

# ALS REPORTER GUIDE

INFORMATION ABOUT AMYOTROPHIC LATERAL SCLEROSIS

**ALS Awareness Month • May 2009**



Muscular Dystrophy Association • ALS Division  
[www.als-mda.org](http://www.als-mda.org)

©Muscular Dystrophy Association, 2009

# TABLE OF CONTENTS

<b>WHAT IS ALS?</b> .....	<b>2</b>
HOW DOES ALS AFFECT PEOPLE? .....	2
WHAT CAUSES ALS? .....	3
<b>ALS WARNING SIGNS AND DIAGNOSIS</b> .....	<b>4</b>
TWO TYPES OF ONSET .....	4
DIAGNOSTIC EXAMINATION .....	5
WHAT NOW? .....	5
<b>LIVING WITH ALS</b> .....	<b>6</b>
MOBILITY .....	6
SPEAKING .....	7
EATING .....	7
BREATHING .....	8
<b>THE ALS CAREGIVER</b> .....	<b>10</b>
PHYSICAL CARE .....	10
EMOTIONAL DEMANDS .....	11
CARING FOR THE CAREGIVER .....	11
LOVING LIFE .....	12
<b>ALS RESEARCH TODAY</b> .....	<b>13</b>
ALS RESEARCH HISTORY .....	13
ALS RESEARCH TODAY .....	13
ALS TREATMENT DEVELOPMENT .....	14
<b>ALS DIVISION — MUSCULAR DYSTROPHY ASSOCIATION</b> .....	<b>15</b>
MEDICAL CARE .....	15
FAMILY SUPPORT .....	16
RESEARCH .....	16
ADVOCACY .....	16
INFORMATION .....	17
PUBLICATIONS & VIDEOS .....	17
<b>MEET THE NIETOS</b> .....	<b>19</b>



## WHAT IS ALS?

You don't have to be a baseball fan to be familiar with Lou Gehrig's disease.

Amyotrophic lateral sclerosis, or ALS, got its nickname from the legendary New York Yankees first baseman who died from the disease in 1941. This disease most often strikes adults in middle age (average age of onset is 58), though it can affect people ranging in age from their teens to their 80s.

Each year more than 5,000 Americans receive diagnoses of ALS, and at any given time there are about 30,000 people in the United States who have the disease.

ALS doesn't discriminate. It affects both men and women, and people of all races and backgrounds.

## HOW DOES ALS AFFECT PEOPLE?

ALS causes progressive disintegration of both upper and lower motor neurons — nerve cells in the brain and spinal cord that control the action of voluntary muscles. As these nerve cells gradually die, the muscles that normally receive signals from them become weak and nonfunctional.

ALS *doesn't* directly affect involuntary muscles, such as those that control the heartbeat, gastrointestinal tract and bowel function, bladder, and sexual functions. Hearing, vision, touch and intellectual ability generally remain quite normal, although mild cognitive impairment is common. Severe impairment occurs in some cases.

Eventually, most people with ALS become almost completely paralyzed and lose the ability to speak and breathe on their own.

Death, usually from respiratory complications, has typically come within three to five years of diagnosis. However, modern technology has challenged these statistics, helping people

with ALS to compensate for almost every loss of function to some degree. Today mechanical ventilation and feeding tubes allow people with ALS to live longer; power wheelchairs enable mobility; and computers provide means of communication.

Receiving a diagnosis of ALS doesn't mean life is over.

Technological advances as well as a dedicated spirit and a supportive family or circle of care all influence quality of life.

For example, ALS doesn't affect a person's creativity, as evidenced by the MDA Art Collection, which includes artwork from a number of artists with ALS. Artists have painted by holding brushes in their mouths, or created computer art using mouthsticks or sensors attached to their eyebrows or foreheads.

### **WHAT CAUSES ALS?**

About 10 percent of ALS cases are "familial," and are clearly linked to hereditary genetic causes, with more than one case in a family.

In the other 90 percent of cases (called "sporadic ALS"), there is no family history, but genetic factors are thought to play a role. MDA's search for these factors took substantial steps forward in 2008-2009.

In 2006, MDA and the ALS Therapy Foundation formed the ALS Therapy Institute, and launched the largest ALS drug discovery program in history, a three-year, \$36 million project to identify biochemical targets in ALS and seek drugs that work on them. In addition, MDA continues to fund dozens of ALS research teams around the world and participate in a number of clinical trials, in order to achieve a greater understanding of the disease and ways to stop its destructiveness.

