Title	What is Lou Gehrig's Disease? (ALS):
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Purpose

The goal of this course is to understand the signs and symptoms of ALS. Also included in this material are some guidelines for caregivers

Objectives

1. Describe the causes and diagnosis of ALS
2. What are the symptoms of ALS
3. What are some of the treatments for ALS
4. What medication currently exists for ALS
5. What research is being done
6. What information is there on genetics and ALS

What causes Amyotrophic Lateral Sclerosis and who is susceptible to it?

Amyotrophic Lateral Sclerosis (ALS) is a motor neuron disease. It is also referred to as Lou Gehrig's disease. He was a baseball player diagnosed with the disease in 1939. This neurodegenerative disease causes degeneration in the brain and spinal cord; consequently, causing progressive paralization due to degeneration in the upper and lower motor neurons. The causes of ALS are not completely understood. What is known about the biological causes of ALS is that a mutated gene (SOD1) produces a defective protein that is toxic to the motor neurons; it is found in 20% of inherited cases which is only 5-10% of cases. Some other causes found during research are excess glutamate oxidative and free radical damage, mitochondrial dysfunction and activation of the cell death pathways. Weakness in muscles are a hallmark sign and occurs in 60% of patients; however, initial symptoms of ALS vary among individuals. Some individuals trip, have twitching muscles, slurred speech or are unable to lift. Later, the patient is unable to breath or swallow. Interventions such as assisted breathing and feeding tubes help improve the quality of life and chances for survival. Therefore, the survival rate varies with the average rate being 3-5 years. Although some live longer. It has also been discovered that ALS can "remit or halt" its progress. However, the progressive muscle weakness and paralyses are "universal." These symptoms affect activities of daily living such as dressing and bathing. Regarding available treatments, they have come into existence and have been limited within the last 10 years.

Medication:

There is a medication on the market called Rilutek (Riluzole). The lab studies found that it is a "neuroprotector" in that it protects nerve cells from damage. Researchers believe that it protects the nerve cells by "glutamate (pronounced glu-tah-mate) inhibition." Rilutek is a tablet made with riluzole (pronounced rill-you-zole). The company that makes it is called Rhone-Poulenc Rorer Pharmaceuticals, Inc.

It is believed that Rilutek works by protecting the nerve cells from overexposure to glutamate. Glutamate is naturally produced by the body. It is found in the process in which the brain tells the muscles what to do. In the case of ALS, researchers believe that parts of the nervous system are overexposed to glutamate. If it builds up, as it does with ALS, it will "burn out" motor neurons so they can no longer carry messages to the muscles. Although researchers do not fully understand how this medication works, they believe it protects nerve cells from overexposure to glutamate.

In the over 1,100 patients in the clinical trials it was found that in the first year of treatment, patients that took Rilutek had increased their chances of staying alive compared to those who took a placebo. After 18 months, the tests showed no difference between the outcomes of the two groups. It is clear that Rilutek is not a cure but rather a treatment for ALS. It will not bring back any existing lost functions.

The recommended dose is one 50 mg table every 12 hours on a regular schedule every day. It must be taken within 1 to 2 hours after a meal as prescribed. The most common side effects of Rilutek are the following: weakness, nausea, decreased lung function, a mild temporary liver disorder and headaches. It is recommended that patients avoid smoking and alcohol while taking this medication. Also, before taking this medication, find out if you have liver or kidney problems. Blood tests are recommended while taking this medication because of the potential effects on the liver.

Research in the areas of stem cell and gene therapy on mice is currently being done. As with treatments, research has been more aggressively conducted in the last 10 years. Environmental factors such as toxins, diet and trace minerals in the soil, toxic metals and solvents, warfare, exercise or pesticides, and viruses are researched as well. Further research is required in both. The disease does not affect hearing, taste, touch, smell and the general muscles of the eyes and bladder are not affected. The mind is not affected either. As mentioned earlier, anyone may be affected by the disorder ALS. Statistically, 5,600 people in the United States alone are diagnosed each year. In the ALS care database it has been concluded that 60% of these cases are men and 93% of cases are Caucasian. The general age group is between 40 and 70 years of age and diagnosed at age 55. Even though the incidence of ALS is higher in men, as age increases the difference narrows between genders.

"With advances in technology, a better understanding of the disease process in ALS, and models systems now available, the time is right to translate research ideas into possible therapies. TREAT ALS (Translational Research Advancing Therapy for ALS), a drug discovery and clinical trials process initiated by The ALS Association, is supporting clinical pilot studies that offer novel clinical approaches for ALS. Several studies are already underway and investigators are encouraged to respond to the request for additional proposals offered in this publication."

