The complications and symptoms of amyotrophic lateral sclerosis (ALS): A literature review

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ABSTRACT

Amyotrophic lateral sclerosis (ALS) is a disease that affects neurons, especially motor neurons, affecting the ability to send electrical impulses correctly, resulting in impaired functioning of voluntary contraction muscles. ALS is a progressive degenerative disease, so as the disease progresses, the symptoms progress and worsen, causing the patient to stop performing movements such as breathing, as a result of the involvement of the diaphragm muscle. The diagnosis is often time-consuming, complex and multifactorial, causing the possible carrier to go through several medical processes until the diagnosis is made. In the treatment, methods are used that help to slow down the progression of the disease, with the aim of increasing the patient's survival period, preserving their autonomy as much as possible. To date, there is still no cure for ALS. However, with the care of a qualified multidisciplinary team, it is possible to provide the patient with comfort and welcome during their treatment.

Keywords: Amyotrophic Lateral Sclerosis, Degenerative, Neurons.

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INTRODUCTION

Neurodegenerative diseases can affect people of any age, but the onset of symptoms is usually between the ages of 50 and 65. These diseases are chronic and affect the body's cells uninterruptedly, irreversibly, and with progressive symptoms, thus, as they evolve, the body's basic functions, such as movement, breathing, and reasoning, are affected (CAVACO, 2016; SANTOS, 2017).

One of the neurodegenerative diseases was first described in 1869 in Paris by Jean-Martin Charcot, a physician, professor and scientist, as Amyotrophic Lateral Sclerosis (ALS), which affects motor neurons. This disease is known as "Lou Gehring's disease" in the United States, due to the baseball player's death in 1941. In BRASIL, ALS was described by the physician Cypriano Freitas in 1909, and in 1919 the symptoms were described in two patients by the physician Gonçalves Viana. (CAVACO, 2016; SANTOS, 2017)

In order to better welcome people affected by neurodegenerative diseases, the Ministry of Health (MS) instituted the National Policy for Comprehensive Care for People with Rare Diseases in 2014. According to the Ministry of Health, in 2018 there were 14 thousand patients diagnosed with ALS in BRASIL, but received care through the SUS. (MS, 2018)

Therefore, the ALS disease has great importance and needs attention, since several studies have already been carried out in order to understand the mechanisms of action of the pathology and to verify the quality of life of patients who are affected by this disease and to ascertain the quality of life of each patient are carried out and yet so much is missing to understand the complexity of this disease. The main objective of this study is to present the complications and symptoms caused by Amyotrophic Lateral Sclerosis (ALS).

METHODOLOGY

The present study was developed through a literature review, as it is based on the survey and analysis of information already published.

According to Cruz and Ribeiro (2009), the bibliographic study is based on structured literature, obtained from books and scientific articles from conventional and virtual libraries.

A search was carried out in virtual health databases, specifically in the Virtual Health Library – VHL, Scientific Electronic Library Online, Google Scholar and reference documents available in specific portals, such as the Ministry of Health (MS). As exclusion criteria, articles in a foreign language, which were outside the theme and which were outside the established time frame, were eliminated.

Based on the reading of the abstracts, the articles were selected according to inclusion and exclusion criteria. Articles and books published in national journals that addressed the topic related to Amyotrophic Lateral Sclerosis (ALS) were reviewed.

WHERE AMYOTROPHIC LATERAL SCLEROSIS (ALS) OCCURS

In the development of the embryonic leaflet (ectoderm) the nervous system originates, this system is anatomically divided into the central nervous system (CNS) and the peripheral nervous system (PNS). The CNS is formed by the brain that is located in the skull, by the spinal cord that is connected to the brain and involves the bones of the spine, while the peripheral nervous system has in its formation 12 pairs of cranial nerves and 31 pairs of spinal nerves. (TORTORA; DERRICKSON, 2017).

