Amyotrophic Lateral Sclerosis (ALS) is a motor neuron disease, that is, a disease that affects the skeletal muscle nerve cells. A network of nerves carries messages from the brain, down the spinal cord and out to various parts of the body. Included in this network are motor neurons which carry messages to the skeletal muscles. In ALS, these nerves progressively degenerate and eventually die. As a result, the skeletal muscles do not receive the nerve signals they need to function properly and the muscles gradually atrophy or waste away from lack of use, leaving parts of the body paralyzed.

“A-myo-trophic” is derived from the Greek language meaning “no-muscle-nourishment.” Lateral refers to the area of the spinal cord where the muscle nerves are located. Sclerosis refers to nerve damage through hardening or scarring.

ALS is frequently referred to as “Lou Gehrig’s disease.” Lou Gehrig was a major league baseball player for the New York Yankees who was diagnosed with the disease and abruptly retired from the game in 1939. His sudden retirement and death in 1941 brought national and world attention to the disease.

Causes, Prevalence and Risk Factors

About 5,600 people are diagnosed with ALS in the United States every year. At any given time there are approximately 30,000 people who have the disease. As these numbers suggest, the average life expectancy after diagnosis is 3-5 years, with ten percent surviving ten years or more.

People who develop ALS are usually between the ages of 40 and 70 with an average age at diagnosis of 55. There are 20% more men than women with ALS at younger ages; however, as age at diagnosis increases, the proportion of men and women equalizes.

There are two known forms of ALS: sporadic and familial.

The sporadic form accounts for 90% of all cases. What causes it to appear is unknown and its risk factors are unknown.

The familial or inherited form makes up the remaining 10% of cases. Families that have ALS appearing more than once in their family tree are in this category. Only one specific gene has been identified: the SOD1 gene. It accounts for about 20% of the familial cases or 1% of all cases.

If there is a family history of ALS, then testing for the SOD1 gene may be an option to pursue. Of course, genetic testing is not a treatment for the disease but it may offer information about an individual’s capability of developing ALS and/or insight into a family’s capacity to transmit the SOD1 gene. Families without a history of ALS are unlikely to have the SOD1 gene present, so testing may not be beneficial to them. In any case, consultation with your neurologist and/or a genetic counselor is advisable.
counselor is advised.

Symptoms and Diagnosis

The symptoms of ALS vary from one person to another but the majority of people experience:

- muscle weakness in arms or legs,
- clumsy hand - dropping/fumbling with items,
- a hoarse voice,
- shoulder dysfunction,
- weak foot-foot drop,
- difficulty walking,
- exercise intolerance, abnormal fatigue,
- muscle cramps or twitches.

The disease progresses with increasing weakness and more muscles affected. Walking becomes strenuous or impossible and personal care may be difficult with limited use of the hands and arms. When the weakness has spread to the trunk of the body, speech, swallowing and breathing become impaired. Eventually ventilator support is necessary. Death usually results from the complications of inactivity or from paralysis of the muscles that control breathing.

ALS is difficult to diagnose because it shares many of its symptoms with a variety of other neurological diseases. Unfortunately, there is no one test that indicates that a patient has ALS. Instead a series of procedures are conducted eliminating other diseases until only ALS remains.

The procedures that may be used in diagnosing ALS include:

- a thorough neurological examination,
- blood and urine studies,
- electrodiagnostic tests including electromyography (EMG) and nerve conduction velocity (NCV),
- x-rays, including magnetic resonance imaging (MRI),
- myelogram of spine,
- muscle and/or nerve biopsy, and
- spinal tap.

Treatment

No effective medical or surgical cure exists for ALS. Only one drug, Rilutek, has been found to be somewhat effective in prolonging (a few months) life. Rilutek works by increasing nerve life by reducing nerve “stress.”

The remaining treatment options are to treat the symptoms. First and foremost, good nutrition is essential so that the body does not use the remaining healthy tissue for fuel. Several medications are available to help control muscle spasms, leg cramps and contractions. As the control of the hands and feet are lost, special grips, utensils, devices and braces are available to extend functionality. Also communication devices can be useful in helping the individual to communicate with others. The ability to communicate with others also aids in combating feelings of isolation and depression. As breathing becomes more labored, a number of devices are available to assist the individual.

More Information

Amyotrophic Lateral Sclerosis Association
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Muscular Dystrophy Association
www.mdausa.org/publications/fa-als.html

ALS Society of Canada
www.als.ca
References:
