that many people with ALS also experience cognitive changes consistent with FTD. Indeed, as many as half of those with ALS exhibit behavioral changes or a decline in language skills similar to those observed in behavioral variant FTD or primary progressive aphasia. Conversely, up to 30% of people diagnosed with FTD develop motor symptoms consistent with ALS.

Over the last 10 years, there has been increasing recognition of a continuum between ALS and FTD that can be characterized on clinical, imaging, and pathological grounds. The recent discovery that mutation of the *C9orf72* gene is the most common genetic cause of both disorders offers further evidence of this continuum. ALS with FTD is an especially complicated and challenging form of FTD, and our understanding of it is still evolving.



## The Case of Cathy R.

### Early Symptoms and Diagnosis

While in her early 60s, Cathy R. began slurring her speech, a condition called dysarthria. (See glossary on page 3.) After six months of these symptoms, she and her husband Michael visited the neuromuscular disorders clinic at an academic medical center for evaluation. Michael explained that she did not like to talk because she was embarrassed by her speech, so he presented most of her history. He said her primary care doctor was concerned about the possibility of a stroke, and ordered an MRI scan of her brain. While the scan came back normal, her symptoms grew more consistent, and her doctor referred her to a neurologist.

The neurologist suspected myasthenia gravis, an autoimmune disorder that causes muscle weakness, and ordered blood work to confirm a diagnosis. As with her MRI scan, Cathy's blood work came back normal. Yet her neurologist continued to suspect myasthenia and prescribed Mestinon, a medication that alleviates its symptoms. She and Michael both thought that the Mestinon improved her speech.

During a follow-up appointment with the neurologist, Michael reported that she had tripped and fallen twice and wondered if this was a common problem in myasthenia. Additionally, she had developed muscle cramps in her legs. The neurologist believed that the Mestinon most likely caused the cramps. He prescribed Robinul to counteract them and discussed adding prednisone, a steroid, to treat Cathy's apparent leg weakness.

At the next monthly follow-up, Cathy reported that she had lost four pounds and had fallen again. Her neurologist noted a new hyperactive knee reflex and consequently questioned his diagnosis of myasthenia gravis. He referred her to a specialist in neuromuscular disorders for a second opinion.

The specialist took note of Cathy's most prominent symptoms: dysarthria, mild dysphagia (difficulty swallowing), muscle cramps, and multiple falls. She had lost a total of seven pounds over three months, which she ascribed to a poor appetite. Asked whether she had been more frequently laughing, crying or yawning, she smiled and shook her head "no," but her husband nodded: Yes, he had noticed that recently. A physical examination showed marked paucity of speech; she largely communicated by nodding and giving one-word answers. She giggled often and looked to her husband to complete her sentences. They examined her family history: Both of her parents died in a car accident when she was 38, while her maternal aunt had been diagnosed with dementia in her mid-50s—"but not the kind that makes you lose your keys or get lost," as Cathy put it. (Her aunt lived eight more years after her diagnosis before dying of pneumonia.)

The specialist evaluated her upper and lower motor neuron responses for indicators of damage to the nerve paths connecting the brain and spinal cord. She observed dysarthria, mild

facial weakness, and evidence of tongue weakness, as well as fasciculations (twitching) in the tongue, right upper arm, and both upper and lower legs. Cathy's gait showed right foot drop due to right ankle weakness. Reflexes were normal in the arms and "brisk" in the legs, meaning they contracted several times when tested; Babinski signs, reflexes that occur when the sole of the foot is stimulated, were not present. The physical exam indicated upper motor neuron involvement in the bulbar region of the brain and the lumbosacral (lower spine) area, and lower motor neuron involvement in the bulbar, cervical and lumbosacral segments with additional features suggestive of primary progressive aphasia.

Privately, the doctor determined that results of the examination were consistent only with a diagnosis of ALS with FTD; no other diagnostic considerations would account for all of her symptoms and signs. However, other conditions could potentially account for some of Cathy's symptoms, so additional tests will be needed to rule out those diagnoses.

