KNOWLEDGE IS POWER...

WHAT IS ALS?

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ALS stands for **Amyotrophic Lateral Sclerosis**, commonly referred to as Lou Gehrig's disease after the famous Yankee baseball player who died of the disease in 1941. It is also called Charcot's Disease, or Maladie de Charcot, after a renowned French neurologist who described it in1869. The word amyotrophic comes from the Greek language and means 'without nourishment to muscles', referring to the loss of electrical potentials, or signals, that nerve cells normally send to muscle cells. 'Lateral' means 'to the side' and refers to the location of the damage in the spinal cord. As this area degenerates, it leads to scarring or hardening ('sclerosis') in the region.

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 and the atrophy or wasting away of the muscles to which those motor neurons are connected.

The motor nerves affected when you have ALS are those that provide voluntary movements and muscle control. **Voluntary muscles work according to your will and are under conscious control.**

Thus, when the motor neurons die, the ability of the brain to initiate and control muscle movement is lost. When voluntary muscle action is progressively affected, people may lose the ability to speak, eat, move and breathe.

Involuntary muscles, such as those that control digestion, heart beating and sneezing, are not under your direct control and are not affected by ALS. The senses, including hearing, sight, smell, taste, and touch, are not affected by ALS.

Often people have symptoms of ALS for many years before diagnosis. Sometimes that happens because we believe that these changes are a normal part of ageing. Sometime we are too busy or just don't want to know. What we do know is that most people will survive three to five years after diagnosis.

ALS can affect anyone of any age, gender or race. Most commonly, ALS is diagnosed in people between the ages of 40 and 70.

About 90% of cases ALS are sporadic, occurring randomly with no known cause. 5-10% of cases are called familial because they occur in families who have particular genetic defects.

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HOW IS ALS DIAGNOSED?

ALS can be very difficult to diagnose and the diagnosis is often made only after ruling out other options. Statistics tell us that many family doctors will NEVER care for a person with ALS, and many general neurologists may only rarely diagnose the disease in their patient population. Neurologist specializing in neuromuscular disorders and other subspecialty doctors such as Physiatrists are the doctors who diagnose and care for most persons with ALS.

There is no single diagnostic tool for ALS.

The first step in the diagnostic process is an examination by a neurologist. This will include detailed family, work, and environmental histories. During the exam, the neurologist will look for typical features of ALS that may include:

• **Muscle weakness** (which is often only on one side of the body, such as one arm or one leg) as well as changes in the character of the individual's voice (especially slurred words or slowness of speech). The exam will evaluate muscles of the mouth, the tongue, and those involved in chewing and swallowing.

• **Lower Motor Neuron** (LMN) is the motor component that connects with the muscles. Features of the disease include muscles shrinking in size or muscle twitches. These twitches are called fasciculations and may occur when muscles contract without the nerve cells fully controlling them.

• **Upper Motor Neuron** (UMN) is the motor component of the central nervous system that transmits impulses from the brain to lower motor neurons. Features of interest include hyperactive reflexes and muscle spasticity (a type of tightness and rigidity of the muscles).

• **Emotional changes resulting in the loss of some control of emotional responses**, such as uncontrolled crying or laughing. The exam will also look at changes in thinking, such as loss of good judgment or loss of common social skills. The examiner will look for problems in verbal fluency (strategic word Þnding) and word recognition abilities. These types of symptoms are less common or may be present but not easily identified.

The neurologist will also look for signs such as pain, loss of sensation, or extra-pyramidal rigidity, which is a different type of muscle rigidity that is most often seen in Parkinson's type disorders.

The next step in diagnosis involves a series of tests, including EMG (electromyography), a MRI (magnetic resonance imaging) of the neck, and sometimes of the head and lower spine, and a series of blood tests.

Sometimes urine tests, genetic tests, or a lumbar puncture (also called a spinal tap) are also necessary.

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EMG

The EMG is a very important part of the diagnostic procedure. Although this test can sometimes be uncomfortable, it is very important to have it done.

In the Þrst part of the EMG, small electric shocks are sent through the nerves to measure how fast they conduct electricity and to find out whether there is any nerve damage. The shocks tend to feel like the kind you get from static electricity but may sometimes feel a bit stronger.

This first part of the EMG determines whether the individual has 'nerve block', which is a feature of a different disease called multifocal motor neuropathy, and also tests whether the nerves that communicate sensation are affected, which may also indicate a disease other t han ALS.

The second part of the EMG tests the electrical activity of selected muscles. This is done by inserting a very fine needle into the selected muscles and using it to 'listen' to the pattern of electrical activity in these muscles. No electric shocks are involved in this part of the test and the needle does not inject anything into, or take anything out of, your muscles.

MRI

An MRI is a painless, non-invasive procedure that offers a very detailed picture of the spinal cord, the nerves that come out of the spinal cord, and the bones and connective tissues that surround and protect the spinal cord. It shows more detail than a CT (computed axial tomography) scan or X-rays. The MRI will help rule out pressure on the spinal cord or major nerves (such as from a herniated vertebral disk), Multiple Sclerosis, and tumors or bony abnormalities that compress the nerves. It can help detect vascular changes and strokes that sometimes affect the spinal cord or brain.

The individual having the MRI lies down inside a machine that is basically a large, rotating magnet. The test, that takes about 30 minutes, is noisy but it is painless. Some people have trouble being in a small, conÞned spaces and it is important to tell the doctor about this before the MRI begins so that medication can be given to help the person relax.

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BLOOD, URINE, AND OTHER TESTS

Blood tests are used to look for evidence of other diseases whose symptoms are similar to the early signs of ALS, such as thyroid and parathyroid disease, vitamin B12 deÞciency, HIV, hepatitis, auto-immune diseases and some types of cancer. Creatine Kinase (CK), a muscle enzyme released when muscles are injured or die, is also measured.

Specialized blood tests, such as autoimmune antibody tests, anti-GM1 antibody tests, and tests looking for high levels of protein in the blood and urine that may be related to some types of cancers are also performed. Depending on the individual's work and environmental history, the doctor may also test their urine for heavy metals.

In some rare cases, genetic tests and tests of hexosaminidase A levels (which can be related to juvenile spinal muscle atrophy) may also be performed. Genetic testing for ALS is usually only done when someone else in the family has ALS.

Occasionally, a lumbar puncture (also called a spinal tap) may be required. For this test, a small needle is inserted into the lowest part of the spine (below the spinal cord) to remove ßuid which will be examined under a microscope for abnormal cells. A lumbar puncture is usually done only if the individual has unusual features of ALS, such as spinal nerve abnormalities, or has no sign of abnormal reßexes or spasticity. Similarly, some people who have uncommon patterns of weakness, pain, or very high creatine kinase (CK) levels may need a muscle biopsy to look for muscle-speciÞc diseases. However, this is rarely necessary.

Once these tests have been completed, the neurologist may be able to tell whether an individual has ALS. Sometimes not all of the symptoms and Þndings that are required to make the diagnosis are present, particularly in the early phase of the disease. In this case, the neurologist will repeat the physical and neurological exams and the EMG at a later date to look for changes over time.

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