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What is ALS?

Amyotrophic lateral sclerosis (ALS) often referred to as "Lou Gehrig's disease", is a progressive neurodegenerative disease that affects nerve cells in the brain and the spinal cord. Motor neurons reach from the brain to the spinal cord and from the spinal cord to the muscles throughout the body. The progressive degeneration of the motor neurons in ALS eventually leads to their death. When the motor neurons die, the ability of the brain to initiate and control muscle movement is lost. With voluntary muscle action progressively affected, patients in the later stages of the disease may become totally paralyzed.

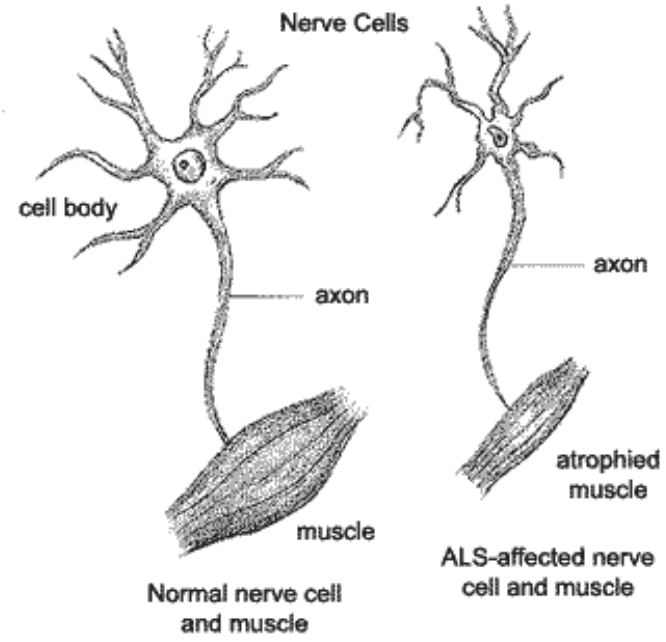
A-myo-trophic comes from the Greek language. "A" means no or negative. "Myo" refers to muscle, and "Trophic" means nourishment---"No muscle nourishment." When a muscle has no nourishment, it "atrophies" or wastes away. "Lateral" identifies the areas in a person's spinal cord where portions of the nerve cells that signal and control the muscles are located. As this area degenerates it leads to scarring or hardening ("sclerosis") in the region.

As motor neurons degenerate, they can no longer send impulses to the muscle fibers that normally result in muscle movement. Early symptoms of ALS often include increasing muscle weakness, especially involving the arms and legs, speech, swallowing or breathing. When muscles no longer receive the messages from the motor neurons that they require to function, the muscles begin to atrophy (become smaller). Limbs begin to look "thinner" as muscle tissue atrophies.

What Types of Nerves Make Your Body Work Properly?

(from Living with ALS Manual I: What's It All About?)

The body has many kinds of nerves. There are those involved in the process of thinking, memory, and of detecting sensations (such as hot/cold, sharp/dull), and others for vision, hearing, and other bodily functions. The nerves that are affected when you have ALS are the motor neurons that provide **voluntary movements** and **muscle power**. Examples of voluntary movements are your making the effort to reach for the phone or step off a curb; these actions are controlled by the muscles in the arms and legs.



The heart and the digestive system are also made of muscle but a different kind, and their movements are not under voluntary control. When your heart beats or a meal is digested, it all happens automatically. Therefore, the heart and digestive system are not involved in ALS. Breathing also may seem to be involuntary. Remember, though, while you cannot stop your heart, you can hold your breath – so be aware that ALS may eventually have an impact on breathing.

Although the cause of ALS is not completely understood, the recent years have brought a wealth of new scientific understanding regarding the physiology of this disease.

While there is not a cure or treatment today that halts or reverses ALS, there is one FDA approved drug, Rilutek®, that modestly slows the progression of ALS as well as several other drugs in **clinical trials** that hold promise.

Importantly, there are significant devices and therapies that can help manage the symptoms of ALS. It is important to remember that ALS is a variable disease; no two people will have the same journey or experiences. No matter what your individual course or situation may be, the ALS Association and your medical team are here to help.

