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Facts About Amyotrophic Lateral Sclerosis (ALS or Lou Gehrig's Disease)

Dear Friends:


Glenn Harwood

When I learned in 1994 that I had ALS, my world changed. I was in my early 50s, had a good career with the U.S. Small Business Administration, and was a husband and father. Suddenly, I faced a serious disease that would affect every aspect of my life.

I decided my best weapon in this battle would be attitude. I've met every challenge ALS has presented with an approach that's unbeatable: I can do it, I will do it. With that conviction, I have a full and rewarding life with the help of a power wheelchair, a voice synthesizer and lots of e-mail. I've continued working, traveling and learning.

I have wonderful allies in this fight: a fantastic family, including my wife, Fran, our two children and beautiful grandchildren, who give me all the help I need and great joy and purpose. My faith in God never fails me, and helps me keep going with hope.

I've also honed my sense of humor. I carry a page of jokes with me at all times and regularly e-mail friends across the country with "My Sunday Bulletin," a compendium of jokes I collect from my e-mail friends. My motto is "Can't Walk or Talk But Can Always Laugh."

Another vital weapon in my arsenal is the Muscular Dystrophy Association, which offers the best doctors and health care professionals in the country. You also can count on MDA for support groups, help in finding and financing special equipment, and support and understanding at every turn.

This MDA booklet offers an introduction to ALS, so you can begin preparing to meet the coming changes. MDA also gives each person with ALS it learns of copies of its very helpful and thorough book, ***Everyday Life with ALS: A Practical Guide***.

From this booklet you'll learn several encouraging things about having ALS: that your diagnosis is in no way your "fault" … that ALS doesn't affect the intellect and that many physical functions remain unaffected … and that better treatments and technological devices are constantly being tested and developed for every aspect of the disease.

It's good to know that society is far more aware of people with disabilities today, and the laws entitle you to equal employment opportunities and access to public places.

By the way, many people with ALS survive much longer than expected — in my case, more than 10 years since the earliest symptoms. I know of others who've had the disease for 15, 20 or more years.

You'll find, as I did, that the love of your family and friends will give you strength. A hopeful attitude and good sense of humor will keep ALS in perspective, as only one part of your life.

And remember: MDA and all its resources are there to help you and your family. *You're not alone*.

Glenn Harwood
Crofton, Maryland

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What is Amyotrophic Lateral Sclerosis?

ALS is a disease of the parts of the nervous system that control voluntary muscle movement.

The word “amyotrophic” comes from Greek roots that mean “without nourishment to muscles” and refers to the loss of signals nerve cells normally send to muscle cells. “Lateral” means “to the side” and refers to the location of the damage in the spinal cord. “Sclerosis” means “hardened” and refers to the hardened nature of the spinal cord in advanced ALS.

In the United States, ALS also is called Lou Gehrig’s disease, named for the Yankees baseball player who died of it in 1941. In Britain and elsewhere in the world, ALS is often called motor neuron disease in reference to the cells that are lost in this disorder.

What happens to someone with ALS?

In ALS, nerve cells that control muscle cells are gradually lost. In most cases, the cause is unknown. As these motor neurons are lost, the muscles they control become weak and then nonfunctional. Eventually, the person with ALS is paralyzed.

Death, usually from respiratory complications, typically comes between three and five years after diagnosis (some studies say after symptoms are noted, so the timing is unclear). About 10 percent of those with the disease live more than 10 years, and some survive for decades.

Modern technology has allowed people with ALS to compensate for almost every loss of function to some degree, making it possible even for those with almost no muscle function to continue to breathe, communicate, move about and use a computer. Longevity statistics may be somewhat out of date because of changes in supportive care and technology. For example, British physicist Stephen Hawking has had ALS since the 1960s and is still able to write and practice his profession.

It’s important to note that the involuntary muscles, such as those that control the heartbeat, gastrointestinal tract and bowel function, bladder and sexual functions are not directly affected in ALS. (However, prolonged inability to move and other effects of ALS can have some indirect impact.)

Pain is not a direct consequence of the disorder, although moderate pain can certainly occur as a result of immobility and its various complications.

Hearing, vision, touch and intellectual ability generally remain quite normal. Some people experience loss of control over emotional expressions such as laughing or crying, a phenomenon thought to be directly related to the disease process.

ALS can strike people of any age, though it usually strikes in late middle age.

Of course, in such a devastating disorder as ALS, it’s natural for people to feel angry, sad or depressed, whether or not they experience unwanted emotional episodes.

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What happens to the nervous system in ALS?