According to Paul Gordon, M.D. of Columbia University and has colleagues, they are testing combined substances to treat ALS. It is felt that it is unlikely that a single compound will be able to treat ALS. They are currently testing celecoxib along with creatine and creatine and minocycline. These two combinations were beneficial in the SOD1 mutant mouse by prolonging its survival. This study was done on a small population. In the future, a larger population will need to be studied to confirm any results of the former study.

Researchers led by Don Cleveland, Ph.D., found that "antisense oligonucleotides" synthesized against SOD1 (copper-zinc superoxide distmutase 1) successfully down regulated the expression of the protein and increased survival in rats modeling ALS. More development is underway for the use of RNAi.

A number of other trials seek to follow "progression and gauge treatment impact. These include comparison of a new measure of muscle function, electrical impedance myography, to a more established test, motor unit number estimation. The drug memantine is also in a TREAT ALS pilot trial that will monitor an MRI signal related to integrity of neurons for changes with disease progression, and compare with a more traditional marker, the ALS functional rating scale."

Diagnosing ALS:

The process of diagnosing ALS is done by a rule out method. The following tests are used to diagnose ALS: EMG, NCV, blood and urine studies, spinal tap, x-rays, MRI, monogram of cervical spine, muscle and/or nerve biopsy and lastly a neuro exam. It is recommended by the ALS Association that a second opinion be obtained from an ALS expert.

Genetic Testing for ALS:

ALS is inherited in a small number of cases. Actually, 90% of cases have no family history. This is referred to as sporadic ALS (SALS). Familial ALS (FALS) is found in 10% of cases. A family history is the best method to distinguish between the two. This is done by either a neurologist or genetic counselor. Generally, the questions are the following:

- has anyone been diagnosed in your family?
- has anyone in your family ever had walking or speech difficulties? If any of these are positive, additional follow up questions are done. It is important to ask if any family member's died young to confirm a lack of medical history. Good resources for information are older family members and medical records that have a release form.

Regarding FALS, it can be inherited in the genes from both males and females. It is a dominant gene which means a child only needs one gene to be at a 50% increased risk for ALS. It is a random process and therefore the parent's have no control over which gene will be passed on to their child. This is important to explain to parents because they feel guilty. A child that inherits a gene for ALS does not guarantee their inheriting ALS. Also, if they do not inherit any gene from their parents, they will not inherit ALS.

Genes are in every cell in the human body. Their functions vary, some contribute to eye and hair color while others are responsible for proteins which determine how our bodies circulate blood or send nerve signals to the muscles. If a gene is mutated (disrupted by a change in sequence) the gene will not function properly.

Basically, genes are "packaged" in chromosomes and these chromosomes are in pairs. One chromosome is inherited from the father and one from the mother. This gives us 23 pairs or 46 chromosomes. The first 22 pairs are shared by both male and female. However, the 23 pair is different between the genders. It is the sex chromosome and females have two Xs and males have one X and one Y.

The University of Miami is collaborating with a Dr. Robert Brown's laboratory in Boston; there they may discover more genes that may be responsible for FALS and genes that make SALS patients either susceptible to or protected from developing ALS.

Basic Care for ALS Patients at Home:

Diet and Nutrition:

As with any patient with a progressive disease, ALS being no exception, a well balanced diet is required. Difficulties may be encountered when chewing and swallowing abilities

begin to decline. First, it is recommended that a multiple vitamin for general good health needs be given. As with any change in medications or treatment consult the patient's physician. If weight gain is a problem, high calorie shakes such as Ensure are available. Particularly once a G.T.(gastric tube) has been put in place. Once again consulting the physician and/or nutritionist is advised. For those not getting their nutrition via a G.T., it is recommended that breaking meals into five or six smaller meals may help with the fatigue a patient may feel as the disease progresses.

In terms of swallowing difficulties, particularly if the patient experiences coughing or choking with food and water, some changes will then be needed. In the earlier stages the following is suggested:

1. allow enough time to eat
2. thin liquids may give you difficulty and "go down the wrong way" as opposed to thicker liquids.
3. Avoid food that gives you difficulty
4. soft foods such as eggs, mashed potatoes and pasta take less energy to eat
5. if the following problems occur: no appetite, weight loss, no longer enjoying food, it takes more than one hour to eat, eating tires the patient, and lastly, food is "going down the wrong way" on a daily basis.