In the brain, spinal cord and ganglia we find cells called neurons, which receive and interpret sensory information, transforming it into nerve impulses (electrical signals). These nerve impulses are transmitted in the region of the synapse where the action of neurotransmitters occurs that transmit command from one neuron to another, or from one neuron to other cells. The nervous system has three main neurons which are: Interneurons that connect the neurons found in the CNS; Sensory neurons that receive stimuli outside or inside the body; Motor neurons that conduct nerve impulses to muscle cells, glands, and endocrine cells. Therefore, when an injury or loss of neurons occurs, neurodegenerative disease arises. (MOREIRA, 2017).

An example of a neurodegenerative disease is Amyotrophic Lateral Sclerosis (ALS), which has in its pathophysiology the cellular and biochemical alteration that triggers the degeneration of motor neurons, affecting the muscles of voluntary contraction. (CAVACO, 2016; PINHEIRO, 2020).

The affected muscles suffer atrophy, so the term amyotrophic is used, which is the lack of contraction due to the absence of nerve stimulus, while the term used for neuron degeneration is sclerosis, because nerve cells are like scar tissue, so this pathology is named Amyotrophic Total Sclerosis because it can affect the upper motor neuron located in the motor area in the brain and the lower motor neuron located in the most lateral region of the spinal cord. (PINHEIRO, 2020).

In pathophysiology, the mechanism of action of the amino acid glutamate is studied, as it is an excitatory neurotransmitter of the CNS, which acts on neural development, synaptic plasticity and memory. Therefore, studies indicate that higher levels of glutamate cause excitotoxicity that lead to degeneration of neurons. (CAVACO, 2016).

According to Cavaco (2016, p.13).

> During glutaminergic neurotransmission, the glutamate released by the presynaptic neuron binds to specific receptors on the postsynaptic neuron. Activation of these receptors results in an influx of sodium (Na+) and calcium (Ca2+) into the cell, leading to depolarization and the generation of an action potential (Figure 1.5A). Excitotoxicity occurs when receptors are overstimulated, causing neuronal degeneration.

In addition, there can be an imbalance in the functions of astrocyte nerve cells, microglia, ubiquitin protein, mutation of the superoxide protein, neurofilaments, and viral infections that can leave motor neurons vulnerable, causing neural damage. (CAVACO, 2016).

RISK FACTORS AND DIAGNOSIS

Although the causes of the disease are still unknown, researchers around the world continue to look into the possible factors that increase an individual's likelihood of acquiring ALS. To date, some studies indicate that hereditary genetic compounds, age group (especially between 40-50 years) and male gender, are attenuating circumstances for the development of neurodegenerative disease. (ALENCAR et al., 2017).

External factors have also been the subject of research. It is believed that smoking practically doubles the chances of ALS, which usually manifests itself more aggressively in smokers. Military personnel may also be more susceptible. It is speculated that exposure to lead present in firearm ammunition may be a triggering factor for the disease. Athletes compared to the general population are more affected by ALS. As a response, studies suggest that the higher prevalence among this population may be related to repetitive head trauma and intense physical activity. Consequently, the more factors the individual accumulates, the greater the probability of a possible development of the pathology. (ALENCAR et al., 2017; BRASIL, 2019).

It is known that the early diagnosis of amyotrophic lateral sclerosis is very difficult to make, since this disease is similar to several other neurological diseases. Nowadays, the diagnosis of amyotrophic lateral sclerosis is based on an international recommendation called El Escorial Criteria, based on factors such as: Clinical history of progressive weakness; Neurological examinations with evidence of weakness and loss of strength, muscle atrophy, fasciculations, lively or exalted reflexes, and preserved sensitivity; Electroneuromyography (upper, lower, back and head limbs); Magnetic resonance imaging of the dorsal spine and brain. In addition, other tests may be requested, such as: Blood and urine tests; Lumbar puncture in order to exclude inflammatory diseases, infections and neoplastic infiltrations; Muscle biopsy, rarely used. (LIMA, 2016)

COMPLICATIONS AND SYMPTOMS

The first symptoms of Amyotrophic Lateral Sclerosis are difficult to identify due to the variation in the manifestation of symptoms in each person. Symptoms begin with simple muscle pain or muscle weakness located bilaterally in the upper or lower limbs. (NEVARES, 2018; RUBIN, 2019).