The muscle-controlling nerve cells, or motor neurons, are divided into two types. The upper motor neurons are located in the upper part of the brain and exert some control over the lower motor neurons, which are in the brainstem and the spinal cord. (See illustration.)

The lower motor neurons are directly attached to muscles through “wires” called axons. Bundles of these axons leave the spinal cord and extend out to the muscles. It’s these bundles that doctors are referring to when they talk about the “nerves.”

The function of lower motor neurons is straightforward. They send “go” signals to muscles. When these cells gradually die, as in ALS, muscles atrophy (shrink) and become progressively weaker and eventually unable to contract, resulting in paralysis.

The lower motor neurons that control most of the body are in the spinal cord. Those that control the muscles of speaking, swallowing and facial expression are in the brainstem. They’re sometimes called bulbar motor neurons, because the part of the brainstem that houses them has a bulblike shape. The term bulbar involvement means that the muscles of the face, mouth and throat are involved in the disease.

The upper motor neurons have more complex functions. It’s harder to study them, and not as much is understood about them, although new techniques are changing that.

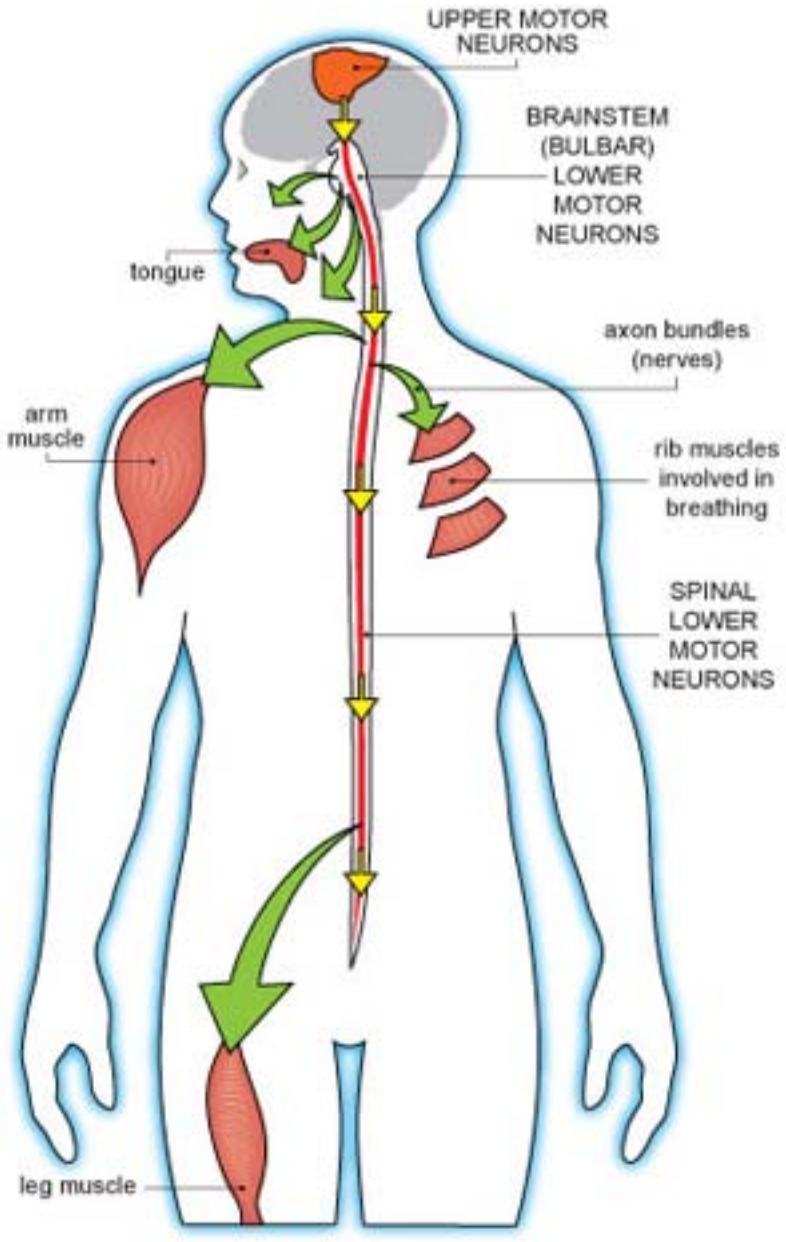
These cells seem to exert complex control over the lower motor neurons that allow movements to be smooth, directed and varied in intensity. (For instance, they’re part of an elaborate system that allows a person to aim a hand at a glass of water, pick it up, estimate its weight, use the right amount of force for its weight, and lift it to his or her mouth, all while thinking about something else.) When upper motor neurons are lost and lower motor neurons remain, movements are still possible but can become “tight” (doctors use the word spastic for this) and less precise.