Although the causes of ALS are still unknown, many families affected by ALS find hope in the fact that MDA is investing \$13.6 million a year in its worldwide research program seeking treatments or a cure for this disease.

In addition to its research program, MDA also combats ALS through comprehensive medical care, individual and family support services, and far-reaching professional and public health education, spending \$11 million on these vital services in 2009.



## **ALS WARNING SIGNS AND DIAGNOSIS**

With ALS, as with other diseases, early diagnosis is important, but the early warning signs can vary significantly.

It's not unusual for people to ignore the warning signs of ALS for some time (perhaps months), or to consult a physician who at first believes there's no serious problem. However, ALS is progressive and symptoms generally will spread from one part of the body to another, almost always in parts adjacent to each other. Eventually the problem can no longer be ignored.

### **TWO TYPES OF ONSET**

The "bulbar-onset" form of ALS commences with the gradual loss of motor neurons, or muscle-controlling nerve cells, in the brainstem.

The first signs include difficulty using the muscles controlling speech, swallowing and facial expressions — i.e., slurred speech, problems in swallowing or a crooked smile.

The "limb-onset" form of ALS begins with loss of motor neurons in the spinal cord.

Early signs of this form may include persistent muscle weakness in an arm or leg, and muscle atrophy (loss of bulk), cramping and twitching. People report first noticing a twitch in the thumb, an inability to lift the foot as high as before, or a sense that one arm or leg was growing weaker.

In both forms of onset, motor neurons in the top portion of the brain are destroyed. The result is gradual progression of muscle weakness, stiffness, slowness of movement, and muscle tightness, or spasticity.

The spread of symptoms is a signal to the general practitioner to refer the person to a neurologist, who then will consider ALS among many other possibilities. Doctors may refer someone they suspect of having ALS to the local MDA office, where an appointment can be made at an MDA clinic or an MDA/ALS center.

## **DIAGNOSTIC EXAMINATION**

There's no definitive test for ALS, so it's diagnosed after other conditions have been ruled out. A thorough medical and family history and physical examination are the starting points of a neurologic workup. The person then undergoes simple, in-office tests of muscle and nerve function.

If ALS is being considered, further tests will be undertaken. These may include an electromyogram, or EMG, which measures the electrical activity inside muscles to see if there's a pattern consistent with ALS; nerve conduction velocity studies, which measure how fast nerve signals move from the spinal cord to muscles; and imaging of the spinal cord and brain, such as with an MRI (magnetic resonance imaging) scan.

## **WHAT NOW?**

Early detection of ALS can lead to interventions that will improve and lengthen the lives of those affected by this disease. MDA supports some 220 clinics and more than 30 MDA/ALS centers throughout the United States. These provide top-quality medical care from leading experts.



## **LIVING WITH ALS**

ALS affects several major body functions, including mobility, speaking, eating (by limiting the ability to chew or swallow) and breathing.

As daunting as this may sound, advances in medical treatments and technology have made it possible for people with ALS to continue leading rewarding lives and remain involved in many activities. In fact, some treatments, such as feeding tubes, ventilators and a medication called riluzole, can prolong survival and improve quality of life.

The medical experts at MDA/ALS centers or MDA clinics are a great resource for information about living with the various aspects of the disease.

### **MOBILITY**

Today's technology provides mobility for almost everyone, no matter how few muscles remain functional.

MDA clinic staff can direct people with ALS to mobility specialists, who help obtain the equipment that's best at each stage of the disease, and who assist in navigating the insurance and funding maze.

In addition, MDA provides financial help with the purchase and repair of wheelchairs and leg braces, and also loans equipment through its equipment loan closets. (Local MDA Loan Closets accept donations of new and used durable medical equipment from the public.)

In the early stages, a cane and/or a supportive brace can help the person with ALS to get around. A leg brace (ankle-foot orthosis, or AFO) can prevent tripping by keeping feet from "dropping" with each step.

Later, additional devices become useful, such as walkers, manual wheelchairs and power wheelchairs. As muscles are progressively lost, different types of switches allow people to operate their power chairs by foot, head or even breath. In addition, power chairs for people with ALS include a tilt-and-recline function for maintaining circulation, avoiding pressure sores and assisting with body stability.

Physicians and therapists recommend that ALS patients look into getting equipment well before it's needed, because some custom equipment can take several months to obtain.