### A SHARED GENETIC MUTATION

The discovery in 2011 that the *C9orf72* gene mutation can cause both frontotemporal degeneration (FTD) and amyotrophic lateral sclerosis (ALS) has transformed a long-held belief that ALS is purely a neuromuscular disorder and that FTD is purely a cognitive or behavioral form of dementia.

It is now recognized that the *C9orf72* gene is the most common gene causing hereditary FTD, ALS, and ALS with FTD. We also now know that several other genes can cause both diseases. FTD is a progressive brain disease that causes changes in behavior, personality and language dysfunction due to loss of nerve cells in the frontal and temporal lobes. ALS is a neurodegenerative disease in which loss of upper motor neurons (located in the brain) and lower motor neurons (located in the brainstem and spinal cord) can lead to paralysis, dysphagia, dysarthria and respiratory failure.

Describing the clinical syndrome where both FTD and ALS occur in the same person has been an area of active research, and our knowledge of the underlying genetics, pathology and clinical features is still unfolding.

ALS is mostly commonly associated with behavioral variant FTD. However, as our case study illustrates, primary progressive aphasia (including both the non-fluent agrammatic and semantic variants) have been reported in association with ALS. ■

The doctor explained that she did not have myasthenia gravis, and that further testing was needed. Michael asked what she thought was wrong with his wife. The doctor explained that the problem appeared to be with her motor nerves or neurons, and that she would have to do a few tests to be sure. Michael then said he had been reading about ALS—could that be the problem? The doctor said she was quite concerned that ALS was, in fact, the diagnosis, and she expected that additional tests would support this suspicion. There is no one specific test to confirm ALS, the doctor explained; additional tests are mostly to exclude other potential causes of the symptoms. Cathy paid intermittent attention during this discussion.

### An Expanded Diagnosis

Over the next two weeks, electrodiagnostic studies showed the expected nerve damage, while MRIs and blood work failed to point toward an alternative diagnosis. The couple returned for a follow-up visit to the multi-disciplinary ALS clinic. They first met with the specialist to discuss a formal diagnosis. She said that Cathy's reduced language production suggested primary progressive aphasia, thus expanding the diagnosis from ALS to ALS with FTD.

Cathy shook her head, and Michael said that her lack of speech production was due to her discomfort with her slurring, not FTD. "She understands everything, you can be sure of that!" he said. When the doctor asked her to write a sentence about the weather, she wrote, "tody sunny cold." However, because visits to the multidisciplinary clinic can be lengthy, the doctor did not pursue the issue of language production at that time.

During their first official clinic visit, Cathy and Michael met with a nurse practitioner, physical therapist, occupational therapist, speech and language pathologist, dietitian, research nurse coordinator, genetic counselor, and a representative from a nonprofit ALS care organization; a follow-up phone call with a social worker was also arranged. At the end of the clinic day, the team met to discuss their observations. All team members said they noticed that she spoke very little and laughed inappropriately. Pulmonology function tests showed her breathing was normal, but a swallow evaluation revealed that her swallowing function was significantly worse than she and her husband had reported. The speech and language pathologist recommended thickened liquids, while the physical therapist recommended

## ALS WITH FTD GLOSSARY

**Babinski sign:** Neurologic examination based upon what the big toe does when the sole of the foot is stimulated.

**Bulbar region:** Area of the brain, including the brain stem and cerebellum, that controls muscles in the face, neck and head.

***C9orf72*:** A mutation of this gene on chromosome 9, known as a hexanucleotide repeat, is the most common genetic cause of both FTD and ALS. The *C9orf72* mutation is associated with an abnormal accumulation of the protein TDP-43.

**Dysarthria:** Slurred or slowed speech that is difficult to understand, caused by difficulty moving and coordinating the muscles that control the lips, tongue and jaw.

**Dysphagia:** Difficulty swallowing, which can lead to gagging or choking.

**Electromyography (EMG):** Neurological test using fine needles to record the nerve impulses within certain muscles. Can detect when muscles start to lose their nerve supply, a symptom of ALS.

**Fasciculations:** Brief, spontaneous, uncontrolled muscle twitches.

**Hyperactive reflexes:** Sudden, involuntary flexing (bending) or extending (straightening) of a limb.

**Nerve conduction studies (NCS):** Neurological test sending electrical impulses through a small pad on the skin. This measures the speed at which nerves carry electrical signals.