In ALS, a combination of these effects is usually seen because both upper and lower motor neurons are dying. People with ALS can have weak and wasted muscles with tightness (spasticity). Muscle twitches and cramps are common; they occur because degenerating axons (nerves) become “irritable.”

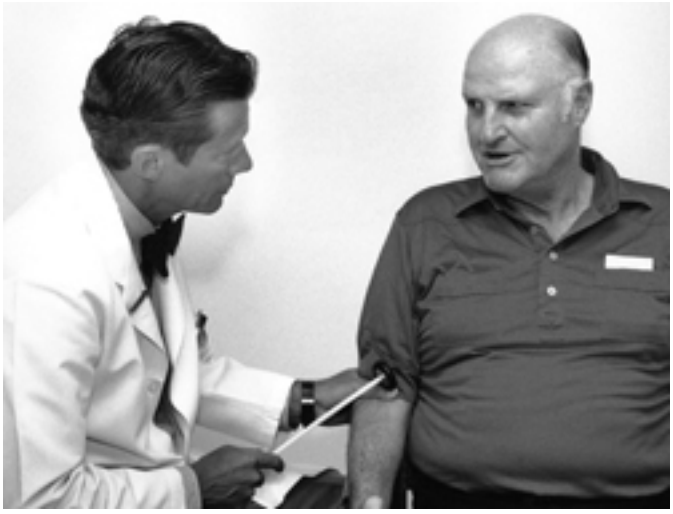
Who gets ALS?

ALS usually strikes in late middle age (the late 50s is average) or later, although there have been cases of ALS in young adults and even in children, as well as in very elderly people. Some genetic forms of ALS have their onset in youth.

Men are somewhat more likely to develop ALS than are women. Studies suggest an overall ratio of about 1.2 men to every woman who develops the disorder.



In ALS, upper and lower motor neurons normally send signals to lower motor neurons, which send signals to muscles.



Diagnosis is based on the medical history, physical examination and exclusion of disorders that may mimic ALS.

Genetic factors are part of the picture in ALS, and the disease can run in families (see "Does It Run in the Family?").

For years, experts have tried to find factors common to people who develop ALS, such as environmental toxins, occupational hazards, places of work or residence, and so forth. So far, the evidence for such risk factors and triggers has been frustratingly unclear, although a recent finding of an association between developing ALS and having served in the Gulf War in the early 1990s has indicated one of the strongest of these proposed risk factors. (For more on causes of ALS, see "What Causes ALS?")

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How is ALS diagnosed?

ALS usually announces itself with persistent weakness or spasticity in an arm or leg, causing difficulty using the affected limb, or in the muscles controlling speech or swallowing, leading to difficulty with these functions. It isn't unusual for people to ignore such problems for some time (perhaps months) at this stage or to consult a physician who may be relatively unconcerned.

However, the disease — if it's truly ALS — doesn't stop there. It generally spreads from one part of the body to another, almost always in parts adjacent to each other, so that eventually the problem can no longer be ignored or treated with exercise or a cane.

It's at this point that the patient is usually referred by a general practitioner to a neurologist, who will then consider ALS among many other possibilities.

MDA's ALS centers offer a team approach to treatment.

A thorough medical and family history and physical examination are the starting points of a neurologic workup. The person will undergo simple, in-office tests of muscle and nerve function.

If ALS is still being considered at this point, the next step is usually an electromyogram, or EMG. This test measures the signals that run between nerves and muscles and the electrical activity inside muscles to see if there's a pattern consistent with ALS. If there is, more tests likely will be ordered.

Additional tests may include imaging of the spinal cord and brain, usually by MRI (magnetic resonance imaging) scan, and sometimes a test of the fluid surrounding the spinal cord (spinal tap or lumbar puncture), which is performed by putting a needle into the back between two lower vertebrae.

Blood tests to exclude disorders that mimic ALS are also performed. In some instances, a muscle biopsy, which involves taking a small sample of muscle under local anesthesia, is performed.

With the exception of a genetic test that can reveal the source of the disorder in a small percentage of cases, the diagnosis of ALS is mostly a "rule-out" procedure. This means ALS is diagnosed after all other possibilities have been ruled out by specific tests.

Among the conditions that resemble ALS are some forms of muscular dystrophy, the neurologic conditions known as spinal-bulbar muscular atrophy and adult-onset spinal muscular atrophy, the nerve-to-muscle

transmission disorder known as myasthenia gravis, and various causes of compression of the spinal cord or brainstem, such as tumors and malformations.