## **SPEAKING**

People with ALS often lose the ability to speak. One of the most distressing aspects of the disease, this may occur when throat and mouth muscles are weakened. In some forms of ALS (bulbar-onset), slurred speech or difficulty swallowing may be among the first symptoms.

A speech-language pathologist (SLP) can provide crucial help in dealing with this loss. An SLP who's up-to-date on the growing choices in life-enhancing technology can help select and find funding for an augmentative, assistive communication (AAC) device. Help with such funding is provided by MDA.

SLPs often advise those whose speech isn't yet affected to plan ahead by recording their own voices to be uploaded later onto an AAC device. They also urge patients to learn how to use this assistive technology before it's urgently needed.



## **EATING**

Adequate nutrition is another major concern for people with ALS, particularly as weakness progresses in the muscles involved in chewing, moving food toward the back of the mouth, and swallowing.

With weaker muscles, eating and drinking can become more hazardous and time-consuming. Choking and aspiration — inhaling food or liquid into the lungs — can be the result.

SLPs and other therapists who specialize in swallowing are important allies here. They recommend early solutions such as changing the consistency of food or swallowing techniques.

Later, physicians and therapists may suggest the insertion of a tube directly into the stomach (not down the nose). Getting a gastrostomy tube, also called a g-tube or PEG (percutaneous endoscopic gastrostomy) tube, can be a great relief to people with ALS. They can continue to eat and drink if desired, but no longer have to rely on swallowing as the sole method of obtaining adequate nutrition.

## BREATHING



ALS gradually weakens the muscles responsible for moving air into and out of the lungs. Most people with ALS end up using some form of assisted ventilation to mechanically improve air movement.

(ALS doesn't impair the ability of the lungs to extract oxygen from air, so supplemental oxygen usually isn't needed for an otherwise healthy person. In fact, supplemental oxygen given to someone whose only problem is weak respiratory muscles can actually cause more breathing difficulties.)

ALS gradually weakens the muscles responsible for moving air into and out of the lungs. Most people with ALS end up using some form of assisted ventilation to mechanically improve air movement.

(ALS doesn't impair the ability of the lungs to extract oxygen from air, so supplemental oxygen usually isn't needed for an otherwise healthy person. In fact, supplemental oxygen given to someone whose only problem is weak respiratory muscles can actually cause more breathing difficulties.)

Symptoms of breathing difficulties can include shortness of breath with exertion or while lying down, difficulty sleeping or dozing a lot during the day, "blunted" or "fuzzy" thinking, or frequent headaches or nausea upon awaking (thought to be due to a buildup of carbon dioxide).

A physician can perform pulmonary function tests to measure the levels of oxygen, carbon dioxide and other chemicals in the blood. He or she may involve the expertise of a pulmonologist and/or respiratory therapist, and discuss the options available to mechanically assist with breathing.

The main types of assisted ventilation are noninvasive and invasive.

Noninvasive ventilation means that air is delivered by a mask over the nose, mouth or both. In invasive ventilation, air is delivered through a surgical hole in the throat, called a tracheostomy (or trach).

Many devices to assist with breathing are on the market today, and each has its own advantages. Whichever system is chosen, the goal is the same: to ensure the lungs can inhale and exhale an adequate supply of air.

*For more detailed information about these topics, go to [www.als-mda.org](http://www.als-mda.org).*



## THE ALS CAREGIVER

ALS doesn't just attack the person with the disease. It has a profound impact on everyone who cares about that person, especially the primary caregiver, who is often the spouse.

Caring for someone with ALS is a physically and emotionally wrenching journey, but many caregivers have found it also a psychologically and spiritually enriching one.

### PHYSICAL CARE

As ALS progresses, caregivers must adapt to their loved ones' steady loss of function.

In the early stages of the disease, a caregiver may simply do a larger share of physical chores at home. In the later stages, an affected person is totally reliant on the care of others.

Caregivers become responsible for all elements of the person's life: hygiene and skin care; bowel and bladder routines; transfers between bed and wheelchair; providing easy-to-swallow nutrition; range-of-motion exercise to prevent contractures (painfully tight muscles and immobilized joints).

Physical care comes to comprise thousands of little care moments. As the disease progresses, caregivers arise many times a night to turn their loved ones to a more comfortable position. They scratch itches and adjust eyeglasses. They cut food into tiny bites and pick up dropped objects. And they try to do it all with grace and good humor.

