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Amyotrophic Lateral Sclerosis

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Amyotrophic Lateral Sclerosis (ALS)

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ALS

- Degenerative disease of upper and lower motor neurons.
- Between 20,000-30,000 Americans currently living with the disease (Bellomo & Chichminski, 2015).
- Estimated 5,000 new cases diagnosed each year (Bellomo & Chichminski, 2015).
- Affects whites more than any other ethnic group in the United States (Rechtman, Jordan, Wagner, Horton, & Kaye 2015).
- Typically affects males more than females. Males often have a longer prognosis as well as those diagnosed younger in life, and those with limb onset versus bulbar onset (Rechtman et al, 2015).
- Prognosis after diagnosis averages three to five years (Bellomo & Chichminski, 2015).
- Most common cause of death is respiratory muscle weakness (Gordon, 2013).
- There is no cure, only palliative support (Gordon, 2013).

Why should clinicians care?

Over the past 10 years there has been no progress in the time it takes for clinicians to recognize and diagnose ALS. The average time from symptom onset to diagnosis is still eight to 16 months. This delay in diagnosis delays the patient and caregivers' access to critical supportive measures.

Misdiagnosis could potentially cause increase in suffering as unnecessary testing or surgeries might be performed.

Patients receiving care for ALS from a multidisciplinary team report higher quality of life.(Williams, Fitzhenry, Grant, Martyn, & Kerr, 2013).

Signs & Symptoms

- Muscle twitching, or fasciculation
- Muscle weakness
- Hyperreflexia
- Positive Babinski and Hoffman signs
- Emotional lability
- Usually begins in the arms in 2/3rds of patients
- Foot drop
- Difficulty walking
- Loss of hand dexterity
- Weakness when lifting arms (Gordon, 2013).
- 80% present with asymmetric limb weakness
- 20% present with bulbar onset
 - Dysarthria
 - Dysphagia
- There is no single definitive test. ALS is diagnosed by ruling out other diseases and the presenting signs and symptoms (Bellomo & Chichminski, 2015).

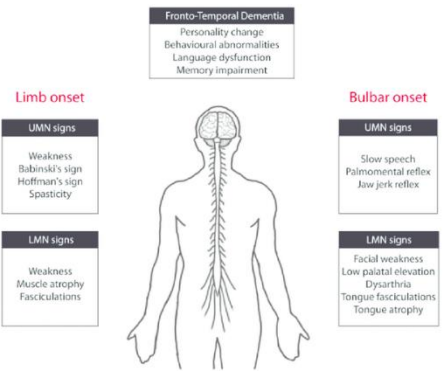
Pathophysiology

- Demyelination of upper and lower motor neurons.
- Loss of myelinated axons in the lateral and anterior columns of the spinal cord.
- Decrease in the size of the anterior horn in the spinal cord.
- Degeneration and loss of large motor neurons in the anterior horn of the spinal cord, lower cranial motor nuclei of the brainstem, and Betz cells in the motor cortex.
- Global reduction of all neurons in the anterior horn.
- Reduction of neuron size as well as atrophy of nerve fibers in ALS.
- Large empty spaces occur near neurons.
- Microscopic holes , which create a sponge-like appearance also occur.
- Bunina bodies are found in ALS, these are small (3-6microns big) eosinophilic inclusions that are found in cytoplasm of motor neurons. Their significance is not understood (Saber, Stauffer, Schulte, & Ravits, 2015).

Pathophysiology Continued

- Reactive astrocytes display increased immunoreactivity for the proteins GFAP and S100B. They express inflammatory markers, like COX -2, neuronal NOS (Saber et al, 2015).
- Microglia are activated in response to neuronal distress and release proinflammatory cytokines and reactive oxygen species as a result (Saber et al, 2015).
- Ubiquitin, a dense, round structure found in the cytoplasm of neurons in the anterior horn cells, is indicative of ALS (Saber et al, 2015).
- TDP-43 is a ribonucleic protein that stabilizes mRNA, and assists with its processing, transport, and translocation (Saber et al, 2015).
- In those with ALS, there is a loss of function in TDP-43 and noted formation of pathological aggregates, like ubiquitin, in the cytoplasm of the cells (Saber et al, 2015).
- The morphology of cell death in ALS is not clearly understood, but research points to cell apoptosis (Saber et al, 2015).
- Axonal regrowth is inhibited by a pathological protein found in muscles fibers called NOGO-A (Gordon, 2013).