If your condition has been diagnosed as ALS outside a major medical center or without extensive testing, it may be worth getting a second opinion. MDA-supported clinics and MDA/ALS centers are staffed by professionals who are highly skilled at diagnosing ALS and the conditions that resemble it.

What can be done about ALS?

Although ALS research is proceeding at an unprecedented pace, only one medication has been found to be somewhat effective against the disease and is approved by the U.S. Food and Drug Administration as an ALS treatment. That medication, riluzole (brand name Rilutek), has a modest effect in prolonging survival.

Several other medications are now in clinical trials (See "MDA's Search for Treatments & Cures.")

Until a definitive treatment or cure is found for ALS, MDA clinics and centers use a team approach to patient care that mobilizes a variety of health care professionals, all of whom aim to alleviate symptoms, maintain function and independence, prolong life and offer guidance for those with this disorder and their families.

Supportive braces can improve function.

Preserving hand function

Special grips for writing implements and eating utensils, devices that fit over keys to make them easier to turn, zipper pulls and button hooks can help make weakening hands more functional.

A professional therapist associated with your MDA clinic or MDA/ALS center can help you with these devices. For more, see MDA's book ***Everyday Life with ALS***.

Preserving mobility

Today's technology allows for mobility for almost everyone, no matter how few muscles remain functional. *Physical* and *occupational therapists* at your MDA clinic can help you obtain the equipment that's best for you at each stage of the disorder.

In the early stages, a cane or a supportive brace (*orthosis*) may be all that's needed to help the person with ALS get around.

Physical therapy can help

An *ankle-foot orthosis*, or *AFO*, can keep the foot from "dropping" with each step and tripping the person who's trying to walk.

with mobility.

Later, additional devices may be useful. These include walkers, manual wheelchairs, and power wheelchairs or scooters.

After the disease has progressed for a few years, a power wheelchair is usually highly desirable. A "tilt-in-space" type allows the seat to be positioned at a variety of angles, which relieves pressure and helps prevent irritation or breakdown of the skin. Some models allow the user to be brought into a standing position, which is generally good for circulation, bowel and bladder function, and bone preservation, as well as providing the psychological benefits of standing.

Careful planning for the type of wheelchair needed and desired, and a thorough knowledge of insurance matters in relation to wheelchairs, is important. Your MDA clinic or center often has a wheelchair specialist on-site or nearby who can help you with these matters.

Custom-fitted power wheelchairs can take many weeks or months to obtain, so plan ahead. Your physician or physical therapist may raise the issue of a power wheelchair before you think you're ready, but this is to avoid long delays between the time the chair is needed and the time it may arrive.

Preserving communication

For many people with ALS, the loss of mobility and strength is less distressing than the loss of the ability to speak. This may occur as the muscles in the mouth and throat that control speech and the muscles that help generate the pressure that moves air over the vocal cords lose power over time.

For this reason, *speech therapists*, or *speech-language pathologists*, are vital members of the ALS care team.

Speech-generating devices help maintain communication.

Early in the disease process, while speech is still normal or nearly so, speech therapists may suggest that a person with ALS record his or her speech. A number of phrases can later be programmed into a computer, or perhaps the person would like to talk about his or her life for future listening by friends and family.

Later, the therapist can teach the person with ALS special techniques for conserving energy and making speech understood as well as possible. In some cases, a dentist can be asked to make a device called a palatal lift that can help compensate for certain types of weakness in the roof of the mouth.

Later still, the therapist can help the person with ALS learn to use an electronic device (there are a variety on the market) that can substitute for speech. Some therapists recommend learning the required skills long before they're needed, preferably while good hand function remains and energy levels are fairly high. Learning to use such a device later may be harder, they say.

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Getting enough to eat and drink

As the muscles involved in chewing, moving food toward the back of the mouth, and swallowing weaken in ALS, eating and drinking become less pleasurable and more hazardous and time-consuming.

Regular measuring of respiratory muscle strength is an important part of ALS care.

The most serious problems are outright *choking* — obstruction of the trachea, or windpipe, by a piece of food — and *aspiration*, which means inhaling food or liquid into the lungs instead of routing it down the esophagus into the stomach. Normally, the throat muscles protect us from aspirating food or drink, but they may lose their ability to do this as ALS advances.

Speech-language pathologists or therapists are also specialists in swallowing, since these functions involve the same muscles as speech. Some therapists specialize more in speech and others more in swallowing, however.

Your MDA clinic has a swallowing specialist on-site or nearby who can specifically assess your swallowing at various stages of the disease and help you address problems as they arise.

Early solutions involve changing the consistency of food and liquids — usually thickening the liquids and avoiding large pieces of food — as well as changing swallowing techniques.