Constant respiratory care and monitoring also are important elements of late-stage caregiving in ALS, especially if the patient is using a ventilator. Even a short lapse in caregiver diligence, day or night, can be fatal at this point.

Private or state-provided nursing services can perform some caregiving duties, depending on the family's resources. But these programs are limited, and some degree of responsibility always rests with family members or other volunteer caregivers.

The staff at a nearby MDA clinic and members of an MDA ALS support group help families learn how to perform caregiving duties, and advise on community resources to help with these demands.

Another great help is MDA's new book, *MDA ALS Caregiver's Guide*, and its 2005 book, *Everyday Life With ALS: A Practical Guide*. Both are available through MDA offices or online.

## **EMOTIONAL DEMANDS**

The psychological demands of caring for someone with ALS can be just as complex and relentless as the physical ones.

Patients and caregivers often are married, but ALS changes their relationship dramatically. Privacy is lost; family responsibilities and activities shift.

A caregiver's employment may be threatened by increased demands at home, and finances can become very tight.

Caregivers' feelings of sadness, fear and grief over the disease may become mingled with resentment, impatience, anger, self-pity and guilt, causing even more psychological stress.

In addition to coping with their own emotions, caregivers must help keep up a patient's spirits. Depression is common in ALS, as is frustration and rage over loss of ability. Although ALS doesn't commonly affect thinking, some people with the disease experience an odd neurological side effect that causes them to burst into uncontrollable laughing or crying for no obvious reason (pseudobulbar affect).

## **CARING FOR THE CAREGIVER**

According to a 1997 survey by the National Family Caregivers Association (NFCA), more than half of all caregivers have experienced sleeplessness and depression, and three-quarters say they

receive no consistent help from other family members. Back pain, headaches, stomach disorders and loss of personal time are common caregiver complaints.

In order to meet their enormous responsibilities, ALS caregivers need to take care of their own health. But most find it very hard to get enough sleep, eat nutritiously, exercise regularly and take time for themselves.

The key to caregiver health is developing a team to “share the care.” Care circles can be groups of people formally organized to help, or loose networks of family and friends who fill in the gaps and provide sporadic support.

## **LOVING LIFE**

As grueling as it is to take care of a person with ALS, caregivers report that laughter, love and fun still are big parts of their lives, and that ordinary moments have become sharper and sweeter in the face of a terminal disease.

Families create new definitions of normalcy, and modify or replace old dreams with new ones. Couples have honest discussions about difficult issues of life and death.

Many caregivers and their loved ones with ALS report that — although they hate ALS — it has caused them to grow in character, spiritual depth, and love and commitment.



## **ALS RESEARCH TODAY**

MDA's ALS Division leads the world in conducting research on ALS, expending \$13.6 million in 2009. Through its hospital-affiliated MDA/ALS centers and clinics, MDA's scientists have developed and tested a number of potential ALS treatments over the years.

### **ALS RESEARCH HISTORY**

Only a little more than a decade ago, no one had any idea what caused ALS, and therapeutic trials were just stabs in the dark. MDA-supported scientists uncovered a primary clue in 1993, with the finding of a flaw in a gene for the protein SOD1 in a small number of people with ALS. Almost immediately, mice with this kind of mutation were bred, giving the ALS research community its first mouse model of human ALS, a prerequisite for testing treatments.

In the 1990s, research showed that at least some people with ALS have defects in the clearance of the neurotransmitter glutamate from the area around nerve cells. MDA research was instrumental in testing riluzole (Rilutek), which partially blocks the release of glutamate, and is the only FDA-approved drug developed specifically for ALS. The drug extends life expectancy by a few months.

Since then, many more genes have been found that may directly cause ALS or influence its risk or severity. All these genes are providing further insights into the disease.

### **ALS RESEARCH TODAY**

MDA-supported researchers at the ALS Therapy Development Institute (ALS TDI) in Cambridge, Mass., are identifying new disease targets, discovering compounds that may act against these targets, and screening potential treatments for clinical development.

The ALS TDI recently partnered with Asklepios BioPharmaceutical, an MDA-supported company in Chapel Hill, N.C., to develop gene therapy to treat ALS.