Because the symptoms are so unassuming, such as a simple trip on the carpet or a difficulty writing, few people identify them. The initial symptoms are, difficulty swallowing and breathing, stuttering, change in voice, (a slower speech than normal), due to the degeneration of the pyramidal bundles (responsible for the initial movements) causing spastic paresis and the reduction in the speed of nerve conduction, also resulting in respiratory failure, dysarthria or dysphasia, emotional lability (known as a pathological cry due to the increase in the mandibular reflex), problems with motor

coordination that occasionally end up hindering simple tasks of daily life, proximal weakness and distal weakness, difficulty climbing stairs (proximal weakness) due to degeneration of lumbar motor neurons, tremors, cramps and muscle spasms, loss of sensation in touch, difficulty holding the head up and weight loss. (NEVARES, 2018; RUBIN, 2019).

Consequently, as the disease progresses, the following complications may occur in isolated or added forms, such as aspiration of food or liquids, pulmonary insufficiency, bedsores, pneumonia, and inability to swallow, speak, or breathe. Therefore, the major complications of this disease are associated with the degeneration of bone marrow cells, causing muscle atrophies, sagging and decreased or absent reflexes, as they are part of the nervous system. ALS can also cause progressive bulbar palsy, which is a degenerative disease of motor neurons, progressively affecting the cranial nerves and affecting the facial nerves. (NEVARES, 2018; RUBIN, 2019).

FORMS OF TREATMENT

Treatment for Amyotrophic Lateral Sclerosis (ALS) aims to slow the speed of disease progression by increasing the survival period and patient comfort. Because it is an incurable and hitherto idiopathic degenerative disease, ALS does not yet have a specific treatment. In BRASIL, the only drug registered by the National Health Surveillance Agency (ANVISA) is Riluzole. Studies indicate that the drug can act as a neuroprotectant, acting to decrease the release of glutamate, resulting in the delay of neuronal death, providing a longer survival to the patient. Thus, it is necessary to use it as immediately as possible after the diagnosis of ALS. The total daily administration of the drug is 100 mg, divided into two doses of 50 mg each. Riluzole is provided by the Unified Health System (SUS) to patients with the disease. In what is checked by the Clinical Protocol and Therapeutic Guidelines for Amyotrophic Lateral Sclerosis, of the Ministry of Health. Article 2 establishes the obligation for the patient, or his/her legal guardian, to be aware of the possible risks and side effects arising from the drug treatment of ALS (BRASIL, 2020; BOSSE et al., 2020)

According to their clinical status, patients with ALS may need comprehensive and multidisciplinary care, in addition to neurological medicine, which aims to contribute to the preservation of their autonomy, quality of life and, subsequently, the relief of their suffering. As the loss of muscle strength is a common symptom, physiotherapy becomes a key part in all stages of the disease, since through specific exercises it will contribute to the optimization of the patient's muscular, motor and respiratory function. Dysphagia, anaarthria and dysarthria also affect most ALS patients, making it essential to follow up with a nutritionist and speech therapist, reducing the risks of tracheal aspiration and providing safer swallowing and preserving the patient's communication as much as possible. Impairments in self-care can cause psychological disorders to patients, caregivers

and family members. Thus, psychological follow-up is indicated, with professionals such as psychologists, psychiatrists and occupational therapists. As palliative therapies that aim to bring comfort during the patient's survival, the Ministry of Health offers Complementary Integrative Practices (PICS), which contribute to their physical and mental well-being. Among them include: Aromatherapy, chromotherapy, homeopathy, acupuncture, floral remedies (ABRELA, 2020; BRASIL, 2019).