Later, if swallowing becomes very hazardous and eating takes a great deal of unrewarding time and energy, the therapist and physician may ask the patient to consider the insertion of a tube that goes directly into the stomach (not down the nose). This type of tube is called a *gastrostomy tube*, *g tube* or *PEG* — percutaneous endoscopic *gastrostomy* — tube. The term *gastrostomy* refers to making a small incision in the stomach. It's usually done *percutaneously*, which means "through the skin," with the help of an *endoscope*, a medical instrument.

If it's still possible to swallow some foods or liquids safely, the ALS patient can continue to eat and drink after placement of a feeding tube, but the swallowing mechanism no longer has to be relied on as the sole method of obtaining adequate nutrition. This can be a relief to those who can't take in enough calories by mouth because they get too tired or are afraid of choking or aspirating food.

Maintaining respiratory function

Perhaps the most serious medical complication in ALS is the gradual deterioration of the muscles involved in breathing. The *diaphragm* is an arched muscle located just beneath the lungs, which moves up and down and allows air to come in and move out. The *intercostals* are muscles between the ribs that contract and relax and also assist with air movement.

As these muscles weaken, the act of breathing, which is entirely automatic for most people, becomes conscious and energy-consuming.

At or before this stage of ALS, the neurologist will probably bring in a *pulmonologist* and/or *respiratory therapist*. These professionals are usually available in or near each MDA clinic or MDA/ALS center.

The physician may recommend that you consider using *noninvasive ventilation* to compensate for weakening muscles by allowing air to move in and out of your lungs as if your muscles were working well. In noninvasive ventilation, no incisions or body invasions are made.

Noninvasive ventilation comes in many forms, but usually consists of two basic elements — an “interface,” such as a mask or nose inserts, and air delivered under pressure by a small, portable machine. Usually, there’s one pressure for inhalation and another pressure for exhalation. This type of machine is often called a BiPAP, for bilevel positive airway pressure. (BiPAP is a registered brand name of Respironics.) There are other types of noninvasive ventilators as well, and professionals at the clinic will help you choose the device that meets your needs.

The device can be used as needed by the person with ALS. It need not be used around the clock, and pressures, masks and other aspects of the device can be changed as desired.

Invasive ventilation has afforded this woman continued vitality.

Another form of breathing support, known as *invasive ventilation*, delivers air through a hole in the trachea, or windpipe. The surgical creation of this hole is called a *tracheostomy*, and the tube through which the air is delivered is called a *tracheostomy (trach) tube*.

Invasive ventilation is thought by most doctors to be a more reliable means of delivering air to the lungs when the disease is advanced and the respiratory and throat muscles are almost entirely useless. But a tracheostomy is a big step, and carries added expense, specialized care by family members and/or hired professionals, a high risk of infection and an altered body image.

Decisions about ventilation aren’t easy to make. Professionals at the MDA clinic are there to help you.

Another aspect of respiratory care that’s important in ALS is *assisted coughing*. As the coughing muscles weaken, it becomes harder and harder to clear mucus from the airways. An assisted coughing device, which pushes air into the airways through a mask and then quickly reverses air flow, can help clear the airways and prevent infection. Your doctor may recommend other methods to assist with coughing and clearance of secretions from the airways.

Emotional and intellectual life

Although ALS shortens life, it doesn’t have to destroy it. An enormous number of people with ALS have rich emotional lives with their families and friends, continue with existing careers or interests or find new ones. People with ALS are artists, writers, readers, computer specialists, physicists, physicians, parents, lovers and poker players.

Many people with ALS — sometimes to the surprise of health care professionals and co-workers — maintain a sense of perspective and humor.

One “emotional” symptom of ALS that some people experience may be related purely to the physiology of the disease. Known as *pseudobulbar affect*, or *involuntary emotional expression disorder*, it involves prolonged laughing or crying spells out of proportion or inappropriate to the situation of the moment.

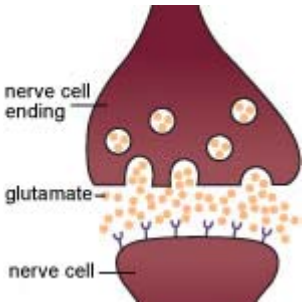
Some experts in neurophysiology believe this symptom arises from the loss of motor neurons in the top part of the brain that normally moderate the activity of the bulbar motor neurons in the brainstem. These motor neurons activate muscles in the face and throat involved in laughing and crying. Without the influence of the upper brain neurons, more “primitive” parts of the brain may take over, experts believe, leading to physical expressions of emotion that adults normally inhibit. The “pseudo” in the term refers to the fact that the location of the problem isn’t in the bulbar neurons themselves but in their loss of connection to neurons elsewhere in the brain.