**Percutaneous endoscopic gastrostomy (PEG tube):** Feeding tube used to provide nutrition when people are unable to swallow safely, or need nutritional supplementation.

**Pseudobulbar affect:** Episodes of uncontrolled, exaggerated emotional responses such as laughing or crying.

**Sialorrhea:** Excessive drooling or salivation.

**Upper and lower motor neurons:** Two sets of neurons that start in the brain and form pathways for transmitting impulses to control voluntary muscles and movement.

a brace for her right leg. The genetic counselor had asked the couple if they were interested in pursuing genetic testing, but—overwhelmed by the length of the visit and failing to fully appreciate that the condition might be genetic, and that that information could be of use to other extended family members—they declined. Together, they scheduled a follow-up visit in three months.

Eight weeks later, concerned that his wife was losing weight, Michael called the nurse practitioner to ask if he could move up their appointment. An evaluation showed that Cathy had lost eight pounds since her last visit.

The evaluation indicated progression in other symptoms. Cathy spoke rarely, and when she did her words were nearly unintelligible; she continuously looked to her husband to speak for her. She barely reacted when Michael wiped her face off after she exhibited sialorrhea (drooling). When Michael reported that she would not drink the thickened liquids, she just smiled and laughed. He said that she seemed uninterested in eating, even when he tried to give her foods she liked. In fact, she did not really seem interested in doing much at all. She had stopped socializing with friends, which Michael attributed to her discomfort with the sound of her speech. Oddly enough, while she liked to watch the Food Network all day, this did not seem to spark an interest in actually eating.

New findings from her physical examination included right hand weakness and worsened bilateral leg weakness, making her gait quite unsteady. At the previous visit, Cathy had rejected the idea of a brace for her right foot. This time, the doctor suggested that braces on both feet and a walker would help stabilize her movement. Michael was quite enthusiastic, but his wife smiled and shook her head no. To demonstrate how well she could still move, she stood up to walk, but as she turned she lost her balance and nearly fell over, causing her to laugh uncontrollably. The doctor suggested medication to enhance her appetite and control her drooling. Michael agreed to try, but said that she generally refused to take pills at this point.

When the doctor again brought up language disorders, Michael reiterated that his wife could still speak but chose not to because of the way her voice sounded. The doctor explained the relationship between ALS and FTD and discussed the different forms that FTD can take, focusing on primary progressive aphasia. (While ALS with FTD is most commonly associated with the behavioral variant of FTD, both the non-fluent agrammatic and semantic variants of PPA can occur in association with ALS.) She was very clear with Cathy and Michael that primary progressive aphasia was playing an important role in her disease process, and that certain symptoms of her FTD—including poor judgment

and lack of awareness—will make his caregiving responsibilities that much harder. Certain therapies and treatments for ALS will likely be unworkable because of her FTD. The doctor encouraged Michael to try not to get discouraged, although she emphasized that frustration would be common.

After Michael expressed a need for time to process this information, the doctor offered him an appointment with the affiliated cognitive clinic to learn more about FTD and connect with the many resources and supports available from AFTD. Still feeling overwhelmed, Michael said he would call back to schedule that appointment. In the meantime, the doctor encouraged him to focus on Cathy's safety and nutrition and to try to encourage her to take the new medication for appetite stimulation.



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### Shifting to Comfort Care

At their follow-up visit four weeks later, Cathy arrived in a transport wheelchair given to them by a member of their church. She looked thinner, having lost an additional six pounds. While she smiled most of the time, she produced no speech, instead making continuous moaning noises. She communicated through facial expressions and by nodding her head. Michael reported that she ate very little food, often holding it in her mouth for a long time before swallowing, and that she would not take any medications at all. She was able to walk minimally with assistance from her bed to the bathroom, and needed help with all activities of daily living. She denied being in pain, and Michael believed her.

Michael did say that he understood that his wife had ALS with FTD after he had done some reading about it. When the neurologist asked if she was interested in doing genetic testing for the benefit of her immediate family, Cathy did not respond. Michael said he did not want to put her through any unnecessary tests—he understood his wife's poor prognosis and wanted to ensure her safety and comfort during her decline. They began a discussion of hospice care and the types of assistance he would need to continue to care for her at home. Just three weeks later, Cathy died at home, with Michael at her side.