Self-feeding aids:

Some modifications in eating utensils may need to be done such as "built-up" handles on utensils for people with a weak grasp. Use of plastic or lightweight cups are easier to hold.

Elimination:

The voluntary muscles that control bowel and bladder function are rarely affected by ALS. However, due to the progressive deterioration, getting too a bathroom on time or on and off a toilet may be problematic. Some suggestions are to use a bedside commode or an elevated toilet seat. Handrails are useful too as well as bedpans.

For bowels to function properly the following is recommended:

1. Drink liquids (6 to 8 glasses a day)
2. Get fiber in your diet
3. Be as active as possible. If needed use a stool softener. The patient should have a bowel movement at least every 2-3 days. An urgency to urinate may be a problem and should be discussed with the physician or nurse. There are medicines that can help. Once again, it is recommended to drink a lot of fluids. This is to help avoid infections. If the urine is dark colored for example, then the patient needs to drink more liquids.

Hygiene:

Given the progressive deterioration of the patients with ALS, the following items may help with bath times: handrails, elevated toilet seat, toilet and tub handrails, bath bench with rail and back, hand held shower and power bath life.

Regularly and complete bathing is recommended. Checking the skin for "bedsores" should be done on a daily basis. Look for reddened areas and places such as elbows, heels, buttocks, and coccyx which are more susceptible to them. If an area is red, massage it gently with lotion. Make sure the patient is being turned if he/she is unable to do so by themselves. Turn every 1 to 2 hours is recommended. Foam cushions help as well as other devices to reduce pressure.

Oral care:

It is recommended to brush the teeth at least once daily. Perhaps an electric toothbrush would help. Lip balm or petroleum jelly may be applied to lips for dry and cracked skin. For swallowing difficulties during this care, a small pink sponge or suction device may be used.

Clothing:

Dressing an ALS patient may require a few different approaches given the progressive limitations that occur. What is recommended is light and loose fitting clothes; pullovers without buttons would work well. Slip on shoes that offer support and are slip resistance, shoe horns may helps; also, pants with elastic tops are easier to slip on and off. All clothing can be altered or modified. Velcro is one way of securing clothes instead of buttons and zippers.

Home Care:

If the patient with ALS chooses to be at home for his care you need to contact a Home Health Agency. The physician for the patient must order specific services to follow. Services ordered may include the following: physical therapy, occupational therapy, nursing care or a home safety evaluation. Generally, insurance companies pay for the "skilled" worker. They must be professionally trained or licensed. However, it is recommended that you check with your insurance company to see what benefits are covered under your specific policy.

Communication: Patients with ALS experience difficulty speaking during their lives. Due to muscle weakness, it is difficult to project the voice and/or form words. There are a number of things that can be done to assist with this:

1. using shorter words
2. speaking slowly
3. facing the patient when talking. It has been found that it is best if family members and staff resist speaking for the patient. It is best for the patient to communicate with others himself if possible. This is done best in a relaxed and unhurried atmosphere.

Alternative communication systems are available. The following are included: a homemade alphabet board, magic slates, and computers. A physician can order a speech evaluation with a speech therapist for the patient.

Psychological Support:

During this illness, there are people to help the patient and family. The patient and family may have the following feelings: denial, anger, withdrawal and depression. If an atmosphere which everyone is comfortable is created, generally everyone feels more comfortable to discuss their feelings. It is advised to enable the patient to discuss their feelings more freely. It is important to listen and encourage the patient to voice their concerns; this will hopefully provide a more honest environment; for example, it is advised not to give false assurances such as "everything will be all right." Acceptance and hope are important goals to achieve in this process. This process is based on each individual with their own symptoms and reactions.

In the case of ALS patients, it is important to know that no impairment of intelligence, judgment or other mental abilities is affected. So it is critical that they still be acknowledged in the family as a respected member who has physical deterioration. Thus, they should be a valued member of the family system.

Physical Therapy for the Patient:

Physical therapy may be needed and is not only good for the physical needs of the patient but their spirit as well. They have experience in the following areas: mobility, exercise, equipment, safety and pain. They can do many things with as recommend equipment and instructions, take measurements for a wheelchair, teach safe techniques for transferring and positioning, teach and/or perform stretching exercises and teach and/or perform range of motion exercises to keep joints mobile and free of pain.