Antidepressants are sometimes prescribed, and a medication developed specifically to combat this problem is being tested (as of 2008).

Many people with ALS and their families find support groups (ask at your clinic or local MDA office about one in your area) or Internet chat groups useful. MDA's support groups can provide important help for spouses and other caregivers, whose job can be very demanding.

Pharmaceutical treatments

While researchers continue efforts to identify compounds that slow or stop motor neuron degeneration in ALS, physicians can prescribe medications to treat troublesome symptoms during the course of the disease. These include drugs to ease cramps and muscle twitches, help in handling saliva, reduce anxiety and depression, treat constipation, help with sleep problems, and alleviate pain associated with prolonged immobility and joint displacements.



Glutamate carries signals between neurons (nerve cells), and there may be too much of it in ALS.

What causes ALS?

Years ago, it was widely believed that there might be one cause to explain all cases of ALS. Today, doctors and scientists know that can't be the case. Together, they're working to identify the multiple causes of the disorder.

The 1993 finding of the SOD1 gene mutation that underlies some cases of ALS (see "Does It Run in the Family?") opened a window into ALS. Even though very few ALS patients have flawed SOD1 genes, their disease (*familial ALS*) looks similar to sporadic ALS, the form that isn't caused by the SOD1 gene mutation. Scientists have concluded that the two types of ALS involve common biochemical and physical changes in the motor neurons.

Several clues to ALS causation have emerged since the early 1990s, and most experts believe these clues are linked to each other. The following possible causes are being studied by ALS specialists.

Free radicals

Free radicals are molecules that carry electrical charges that make them unstable and liable to damage cellular structures. They're a normal part of cellular life, and cells are usually able to neutralize most of them and keep their numbers in check. But in ALS, free radicals may build to toxic levels and damage cells, through an attack process called *oxidative stress*.

Excess glutamate

Glutamate is a common chemical in the nervous system, which neurons use to send signals to other neurons. But, like many things, glutamate has to be present in the right amount to work: Too little leads to a lack of signaling, too much to the death of the nerve cells that receive the signal.

Evidence from studies of people with ALS points to an overabundance of glutamate in the nervous system. This may result from inadequate transport of glutamate away from nerve cells after it has finished its signaling work.

Experiments suggest a defect also could lie in excess production or release of glutamate by the sending cells, or it could result from defects in glutamate receptors on the receiving cells.

Buildup of neurofilaments

Proteins known as *neurofilaments* form the "scaffolding" that helps nerve cells hold their shape. In ALS, these neurofilaments tend to clump up near the body of the cell instead of moving down the "tail" (axon) of the cell. This may be causing a cellular traffic jam, not allowing nutrients and other vital materials to move up and down the axon.

Defects in mitochondria

Of all the working parts of a cell, the energy-producing “factories” known as *mitochondria* are arguably the most crucial — especially for high-energy cells like motor neurons. They’re also among the most complex and most studied parts of the cell.

Mitochondria have their own genetic material (DNA). It bears some resemblance to the cell’s other DNA, which is organized into chromosomes in the cell nucleus. But mitochondrial DNA is organized differently, packaged into microscopic rings of genetic material that lack many of the protections against damage that chromosomes in the nucleus possess.

For this reason, and because processes inside the mitochondria produce dangerous free radicals, mitochondrial DNA is always in danger of being damaged. Some amount of damage occurs as part of the aging process, but in ALS, there may be more damage to mitochondria than the average aging cell sustains.

Cell suicide

Most cells have a built-in “suicide” program known as *programmed cell death*, or *apoptosis*. Under some circumstances, programmed cell death is normal. But in ALS and other degenerative diseases, it’s possible that the cell death program is activated inappropriately.

Immune system abnormalities

Many disorders that affect the nervous system are autoimmune in nature, meaning they occur when the body’s immune system mistakenly attacks its own tissues. Microglia, immune system cells found in the nervous system, appear to play a role in ALS. None of the treatments used for other autoimmune diseases has been effective against ALS.

Adhesion molecule abnormalities

In 2006, a large-scale study of people with and without ALS identified differences between these two groups in several genes for adhesion molecules, proteins that keep cells in the right place. Further studies are needed to understand the role of these genes in ALS.

Viruses and other infectious agents

For decades, scientists have guessed that viruses may play a role in ALS and other disorders that involve degeneration of nerve cells. So far, there’s no proof of a viral trigger.