## Questions for discussion:

### What physical symptoms prompted the evaluation with a neurologist who specialized in neuromuscular disorders? What signs of ALS were found?

Initially, Cathy's most prominent symptom was dysarthria, or slurred speech. An MRI ruled out stroke early, but her dysarthria continued. Soon she started falling occasionally, and experienced muscle cramps and leg weakness. A neurologist suspected myasthenia gravis, an autoimmune disorder. During a follow-up appointment, the neurologist noted weight loss and abnormal reflex responses, and referred her to a specialist.

### What language and behavioral symptoms did Cathy exhibit that led the specialist to suspect ALS with FTD?

Early in her evaluation, the specialist noted her consistent lack of speech and inappropriate laughter. Cathy reported some family history of dementia. Michael believed his wife understood everything even though she did not respond, which can be consistent with primary progressive aphasia (PPA). The poor syntax in her simple written sentence was another indicator. She also demonstrated poor self-awareness and judgment when she impulsively tried to prove she could walk and almost fell down. The team at the ALS clinic all noted that lack of speech and inappropriate laughter are not characteristic of ALS alone.

### What contributed to Michael's difficulty recognizing the signs of primary progressive aphasia and their impact on Cathy?

Michael attributed his wife's reduced speech production to her disliking the sound of her voice, due to dysarthria. They both thought the medication, Mestinon, improved her speech. Her communication during appointments was mostly limited to nodding or smiling, which Michael would "translate" into speech for her. They both minimized her family history of dementia because "it was not like the memory kind." Quickly, however, her rapid progression became overwhelming for her husband. Absorbing her diagnosis and meeting her changing physical needs required all his focus and energy. He read about ALS with FTD and came to terms with his wife's diagnosis, but chose not to pursue genetic testing.

### What was the focus of treatment following diagnosis? When and why did it change?

Cathy's physical condition deteriorated rapidly. The ALS multidisciplinary clinic evaluation identified needs of ALS symptoms and recommended interventions based on standards of care in ALS—braces and a walker for movement, thickened liquids for easier swallowing, medication to counteract her drooling and encourage her appetite. Michael reported that she refused to wear braces, take her medicine or consume thickened liquid—all signs of cognitive impairment separate from ALS. Her obstinacy added to her husband's stress—he was trying to follow recommendations and be a good caregiver, but FTD kept interfering. By that point, Michael became more accepting of his wife's diagnosis. The doctor did not recommend standard ALS interventions, including breathing procedures and a PEG tube for feeding, as Cathy's PPA and rapid progression made compliance impossible. Michael shifted his focus to comfort care and hospice. It was just 15 months between his wife's initial presentation of dysarthria to a discussion of hospice care and her passing.

### Partners in FTD Care Advisors

The Partners in FTD Care initiative is the result of collaboration among AFTD, content experts and family caregivers. Advisors include:

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AFTD extends special thanks to this issue's special guest contributors. Lauren Elman, MD, an associate professor of neurology at the University of Pennsylvania Medical Center. Dr. Elman devotes most of her time to caring for people with ALS and other neuromuscular diseases. A member of the Penn Comprehensive ALS Center, Dr. Elman works closely with the Penn FTD Center and has spoken at several of their FTD conferences.

Miki Paul, PhD, is a licensed psychologist who cared for her husband Charles Hill through FTD/ALS until his passing in February 2016. Miki's search for support was instrumental in AFTD starting our telephone support group for FTD/ALS caregivers, and she has recently taken over as facilitator.

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## A CAREGIVER'S PERSPECTIVE... ON SLOW-DEVELOPING ALS WITH FTD

**by Miki Paul, PhD, psychologist, former caregiver of husband who had ALS with FTD, and facilitator of the AFTD phone support group for caregivers of loved ones who have ALS with FTD**

I lost my husband years before I lost him.