Equipment:

As the ALS patient enters later stages of his or her disease, the following special equipment may be useful for them. Wheelchairs, form-fitted seats for wheelchairs, walkers, wheelchair lifts and stair lifts, patient lifts, sliding boards, electric hospital beds, page turners, bathroom aids such as raised toilet seats and commode toilet seats; sheepskin, wheelchair ramps and external urine catheters. Equipment such as the above should be used on the advice of a medical professional. A hospital bed is critical to the ALS patient for the following reasons: movement in bed, poor circulation, fluid retention, stiffness and bedsores may occur. Hospital beds are available for rental from medical supply houses.

Recreation:

The following are some recommended recreational activities: Cards, radio, T.V., reading (automatic page turners are available as well as book services). Volunteers or family members may take turns reading to the patient. Also, there is a Braille Institute Library that offers a free talking book service and lastly, playing chess by mail or other board games at home. Naturally when selecting activities keep the patient's interests in mind.

Activity:

You should be active as much as the patient's body tolerates yet not to the point of fatigue. A physical therapist can provide instructions for caretakers on exercises that are necessary and can be done safely. A walker may be used for some patients depending on the stage of their illness. A walker is prescribed for the patient along with the proper use. Walkers with wheels and a seat is safest.

If the patient is going to use a wheelchair it should be measured and properly fitted for him or her. There needs to be appropriate back and seat cushions. Whatever the needs are, it is important to have a professional instruct the individual on the use of the chair. Some of the chairs may vary due to features that can be modified for individuals such as: high back, head support removable arms, removable leg supports and tilt. Mobility of some kind whether a cane, walker or wheelchair is used because involvement with social activities is recommended and beneficial.

Pain:

ALS patient's may typically have pain in joints and muscles from reduced mobility. The actual nerve damage to ALS does not cause pain. Once again, physical therapists can teach exercises that stretch those areas. Pain management does not have to be difficult. Over-the-counter medications can be helpful. Talk to a doctor, therapist or nurse about this area.

Breathing:

It is predictable that losing your breathing will happen in ALS. The diaphragm, the most important muscle of breathing, has motor nerves that stimulate it. Once the motor nerves lose their function in this disease process, the patient will cease to breath on their own. The early signs of loss of breathing are the following: soft voice or fatigue with talking. Later signs are the following: fatigue with activities and breathing just weakens at this later stage. At some point, interventions people use are pillows to elevate them instead of lying flat.

At some point the patient will need to have their physician test their breathing. There are a variety of "non-invasive" machines that can help you take bigger breaths. These machines use a small mask or mouthpiece they are called non-invasive positive pressure

ventilation (NPPV). After using this machine at night for example, you can have more energy during the day because it has given your diaphragm a rest.

Once a patient cannot breath on their own, there are breathing machines that completely do the work of the diaphragm. Generally a tracheotomy is done and an invasive positive pressure ventilation (IPPV) or tracheotomy invasive positive pressure ventilation (TIPPV) is used. In any case, this should be discussed with the patient's physician.

Advance Directives:

It is advised to plan ahead so that in a crisis people will know what the patient chose to have or not have in your medical care. In the case of ALS, it is best to get information about assisted breathing options. It is helpful to have staff and family help with details and have frequent reviews of resources. After these discussions, a good understanding of the pros and cons should make things clearer.

Once the patient has made any plans in this matter, make them clear to family members and physician(s) so that they can honor them. Health care decisions should be put in writing in the form of an advance directive so decisions are known. Review the Advance Directive from time to time so that you are aware of it and may or may not want to make changes.

Hospice:

Services are provided in the home in the final stages of the ALS disease process in the following scenarios: a disease will shorten your life, you choose to not have artificial life support and your physician certifies there is a likelihood that your life will come to a close within six months.

The staff for hospice is trained to be sensitive to the physical and emotional needs of the patient and family. They provide skills, basic care and support for your family.

It has been found that for most ALS patients, they peacefully and comfortable pass at home. The team of doctors, nurses and hospice work together to ensure comfort for the family.

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Course Exam

1. ALS is not a motor neuron disease.

   ○ True ○ False

2. Rilutek (Riluzole) is a medication taken for ALS.

   ○ True ○ False

3. Lou Gehrig is the name of the baseball player that had ALS.

   ○ True ○ False

4. Diagnosing ALS is done by a "Rule Out" method.

   ○ True ○ False

5. 90% of the cases of ALS have a family history.

   ○ True ○ False

6. Chewing and swallowing may become difficult.

   ○ True ○ False

7. Lots of fluids are recommended for elimination.

   ○ True ○ False

8. An Advance Directive is advised so instructions are clear.

   ○ True ○ False

9. Patients with ALS experience difficulty speaking.

   ○ True ○ False

10. Intelligence and judgment are impaired with ALS.

    ○ True ○ False