The *HIV (human immunodeficiency)* virus, which causes AIDS (acquired immunodeficiency syndrome), can cause an ALS-like syndrome that improves with treatment with antiviral drugs. Most ALS patients aren’t HIV-positive, but the connection bolsters the idea that other viruses could also inflict motor neuron damage.

Traces of an *echovirus* were found in ALS spinal cord tissue in one study, but subsequent studies have so far failed to confirm this finding. Years of study have produced no evidence of any connection between exposure to the polio virus and the development of ALS, but viruses remain on the list of potential triggers of ALS.

Other microorganisms could be involved in ALS. Lyme disease, a bacterial disorder transmitted by infected ticks, can damage motor neurons. There is no evidence that Lyme disease causes ALS.

Prions, proteins that can act like viruses, change the way other proteins are shaped, turning them from beneficial to highly toxic molecules. Prions seem to have a preference for the nervous system, so they’re being examined for a possible role in ALS.

Toxins

The *heavy metals* lead, mercury and arsenic, although they can be toxic to the nervous system, haven't been shown to be causative agents in ALS.

Lead can damage upper and lower motor neurons, but, in the United States, exposure to lead has been monitored and limited for most people for about 25 years. In some circumstances, it may be worth testing for these exposures.

Prolonged contact with agricultural chemicals, such as pesticides, may be an ALS trigger in some cases.

The association of service in the Gulf War of 1990-91 with ALS may yield some clues. Some studies suggest that service in the military in general is a risk factor, in which case a broad range of factors will need investigation.

A high incidence of ALS on the island of Guam has led to the idea that the cycad seed, ingested on the island, could be an ALS trigger.

Electrical injuries

Some people have developed ALS following electrical injuries. These cases need further study.

Genes

In addition to those genes that can lead directly to ALS (see "Does It Run in the Family?"), there are almost certainly genetic risk factors, or "susceptibility factors," that may influence whether someone will develop ALS in the presence of a second or third circumstance (for example, exposure to a certain virus or environmental substance).

In August 2007, MDA-supported scientists at the Translational Genomics Research Institute in Phoenix identified several genetic differences in people with ALS compared to those without ALS. MDA is continuing to pursue this line of inquiry.

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Does it Run in the Family?

Family members can be tested for some genes known to be involved in ALS.

ALS is "familial" — that is, it shows a family history — about 10 percent of the time. Several genes associated with ALS have been identified or at least mapped to a specific region of a chromosome.

The SOD1 gene

In 1993, MDA-supported researchers identified a gene on chromosome 21 that, when flawed (mutated), causes ALS.

Mutations in this SOD1 gene account for some 10 percent to 20 percent of familial ALS cases and also perhaps 1 percent to 3 percent of cases with no family history. (Since ALS can be a very late-onset disease, some people with SOD1 mutations probably die from other causes without ever developing ALS, so absence of a family history in ALS can be misleading.)

SOD1 mutations usually lead to ALS that's inherited in an autosomal dominant pattern, which means the flaw isn't on a sex chromosome (it's on an "autosome") and that it takes a flaw in only one of a person's two SOD1 genes to cause disease. (For more information about inheritance patterns, see the MDA publication "Facts About Genetics and Neuromuscular Diseases.")

SOD1-related ALS sometimes assumes an autosomal recessive inheritance pattern, meaning that two mutated genes — one from each parent — are required before symptoms appear.

Other ALS-causing genes

Other genes that, when flawed, can lead to ALS, have been noted on chromosomes 2, 9, 14, 15, 18, 20 and the X chromosome, but not all these genes have been specifically identified.

The chromosome 2 gene codes for the alsin protein. When a child inherits a flaw in this gene from both parents (the recessive inheritance pattern), he or she can develop a childhood-onset form of ALS.

A gene on chromosome 9 that codes for the senataxin protein, when mutated, can cause a juvenile-onset form of ALS that can be inherited from only one parent (a dominant inheritance pattern). Onset is usually in the teen years.

Mutations in a gene for the protein VAPB, on chromosome 20, cause an adult-onset form of ALS that's inherited in a dominant pattern, as do mutations in a chromosome 14 gene that codes for the *angiogenin* protein.

More mappings, identifications and greater understanding of specific genes, such as those that carry instructions for adhesion molecules, are expected as research proceeds.

Genetic testing and counseling

Most commercial laboratories test only for the SOD1 gene mutations. A small number of labs test for mutations in some of the other ALS-related genes. Research studies may provide additional testing.

Genetic mutations can, of course, be passed from parent to child.

A genetic counselor can help interpret test results and discuss their implications for the person with ALS and his or her family.

MDA ALS Division

MDA is the world leader among voluntary agencies in fighting ALS.