MDA is supporting a clinical trial of lithium carbonate, which showed some promise in slowing the disease in a small trial in Italy. MDA clinics, ALS Centers and physicians are involved in dozens of other clinical trials, such as those studying *arimoclomol*, a drug that may improve cellular defenses against misfolded, toxic proteins; *ceftriaxone*, a drug that may help transport potentially toxic glutamate molecules away from nerve cells; *talampanel*, a drug designed to block cellular landing sites for glutamate; and *SB509*, a compound designed to activate a substance called VEGF that has neuroprotective properties.

The MDA-funded ALS Connection ([www.alasconnection.com](http://www.alasconnection.com)), a Web-based database, continues to gather long-term follow-up data on people with ALS that will be useful for research.



## **ALS DIVISION**

### **MUSCULAR DYSTROPHY ASSOCIATION**

Every May, the nation observes ALS Awareness Month to inform the public about amyotrophic lateral sclerosis (ALS, or Lou Gehrig's disease). The Muscular Dystrophy Association is the world leader among voluntary agencies fighting ALS.

MDA's ALS Division offers the most comprehensive range of services of any voluntary health agency in the nation, with clinics serving people with ALS in communities across the country. MDA leads the search for a treatment or cure through its aggressive, worldwide research program.

In 2009, MDA is spending \$24.6 million on its programs of research, services and information for those with ALS.

Since its inception, MDA has invested more than \$250 million in its ALS program, and is committed to continuing to lead the fight.

For more information, go to [www.als-mda.org](http://www.als-mda.org) or call (800) 572-1717.

#### **MDA's ALS Division provides:**

##### **MEDICAL CARE**

Anyone whose doctor suspects ALS may call a local MDA office and get an appointment at one of MDA's 220 hospital-affiliated clinics or its 36 major medical institutions across the country. There, a multidisciplinary team of the area's local ALS experts, including neurologists, other physicians, nurses, therapists, social workers and more, will provide a diagnostic examination.

People with ALS may register with MDA (it's free), and throughout the course of the disease will be seen for regular follow-up examinations by these experts.

## **FAMILY SUPPORT**

Through its local offices, MDA offers support groups for those with ALS and their caregivers. The MDA ALS Web site ([www.als-mda.org](http://www.als-mda.org)) also hosts regular online chats for individuals affected by the disease.

Devices to help with day-to-day living may be borrowed from MDA's equipment loan closets. MDA helps with the cost of wheelchairs, leg braces and communication devices, as well as wheelchair and brace repair.

The MDA ALS Division also offers educational seminars, home visits, community outreach and advocacy, and a "phone friends" support network.

## **RESEARCH**

MDA's ALS Division funds more research into the causes of, and treatments and a cure for, ALS than any other voluntary health agency in the country.

MDA funds top researchers around the world and conducts or indirectly supports clinical trials of drugs and other potential ALS treatments. Anyone interested in participating in a clinical trial can find details at [www.mda.org/research/ctrials.aspx](http://www.mda.org/research/ctrials.aspx) or by following news reports on MDA's ALS Web site.

## **ADVOCACY**

MDA's Advocacy program provides a unified voice for the hundreds of thousands of Americans affected by ALS and other muscle-wasting diseases. MDA advocates make a difference by helping shape governmental decisions in such areas as public policy and research advancement, as well as raising public awareness of the concerns and priorities of people with chronic, debilitating and fatal conditions.

MDA's Advocacy program offers practical ways to become involved through its "Take 5!" initiative, which provides the tools for people to take just five minutes and contact their elected officials about important legislation. For more information, visit MDA's Advocacy Web pages ([www.mda.org/advocacy](http://www.mda.org/advocacy)).

## **INFORMATION**

MDA is an excellent source of breaking news about ALS research developments, as well as information on coping with the medical, financial, emotional and other challenges presented by the disease. ALS is always prominently represented on the Jerry Lewis MDA Labor Day Telethon. People across the country are available for interviews during ALS Awareness Month and year-round.

The starting point for this information is MDA's ALS Web site, [www.als-mda.org](http://www.als-mda.org), or local MDA offices, which can be found by calling (800) 572-1717.