In more severe cases that progress to hospitalization, the patient loses respiratory autonomy due to impaired function of the muscles involved in breathing, requiring mechanical support, or invasive mechanical respiratory ventilation (IMV), which should be performed with their consent. In addition to MV or IMV, patients with severe dysphagia may require enteral or parenteral feeding (RODRIGUES et al., 2017; SANTOS et al., 2020). The number of national studies on specific therapeutic treatment for ALS compared to international studies presents a profound disparity, which reveals the lag in the encouragement of BRASILian scientific research. Studies related to high technology, such as the use of the Brain-Computer Interface (CCI) device, are still utopian for the BRASILian reality (LEITE, 2021).

CARE PROVIDED BY NURSING

In order to provide quality of life to ALS patients, joint action between the family and the health service is necessary. The role of nurses in care goes beyond techniques and procedures, and it is very important to promote comfort for this patient (ESCOBAR et al., 2016).

The goal of treatment is to control symptoms and minimize disease progression. The treatment aims at the use of palliative measures, since there is still no possibility of cure and no way to delay the progression of the disease. Because it is a rare disease that is difficult to diagnose, it is necessary to be better prepared by the nursing team to establish continuous and direct care for these patients. Through the NCS and the nursing process, nurses should develop an individual and systematized care plan that meets the needs of each patient at each stage of the disease's worsening (ESCOBAR et al., 2016).

Humanized care occurs when nurses have the ability to understand the patient's needs and complexities, developing communication skills with them, whether verbal or non-verbal (ESCOBAR et al., 2016).

Among the procedures, oral and/or orotracheal aspiration is used to reduce the chances of bronchoaspiration and respiratory complications; respiratory physiotherapy associated with other care. Promotion of barrier-free environments for fall prevention; prevention of complications; nutritional follow-up and offer psychosocial support. Techniques such as reading, use of music and

change of position should also be used, as they promote comfort and well-being to the patient (ESCOBAR et al., 2016).

The nurse should monitor the appearance of new symptoms, always be aware of the patient's emotional conditions, so that referral to other professionals is anticipated (OMENA et al., 2019).

In addition to care for the patient's physical and emotional well-being, the nurse establishes care that includes the family as well, providing all the information about the disease, the options related to treatment, and which is the most suitable within their possibilities, informing them about the available social resources, providing emotional support, maintaining effective communication, in addition to preparing them for death in a timely manner. It is also important to educate the technical professionals who will provide care to the patient, implementing all the recommendations of the multidisciplinary team (OMENA et al., 2019).

The nurse must understand each person involved and explain the consequences of the decisions that will be made over time. For the patient who is cared for in Home Care, nursing visits should be regular, for the proper monitoring of medications, skin and gastrointestinal tract care, pain control, and the provision of the report with the evolution of symptoms to the doctors (OMENA et al., 2019).

Thus, the care of the nursing team and a multidisciplinary team is essential throughout the disease process, since it promotes satisfactory moments and quality of life for patients with ALS. (ESCOBAR et al., 2016).

FINAL THOUGHTS

Based on the researched literature, it is possible to understand that in an initial aspect, the main symptoms of ALS unfold from muscle weakness, which can be presented in isolation or associated with other signs.

The symptomatology of ALS is heterogeneous and can be confused with other neurological dysfunctions, resulting in a delay in diagnosis. In addition, it is clarified that because it is a progressive neurodegenerative disease, the complications of the patient's condition are expected and varied, requiring a multidisciplinary team to work on the specific control of these diseases, since the pathology itself is irreversible. In the context of rehabilitation, the aim is to preserve the patient's autonomy and provide comfort regarding their clinical manifestations.

It is up to the professionals involved to provide the appropriate competent care, which aims to provide the patient with the highest possible quality of survival, uniting their physical, mental and social well-being. In addition, the study also reveals the need for greater investment in public policies and BRASILian research related to ALS, which to date is fatal. Therefore, ALS is a physically and

psychologically devastating disease for the patient and family members around him, characterizing the need for specialized and intersectoral care.

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