Since the early 1950s, when Eleanor Gehrig served as a national volunteer leader of MDA, the Association has led the effort to assist those affected by the disorder that takes its name from her husband, baseball great Lou Gehrig, who died of ALS in 1941.

MDA's ALS Division offers the most comprehensive range of services of any voluntary health agency in the nation, and leads the search for better treatments and a cure through its aggressive worldwide research program.

In 2007, MDA allocated $12 million to ALS research and $11 million to services. Since 1950, the Association has invested more than $230 million in its ALS program.

If you've recently received an ALS diagnosis, this booklet will help you understand the disorder, while guiding you to the many services MDA provides.

"MDA Is Here to Help You" describes in more detail MDA's ALS Division program, which includes MDA/ALS research and clinical centers, an ALS Web site and several publications geared to those affected by ALS. We invite you to contact your nearest MDA office for help at each step of the way.

MDA's Search for Treatments & Cures

Many medications are being tested for potential benefits in ALS.

Riluzole, which has been on the market for ALS since the mid-1990s, is thought to work as a glutamate inhibitor, and other drugs that interfere with the synthesis, release or cellular reception of glutamate are being studied or tested (see "Excess glutamate").

Scientists and physicians are also investigating these and other compounds for possible benefits in ALS:

- Ceftriaxone, an antibiotic that may decrease levels of potentially toxic glutamate around nerve cells (see "Excess glutamate")
- IGF1 (Myotrophin), a neurotrophic (nerve-nourishing) protein
- Medications, such as coenzyme Q10, that limit oxidative stress and free radicals (see "Free radicals")
- Atorvastatin (Lipitor), for its anti-inflammatory effects (see "Immune system abnormalities")
- Arimoclomol, a compound that may help proteins, such as neurofilaments and others, fold into their proper shapes and avoid clumping (see "Buildup of neurofilaments")
- Zenvia (dextromethorphan and quinidine), a compound to treat unwanted emotional episodes (see "Emotional and intellectual life")
- Lithium, a compound used to treat bipolar disorder that has shown promise in a small study in ALS

MDA's Web site is constantly updated with the latest information about the neuromuscular diseases in its program. See the latest research news.

Another focus of MDA research is stem cells, primitive cells that can be programmed to generate specific cells the body needs. Early studies show that stem cells may have the potential to replace or repair motor neurons damaged by ALS. Scientists are now exploring the safety and practicality of this approach.

Augie's Quest

Fitness pioneer Augie Nieto and his wife, Lynne, are serving as co-chairpersons of MDA's ALS Division in 2008. Nieto, of Corona del Mar, Calif., received a diagnosis of ALS in March 2005.

Augie's Quest (www.augiesquest.org) is a concentrated MDA research initiative speeding the search for ALS treatments and a cure.

In 2006, Augie's Quest and MDA funded a landmark scan of the human genome that identified some 50 differences between the DNA of people with ALS and the DNA of people without ALS, one of which looks particularly significant.

In 2007, MDA and Augie's Quest joined forces with the ALS Therapy Development Institute (ALS TDI) in Cambridge, Mass. The three-year, multi-million-dollar partnership has created the largest ALS drug discovery project to date, a thorough review of biochemical targets in ALS and drugs that may work on them.

Augie's Quest enables the ALS TDI to apply industrial-scale technology to ALS research in a way that has not been previously attempted.

MDA is Here to Help You

The Muscular Dystrophy Association offers a vast array of services to help you and your family deal with ALS. If you've just received an ALS diagnosis, the staff at your local MDA office is there to assist you in many ways. The Association's services include:

- a nationwide network of hospital-affiliated clinics staffed by top neuromuscular disease specialists
- 36 MDA/ALS centers
- professionally facilitated support groups for those affected, spouses, parents or other caregivers
- assistance with purchase and repair of wheelchairs, leg braces and communication devices
- evaluations for physical, occupational, speech and respiratory therapy
- flu shots to help protect the respiratory system
- equipment loan closets

MDA's ALS Division offers an extensive program of up-to-date information about ALS research and care, such as the monthly MDA/ALS Newsmagazine. MDA's dedicated Web site at www.als-mda.org offers news and a series of online chats, including conferences with ALS specialists.

The book ***Everyday Life with ALS: A Practical Guide*** is offered free to anyone with ALS who's registered with MDA.

MDA also offers a booklet and a video on respiratory issues; a book of recipes for easy swallowing; and an ALS Caregiver's Guide. Go to www.als-mda.org, or check with your local MDA office for these materials.

This booklet is available in Spanish, and MDA has many other brochures and booklets about living with neuromuscular diseases available in English and Spanish.

If you have any questions about ALS, someone at MDA will help you find the answer.