Chuck and I were living our dream life, spending weekends at our cabin with our dogs. But over the course of four and a half years, my gentle, even-tempered husband began exhibiting mood swings, personality changes, and cognitive difficulties. He became frustrated easily and was quick to anger; his ability to empathize or feel compassion disappeared. A formerly confident man, he became insecure and fearful, accusing me of having an affair when I was a little late coming home from work. He had trouble concentrating; tasks that he used to be able to do without thinking (using a remote control, turning on the windshield wipers) now took significant effort. Holding a job became impossible; he was fired seven times in four years. Chuck neglected his hygiene, often going days without showering. He started speaking slowly, his voice sounding soft and thick. A friend asked if he was drunk.

A large, physically strong man, he progressively became weaker, unable to open his usual ice tea bottles, carry luggage or lift up pans from the stove. He could no longer take long walks due to leg weakness. He slept more, and lost 25 pounds without even trying.

I felt confused, anxious, and helpless. Who was this man? What happened to my sweet husband? I begged him to see a doctor. It took literally years for him to agree.

Eventually Chuck was diagnosed with ALS with FTD. I was heartbroken, because I knew it was a death sentence (he died just 16 months after diagnosis). Yet even though this was the worst possible news I could ever imagine, I was relieved that his changed behavior was not willful, but rather the fault of his disease. Keeping this in mind was critical as the disease progressed.

ALS with FTD is the cruelest of diseases, relentless and unpredictable. Watching Chuck wither away in mind and in body, trying to get him to drink thickened liquid (which he hated), and seeing him gasp for air was extremely upsetting and nerve-racking. I thought being a single parent was the hardest job in the world, but it turned out being a caregiver to someone who has ALS with FTD is even more challenging. I grieved for the loss of Chuck's quick wit, his intelligence, his reliability, his friendship. He could no longer be my rock, so I became his.

Early on, I promised him I would keep him at home. So I scaled back my workload to 12 hours a week and hired reliable home-care providers (after firing a few) to assist my husband while I worked.

## COMMON PRESENTATIONS OF ALS WITH FTD

ALS can present in myriad ways, with weakness in any segment of the body. Roughly 75% of patients present with limb weakness, 25% present with bulbar weakness (trouble speaking or swallowing), and a small number present with respiratory insufficiency. Similarly, FTD can manifest early on as a predominantly behavioral syndrome or as a disorder of language; these patterns of disease can look very different. Both ALS and FTD can have variable disease courses as well.

Unsurprisingly, ALS with FTD presents and develops in many different ways—no two cases are the same. Here are some examples of persons diagnosed:

- A 58-year-old man develops walking difficulty due to stiffness in his leg. His wife reports that subtle personality changes, which she first noticed five years ago, have now begun to cause problems at his job.
- A 61-year-old man, already diagnosed with severe bvFTD, is referred by a cognitive clinic for further neuromuscular evaluation because of new onset right-hand weakness.
- A 53-year-old woman with dysarthria and hand weakness, whose sister has ALS, shows a lack of insight into her disability along with inappropriate public affection to her husband in the doctor's office during an examination.
- A 69-year-old man presented with dysarthria and over the course of two years began to exhibit anarthria (no audible speech) and developed a need for a feeding tube and power wheelchair. He then developed a language disorder that manifested in his writing: He confused "yes" and "no," showed a lack of grammar ability and made frequent spelling errors.

ALS is unpredictable—it can first manifest in any part of the body, then spread in a variety of patterns. One common presentation is when weakness starts in one arm: It will then spread to the opposite arm, then to the leg on the side of the body where it initially started. But it is harder to predict when respiratory or bulbar muscles may become involved. In both ALS and FTD, the speed of progression can be variable and is hard to predict. The best predictor of rate of progression is the patient's individual history, as rate of progression tends to remain constant within an individual. ■

I became his advocate, scheduling and accompanying him to all medical appointments (by then he had 10 different providers, including an ALS neurologist and a separate FTD neurologist).

The daily grind of caregiving duties exhausted me—once, I determined that I had completed 31 care-related tasks for him in a single day. I helped him with toileting, grooming, dressing and eating. I had to learn how to work all of his equipment, from his BiPAP machine to his lift chair to his wheelchair van—a steep learning curve for my non-mechanical brain. I was also responsible for organizing and administering all his medication. On top of that, I had to make all the household and home-maintenance decisions by myself, as well as medical decisions on Chuck's behalf. It was incredibly stressful.