## **PUBLICATIONS & VIDEOS**

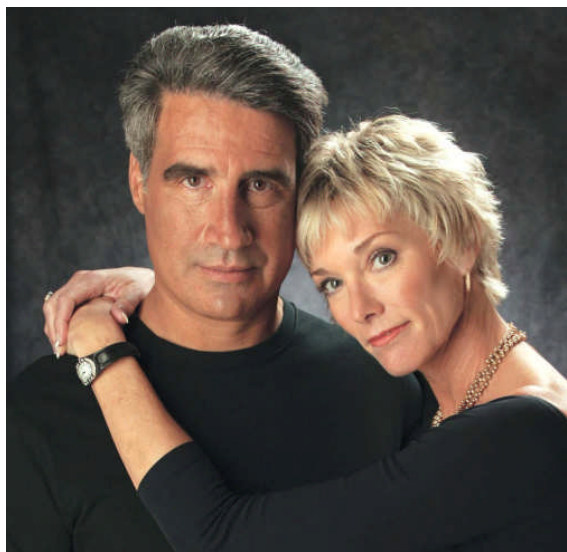
Several ALS-specific publications and videos are available through the Web site, local MDA offices or MDA National Headquarters in Tucson, Ariz., including:

- **ALS: Anyone's Life Story series** -- Online features on individuals with ALS during ALS Awareness Month
- ***Everyday Life With ALS: A Practical Guide*** — A 150-page book that assists readers with ALS in coping with home modification, mobility, breathing, speech, exercise and more
- ***MDA ALS Caregiver's Guide*** — A comprehensive illustrated manual filled with practical advice for caregivers and including extensive resources. New in 2008.

- “Breathe Easy: Respiratory Care in Neuromuscular Disorders” – A booklet with a special section on ALS, available in English and Spanish
- “Facts About Amyotrophic Lateral Sclerosis” — An introductory pamphlet for those with new diagnoses, available in English and Spanish
- MDA ALS Division brochure -- A concise description of MDA’s ALS program, including a summary of MDA services, Web site addresses and contact information.
- MDA/ALS Newsmagazine — Articles include news on ALS research, profiles of people with ALS, and practical information about health care and caregiving
- *Meals for Easy Swallowing* – A book of recipes for those with weakened mouth and throat muscles. Available only online.
- “Milestones in ALS Research” – A booklet tracing major research developments since the 19<sup>th</sup> century. MDA has led the worldwide ALS research effort since the 1950s.
- Quest — MDA’s quarterly national magazine for people coping with any of the 40 neuromuscular diseases under MDA’s umbrella, including ALS (with additional online stories, called Quest Extra!, at [www.mda.org/questmagazineonline](http://www.mda.org/questmagazineonline))
- “With Hope and Courage: Your Guide to Living with ALS” – A new 2008 video to orient and inform people with new ALS diagnoses and their families
- Emergency care envelope: An easy-to-find organizer for all the important documents that should accompany a person to the hospital in an emergency

# MDA<sup>®</sup>

## ALS DIVISION



### MEET THE NIETOS

Augustine (Augie) L. Nieto II and his wife, Lynne, are co-chairs of MDA's ALS Division for the fourth consecutive year. The two raise awareness of ALS and MDA's mission through media interviews, public appearances, speaking engagements and public service announcements.

In addition, they spearhead Augie's Quest, a MDA fund-raising program for its ALS research program.

"We intend to win this war against ALS by building the best scientific and technologic army in the world," said Nieto, 51. "Amazing things can happen when you combine MDA's drive to succeed with money and technology."

Nieto received an ALS diagnosis in March 2005. The disease has affected the muscles throughout his upper body, including those used for speech, swallowing and mobility.

Ironically, since 1977 Nieto has guided the fitness industry to new heights, most notably as the co-founder and former president of Life Fitness. He currently is chairman of Octane Fitness.

In September 2005, he received the Lifetime Achievement Award presented by the International Health, Racquet & Sportsclub Association and the National Fitness Trade Show. Nieto transformed the Las Vegas award dinner and reception into a benefit for MDA's ALS Division, raising more than \$1 million — the largest amount ever raised by an MDA inaugural event of its kind.

Since 2005, Augie's Quest has raised more than \$18 million for MDA's ALS research program.

The Nietos live in Corona del Mar, a Southern California seaside community. They have four grown children. Lynne Nieto has a degree in accounting and worked for more than 20 years in her family's real estate development business.

In addition to supporting research, the Nietos also want to increase public understanding of MDA's primacy in the battle against ALS.

"We want MDA to be viewed as the ultimate resource for people looking for answers about anything from care needs to insurance issues," said Nieto.

"We're going to help put a face on this disease, share crucial information about ALS, help empower others dealing with this disease, raise public awareness and raise money for research in a fast-track format."

- MDA -