Picture retrieved from Picher-Martel, Vladmanis, Gould, Julien, & Dupre, (2016).



Sporadic ALS

- Sporadic ALS occurs in 90% of all cases (Saber et al, 2015)
- Increased age, cigarette smoking, and pesticide exposure, service in the first Gulf War, and high levels of athleticism have been linked to increased risk for ALS. Trauma also shows an associative link. (Gordon, 2013).
- There is very little difference in disease signs and symptoms, progression, or prognosis in sporadic v. familial ALS. There is no cure for either variation, and all measures are supportive.

Familial ALS

- Superoxide dismutase-1 (SOD1) mutations accounts for 20% of familial ALS (fALS). (Saber et al, 2015)
- SOD1 has deposition of ubiquitinated TDP-43 negative SOD1 proteins in neurons of cells (Gordon, 2013).
- SOD1 has younger average age for onset, predominately starts in the legs with lower motor neuron disturbances, and has low occurrence of cognitive disturbances (Gordon, 2013).
- TARDBP mutations account for 5% of fALS cases (Gordon, 2013).
- TARDBP gene causes TDP-43 positive inclusions in the brain which leads to defects in RNA processing (Gordon, 2013).
- Whites with TARDBP often have onset of ALS in the arm while Asians with this mutation often have bulbar onset and longer disease duration (Gordon,2013).
- FUS mutations make up 5% of fALS, and affects amino acids in the FUS gene (Gordon, 2013).
- FUD mutations lead to onset of ALS at ages less than 40 in one third of the cases, typically present in the arms, and survival time is typically less than two years (Gordon, 2013).
- c9ORF72 is the most frequently occurring gene mutation in fALS, accounting for up 40% of cases (Gordon, 2013).
- TDP-43 accumulation in the brain and deposits of p62 show up predominately in c9ORF72 (Gordon, 2013).

Significance of Pathophysiology

Because ALS affects upper and lower motor neuron function, it eventually affects the entire body. The affected person may start out needing minimal help with activities of daily living (ADL), but eventually, loss of motor neuron function impairs body muscle movement to the point that the affected person is completely dependent on others for all ADLs

The most common complication leading to death is respiratory failure. Some affected by the disease may choose to initiate life prolonging measures such as placement of a PEGG or intubation and ventilation, but others may seek palliative care earlier on. Since there is no cure for ALS, and only one drug available on the market that has been shown to slow disease progression minimally, getting multiple supportive services into the home early has been proven to provide great benefit to the affected person and their loved ones through greater quality and quantity of life. (Williams et al., 2013)

Implications in Nursing

The implications of caring for someone with a life limiting and incurable disease shift the focus to supportive measures such as diet, respiratory therapy, home medical equipment, physical therapy, or counseling.

Ensuring that nursing care focuses on comfort, aligns with the patient and family's goals of care, and employs a multidisciplinary approach is key. Nurses need to use their resources and make referrals as appropriate.

Nurses will also need to be vigilant in assessing for skin breakdown for the bedbound and nutritionally impaired, assessing for respiratory infections and encouraging good pulmonary habits, and guiding patients and family's on food choices and safe swallowing techniques as dysphagia presents itself.

Communication in later stages of ALS can become problematic for many patients. Coming up with creative ways to communicate and encouraging early discussions about what a person wants for their future care will help guide families and clinicians alike through the terminal stages of the disease.

Conclusion

Amyotrophic lateral sclerosis is a disease characterized by the demyelination of upper and lower motor neurons. This may start locally, in the arm or leg, or could start with bulbar presentation, but eventually spreads to affect the entire body.

Most cases of ALS are sporadic, but up to 10% can have a genetic component. More research is needed in both types of ALS to help the medical field better understand and treat the disease.

There is no known cure for ALS. All treatment is supportive in nature with an important focus on a multidisciplinary approach to support patients and their families. Prognosis after diagnosis is most commonly three to five years, but can be longer or shorter. Getting a quick diagnosis and supportive resources in sooner has been shown to have better patient reports of quality of life as well as longer prognosis.

References



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