Caring for my husband was a privilege, a heartache and a burden—he relied on me for everything. I knew I needed support, and attended support groups for both FTD and ALS care partners, but I met no one else who cared for someone with both FTD and ALS. I felt so alone. I begged AFTD and the national ALS organization to start a national phone-based support group for caregivers with this dual diagnosis. In the months before my husband died, I was able to participate in the new group a few times, which made me feel so grateful. The group was a ray of sunshine for me; I felt so desperate to connect with others going through a similar experience.

My caregiving experience changed me, and taught me much about myself. I became more organized, more assertive (on behalf of both my husband and myself), and better at time management. I learned to give up perfectionism, to be more patient, to stay in the present moment, rather than project into the unknown future. I learned I am more resilient than I ever imagined. I learned my body could become stronger.

I learned that a caregiver must care for herself as she cares for another. I planned for two much needed respites for short visits to my daughters and my grandchildren, which helped me cope. I received much appreciated stipends for these respite breaks from AFTD to help cover the costs of hiring in-home caregivers. Having some fun away from the sadness at home was critical for maintaining my personal well-being. I realized that I could not do it all without saving some energy, care and love for myself, so I learned to make time for myself without feeling guilty.

The cure for despair is hope. Even though my husband had two terminal diagnoses, I hoped for a new treatment or cure—if not for him, then for future generations. Donating his brain to science was at least a step towards one of those possibilities.

Throughout his journey, I hoped for a peaceful death for my husband while maintaining hope for my own future, even one without my beloved. Fifteen months after my husband died, I relocated to a different part of the country, near family, to create the next chapter of my life.

## A GENETIC PERSPECTIVE ON ALS AND FRONTOTEMPORAL DEGENERATION

Approximately 40% of affected individuals with FTD have a family history that includes at least one other relative diagnosed with a neurodegenerative disease. Their FTD is said to be familial, or hereditary. Hereditary FTD is caused by harmful gene mutations that affect proteins essential to the normal functioning and survival of brain cells. In FTD, autopsy can usually identify abnormal accumulations of either the protein tau, TDP-43 or FUS. The most common finding in ALS with FTD is TDP-43, with a small portion of cases associated with FUS.

Researchers have identified three genes that account for the majority of mutation-associated hereditary FTD cases. The most common genetic mutation in hereditary FTD and ALS is a mutation of the *C9orf72* gene. Approximately 10% of all cases of ALS are familial, and of these, 25% are accounted for by mutations in *C9orf72*. Similarly, 10% to 25% of patients with FTD demonstrate an autosomal dominant family history. Of these, 12% demonstrate mutations in *C9orf72*. Presently it is unclear how the mutation in this gene leads to the pathology that causes ALS or FTD.

Because they share a common inheritable mutation, ALS with FTD can present itself in many different ways within the same family. Members of the same family may present symptoms of just ALS, or just FTD, or innumerable combinations of ALS and FTD symptoms.

For more information, visit the [Genetics of FTD](#) section of the AFTD website. ■

## What to Do About...ALS with FTD

Over the last ten years, there has been increasing recognition of a continuum between ALS and frontotemporal degeneration (FTD). Up to half of people with ALS also show symptoms of FTD, while up to 30% of people diagnosed with FTD develop motor symptoms consistent with ALS. People diagnosed with either ALS or frontotemporal degeneration (FTD) may feel overwhelmed by their situation and miss or overlook possible signs of the other syndrome. But a potential dual diagnosis is important to consider due to its impact on treatment, prognosis, and understanding the genetic risks involved.

### Evaluation and Diagnosis

- Know the **signs and symptoms** of ALS and FTD, and be observant for changes consistent with both.
- Prepare for doctor visits. Keep a log of troubling behavior, language difficulties and muscle or motor changes to take to the doctor. ALS and FTD are both diagnosed by ruling out other possible disorders.
- Seek a second opinion at an academic medical center with specialists in cognitive neurology and neuromuscular disorders, if possible.