Initial Symptoms of ALS

At the onset of ALS the symptoms may be so slight that they are frequently overlooked. With regard to the appearance of symptoms and the progression of the illness, the course of the disease may include the following:

- muscle weakness in one or more of the following: hands, arms, legs or the muscles of speech, swallowing or breathing
- twitching (fasciculation) and cramping of muscles, especially those in the hands and feet
- impairment of the use of the arms and legs
- “thick speech” and difficulty in projecting the voice
- in more advanced stages, shortness of breath, difficulty in breathing and swallowing

The initial symptoms of ALS can be quite varied in different people. One person may experience tripping over carpet edges, another person may have trouble lifting and a third person’s early symptom may be slurred speech. The rate at which ALS progresses can be quite variable from one person to another. Although the mean survival time with ALS is three to five years, many people live five, ten or more years. In a small number of people, ALS is known to remit or halt its progression, though there is no scientific understanding as to how and why this happens. Symptoms can begin in the muscles of speech, swallowing or in the hands, arms, legs or feet. Not all people with ALS experience the same symptoms or the same sequences or patterns of progression. But, progressive muscle weakness and paralysis are universally experienced.

Muscle weakness is a hallmark initial sign in ALS, occurring in approximately 60% of patients. Early symptoms vary with each individual, but usually include tripping, dropping things,

abnormal fatigue of the arms and/or legs, slurred speech, muscle cramps and twitches and/or uncontrollable periods of laughing or crying.

The hands and feet may be affected first, causing difficulty in lifting, walking or using the hands for the activities of daily living such as dressing, washing and buttoning clothes.

As the weakening and paralysis continue to spread to the muscles of the trunk of the body the disease, eventually affects speech, swallowing, chewing and breathing. When the breathing muscles become affected, ultimately, the patient will need permanent ventilatory support in order to survive.

Since ALS attacks only motor neurons, the sense of sight, touch, hearing, taste and smell are not affected. For many people, muscles of the eyes and bladder are generally not affected.

Who Gets ALS

ALS is a disorder that affects the function of nerves and muscles. Based on U.S. population studies, a little over 5,600 people in the U.S. are diagnosed with ALS each year. (That's 15 new cases a day.) It is estimated that as many as 30,000 Americans have the disease at any given time. According to the ALS CARE Database, 60% of the people with ALS in the Database are men and 93% of patients in the Database are Caucasian.

Most people who develop ALS are between the ages of 40 and 70, with an average age of 55 at the time of diagnosis. However, cases of the disease do occur in persons in their twenties and thirties. Generally though, ALS occurs in greater percentages as men and women grow older. ALS is 20% more common in men than in women. However with increasing age, the incidence of ALS is more equal between men and women.

There are several research studies – past and present – investigating possible risk factors that may be associated with ALS. More work is needed to conclusively determine what genetics and/or environment factors contribute to developing ALS. It is known, however, that military veterans, particularly those deployed during the Gulf War, are approximately twice as likely to develop ALS.

Half of all people affected with ALS live at least three or more years after diagnosis. Twenty percent live five years or more; up to ten percent will live more than ten years.

There is some evidence that people with ALS are living longer, at least partially due to clinical management interventions, riluzole and possibly other compounds and drugs under investigation.

Forms of ALS

Three classifications of ALS have been described:

- Sporadic – the most common form of ALS in the United States – 90 to 95% of all cases.

- Familial – occurring more than once in a family lineage (genetic dominant inheritance) accounts for a very small number of cases in the United States – 5 to 10% of all cases.
- Guamanian – an extremely high incidence of ALS was observed in Guam and the Trust territories of the Pacific in the 1950's.

The most common form of ALS in the United States is 'sporadic" ALS. It may affect anyone, anywhere. "Familial" ALS (FALS) means the disease is inherited. Only about 5 to 10% of all ALS patients appear to have a genetic or inherited form of ALS. In those families, there is 50% chance each offspring will inherit the gene mutation and may develop the disease.

Diagnosing ALS

ALS is a very difficult disease to diagnose. To date, there is no one test or procedure to ultimately establish the diagnosis of ALS. It is through a clinical examination and a series of diagnostic tests, often ruling out other diseases that mimic ALS, that a diagnosis can be established. A comprehensive diagnostic workup may include:

- Electrodiagnostic tests including electromyography (EMG) and nerve conduction velocity (NCV)
- Blood and urine studies including high resolution serum protein electrophoresis, thyroid and parathyroid hormone levels and 24 hour urine collection for heavy metals
- Spinal tap
- X-ray, including magnetic resonance imaging (MRI)
- Myelogram of cervical spine
- Muscle and/or nerve biopsy
- Thorough neurological examination

These tests are done at the discretion of the physician, usually based on the results of other diagnostic tests and the physical examination. There are several diseases that have some of the same symptoms as ALS and most of these conditions are treatable. It is for this reason that The ALS Association recommends that a person diagnosed with ALS seek a second opinion from an ALS "expert"-a neurologist who diagnoses and treats many ALS patients and has training in this specialty. The National ALS Association (ALSA) maintains a list of recognized experts in the field of ALS.