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If you are a military service member who has ALS with FTD, the VA may cover the cost of your care.

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- Request testing to rule out ALS if troubling signs are noticed (slurred speech, muscle weakness, stiffness or muscle cramping, early swallowing difficulty, etc.). Note that some of these symptoms can also be part of FTD without ALS, so a specialist is the best option for obtaining a diagnosis.
- Advocate for diagnostic clarity, as diagnosis of ALS with FTD impacts prognosis, treatment decisions and the support needed.
- Apply for Social Security benefits. The Compassionate Allowances program expedites the review process for people with both FTD and ALS diagnoses.
- If you are a military service member who has ALS with FTD, contact the Department of Veterans Affairs (VA). The VA may cover the cost of care for veterans diagnosed with ALS.
- Become informed on the genetics of ALS with FTD and consider possible family inheritance. Consult with a genetic counselor.

### Care Planning

- Discuss with the individual their wishes regarding the use of a feeding tube, ventilator, or other standard treatments in ALS as soon as possible. FTD's cognitive impairment complicates these decisions as the disease progresses.
- Complete key documents (healthcare power of attorney, end-of-life wishes) to maximize the involvement of the person who has ALS with FTD.
- Seek support from an ALS Association or Muscular Dystrophy Association clinic for a team approach to care and adaptive equipment related to the muscle disorder.
- Inform all members of the care team that treatment decisions must include the caregiver/healthcare power of attorney due to the individual's cognitive impairment.
- Seek an early palliative care consultation for help with key decisions, recommendations for appropriate care in degenerative motor disease, and a discussion about hospice.
- Think through recommended ALS interventions in light of the FTD diagnosis. A person with dementia may not understand how to use adaptive equipment and/or lack judgment to use it safely. Work with your care team to find alternate solutions if needed.
- FTD can increase dependency on the primary caregiver, so introduce home health services, physical therapy, occupational therapy and potentially hospice early to assist with activities of daily living. The earlier you can bring in help, the more time the person diagnosed has to adjust to people in their home. The person diagnosed may not be as agitated or resistant to accept assistance as the routine is being established.



## Care Planning (cont.)

- Consult a speech-language pathologist (SLP) for a bedside evaluation of swallowing difficulties (is it due to muscle weakness or inattention?) and recommendations about nutrition, food consistency and assistance with meals. An SLP can also provide strategies to maximize communication in persons diagnosed with primary progressive aphasia. Conduct research into residential care early to identify nursing homes that, if needed, would consider someone who has ALS with FTD.
- Interview hospice providers long before their services are needed. Respite for the family care partner is one benefit of hospice, among others, but the details can vary by provider. For more information, consult the Fall 2016 edition of Partners in FTD Care, entitled “**Comfort Care and Hospice in Advanced FTD.**”
- Focus on, and advocate for, quality of life.
- Establish a backup caregiving plan in case the primary care partner has health needs and is unable.



Complete key documents (healthcare power of attorney, end-of-life wishes) to maximize the involvement of the person who has ALS with FTD.



## The Importance of Support

- Identify sources of emotional support. Managing the logistics of care and making decisions on behalf of a person diagnosed is stressful for family care partners.
- Consider joining AFTD's telephone support group for care partners of someone who has ALS with FTD (contact AFTD's HelpLine at 866-507-7222 for more information), or an ALS, FTD or general caregiving group.
- Tell everyone what is going on.
- Ask for and accept help in all forms. Contact the Area Agency on Aging, the ALS Association or the Muscular Dystrophy Association.
- Remind caregivers they cannot do everything. Support their thoughtful choices, which they are trying their best to make in the face of numerous treatment recommendations and often feelings of guilt.
- Use respite services for the health and well-being of the caregiver (free with hospice).
- Empower the family to understand how an FTD diagnosis may impact standard ALS interventions. Listen to their concerns. Help a care partner decide to accept, request or refuse interventions based on their understanding of the wishes of the person diagnosed.
- Recognize that accidents (falls, choking spells, etc.) will happen in this complicated care situation, and that they are no one's fault.
- Encourage the caregiver to take time to consider how they prefer to spend time with their loved one.